Heike I. Petermann · Peter S. Harper
Susanne Doetz *Editors*

History of Human Genetics

Aspects of Its Development and Global Perspectives

Springer

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Introduction

Heike I. Petermann, Peter S. Harper, and Susanne Doetz

> The history of human genetics is particularly interesting since, unlike in many other natural sciences, concepts of human genetics have often influenced social and political events. At the same time, the development of human genetics as a science has been influenced by various political forces.[1]

This statement by the human geneticists Friedrich Vogel and Arno Motulsky characterised the interaction of human genetics with society, which also forms part of this volume. The process of developing knowledge has become a major topic in the twentieth-century historiography of science and medicine. This is a multifactorial history, of which some aspects are presented in this volume, too.

The contributions are based on seven workshops that took place over the past 15 years. There, human geneticists have met historians to discuss the history of heredity and human genetics.

Six workshops were organised by the *Genetics and Medicine Historical Network* and held as satellite meetings of **the European Human Genetics Conference** of *The European Society of Human Genetics* (ESHG). The programmes are described in Chap. 2 and can be found in full in the appendix.

[1]Vogel and Motulsky 1986, 9—Considering also the interactions of other natural sciences with society, one may question the emphasis that Vogel and Motulsky put hereby on human genetics in opposite to other natural sciences.

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Parts I–VI reflect on the workshops as satellite meetings of the ESHG conference, from the beginnings to narrated history. The topics varied and highlighted many developments in the history of human genetics such as the origins of human heredity, diagnostic applications and the development of the field in different countries. The workshops were funded by the *Wellcome Trust*, ESHG and the *Deutsche Forschungsgemeinschaft* (DFG).[2]

The contributions to Part VII on genetic counselling are based on the workshop *The Establishment of Genetic Counselling in the Second Half of the Twentieth Century*. This workshop took place at the *Institute of the History of Medicine and Ethics in Medicine* of the *Charité* in Berlin in February 2016 and was funded by the DFG.

This volume reflects on different topics and starts with the beginnings of human genetics. Already in ancient times first considerations and thought on the inherited differences of man were made. Plato (428/423–348/347 BC) wrote in his state utopias how carefully a partner for producing children should be selected. Also in Epicurean philosophy ancestral concepts were found (C. Yapijiakis). Then for a long time, the question of heredity was no longer of interest until in 1605 Luis Mercado (ca. 1520–1606) published *De Morbis Hereditariis*. This publication was followed by similar ones in the following years. More than two hundred years later, Joseph Adams (1756–1818) published his influential book *A Treatise on the Supposed Hereditary Properties of Diseases based on Clinical Observations* (1814). But the question, how does heredity work, was still unsolved. In the middle of the nineteenth century, two important publications were made: polymath Francis Galton (1822–1911) published *Hereditary talent and character* (1866) and therefore is regarded as the founder of biometry.[3] The monk Gregor Mendel (1822–1884) experimenting with peas announced his attempts with plant hybrids in 1866 (*Versuche über Pflanzenhybride*) and set up the paradigm of Mendelism. But it was not until the twentieth century that William Bateson (1861–1926) named in 1906 the new science "genetics" and influenced the British Medical Societies (A. Rushton).

After the heredity of characteristics and qualities was recognised, the first attempts were made to diagnose dispositions and diseases as genetic. This was done using pedigrees (P. Wilson) and family histories (T. Pieters). But the results could lead to genetic discrimination of patients (S. Snelders). The description of *Genetic implication of the structure of deoxyribonucleic* acid by Francis H. C. Crick (1916–2014) and James D. Watson (b. 1928) was the starting point for the genome concept. This began to influence human genetics as well as molecular biology and raised the questions of its influence (R. Noguera Solano). At least, uncertainty gained importance in diagnosis (R. Pyeritz).

Did human genetics deal in all countries with the same questions? The special situation in Switzerland is reflected in the research on alpine isolates (P. Germann). In Scandinavia, the interaction between genetics and politics is explored (N. Roll-Hansen). The situation in Germany after 1945 is described for the western

[2]See programmes in the appendix of Chap. 2.

[3]Vogel and Motulsky 1986, 11.

(H. Peterman) and the eastern part (J. Pittelkow). The situation in Russia is depicted by the history of prenatal diagnostics (V. Baranov). Scientists not only from those countries meet at different international congresses, so the foundation of the International Federation of Human Genetics Societies is not surprising (K. Birmingham).

Already in 1888 H. Wilhelm G. von Waldeyer-Harz (1836–1921) had introduced the term "chromosome". At the beginning of the twentieth century, many articles were published about these, though the human chromosome number was late (1956) in being established. Gene mapping was important for the development and practice of human genetics. The first human genetic linkage was reported in 1936 (A. Rushton) and on this topic of human gene mapping the scientists of Glasgow (where the sixth workshop was held) had a considerable influence (M. Ferguson-Smith). But also in other countries like Greece, this topic was discussed (C. Morfakis).

Human genetics as a science was established in the twentieth century; therefore, we have the opportunity to gain information by interviews. For example, an oral history programme has been established at the *National Human Genome Research Institute* (NHGRI). (C. Donohue). But this method can also be used to obtain information about patients and their family (D. Mahr).

The first diseases were classified as hereditary early, for example polydactylia (1745) and haemophilia (1803). A path-breaking development was the discovery of the inheritance of alkaptonuria by Archibald E. Garrod (1857–1936), in conjunction with William Bateson in 1902. As biochemical and chromosomal diagnosis of genetic disorders progressed, the need for counselling for families and patients was soon recognised. The first centre (*Dight Clinic)* was founded in 1941 at the University of Minnesota, USA. This was the starting point, and by 1955, there were already 20 institutions in the USA that offered counselling and information free of charge. The establishment of genetic counselling is reflected in Part VII.

In 1964, the WHO Expert Committee on Human Genetics referred to genetic counselling as "the most immediate and practical service that genetics can render in medicine and surgery."[4] Given the low potential until very recently for an actual cure of most genetic diseases, genetic counselling became of special significance: it was the place where human genetic knowledge was put into practice—an important interface where genetic research, patient care, and the laboratory met each other. Moreover, the non-directive manner in which genetic counselling has been performed in recent years has granted legitimacy and created a necessary distance from eugenic practices in the first half of the twentieth century.

While research on the history of eugenics as well as human genetics and medical genetics has been conducted for some time, the history of genetic counselling has drawn the interest of historians of science and medicine only recently. This section includes essays by practising genetic counsellors, clinical geneticists, bioethicists and historians of medicine and science who have developed different perspectives

[4]WHO 1964, 27.

on the history of genetic counselling. By means of country case studies, we demonstrate how global, national and local factors influenced the establishment of genetic counselling and shaped its further development. The Cold War, religious and ideological concerns, adequate funding and the availability of technical resources were all reasons that could hinder or promote this process. Although the overall goals and the justification of genetic counselling were quite similar in all analysed countries, there were also some differences: abortion regulations had consequences for the outcome of genetic counselling; the incidence of genetically caused diseases and disabilities varied from country to country and guided the focus of genetic counselling: Sweden (M. Björkman, A. Tunlid), FRG (G. Moser, B. Nemec), GDR (S. Doetz), Czechoslovakia (M. Simunek), Austria (K. Geiger, T. Mayer), Belgium (J. Vandendriessche) and Mexico (A. Barahona). In Greece, for example, it was a priority to detect the carriers of thalassaemia (A. Barmpouti).

Even until the present genetic counselling has been an ethically controversial field that has met with critique by several groups. Therefore, we have included articles that explicitly address this topic: on personal counselling (M. Brusa, M. Barilan), by feminist criticism (S. Zuckerman) and the method of non-directiveness (A. Clarke). We conclude the section with a comment by Jean-Paul Gaudillière, who discusses the results of the contributions on genetic counselling and points out desiderata.

The aim of this volume is to present an overview of topics that have been discussed in the history of human genetics. We are aware that many subjects are missing; perhaps these will be discussed at future workshops. Therefore, this volume is an intermediate step, whose results raise many more questions that should be discussed in the future.

We proceed with the Seventh International Workshop on the History of Human Genetics from May 25 to 27, 2017, in Copenhagen to look at 50 years of the *European Society of Human Genetics*.[5]

As always, the authors are responsible for the content of their own contributions.

We should like to thank all of them for making this volume possible, and we hope that we have compiled a book worth reading.

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[5]For more information, see: www.eshg.org

Part I
Workshops on the History of Human Genetics

The International Workshops on Genetics, Medicine and History: An Overview, 2003–2015

Peter S. Harper and Heike I. Petermann

Abstract Between 2003 and 2015, a series of six international workshops on the broad theme of Genetics, Medicine and History has been held, under the auspices of the Genetics and Medicine Historical Network (*Genmedhist*), with a seventh workshop planned for 2017. The principal aim of the workshops was to promote mutual understanding and awareness between historians and those scientists and clinicians working in the field of human and medical genetics. This understanding and its practical consequences of collaboration and the greater preservation of both written records and the oral history of the field have been highly beneficial to all involved. The individual workshops were not published, though some of the material presented has appeared elsewhere. The book *History of Human Genetics* contains contributions to different workshops in the series and to a workshop in Berlin on the history of genetic counselling. The programmes of the different workshops are published in the Appendix, while this chapter provides a more general account of the workshop series as a whole, so that other chapters can be placed within the wider context of the topics to which they contribute.

Keywords History • Human genetics • International workshops • ESHG

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1 Introduction

When the Genetics and Medicine Historical Network (*Genmedhist*) was set up in 2002, one of its main aims was to bring together human geneticists (both laboratory based and clinical) with historians, archivists and others in the humanities. Small and informal international workshops seemed an obvious way to do this, and the success of the initiative can in part be judged by the fact that seven such workshops will have been held by the time that this volume appears. That this was actually achieved, rather than remaining an idea, is largely due to the support over this 14-year period from two bodies, The *European Society of Human Genetics* (ESHG) and the *Wellcome Trust*, as well as to the enthusiasm and hard work of all those involved in the workshops themselves (Table 1).

From the start, the emphasis was on informality and interaction, and since at the beginning their planning was led by geneticists rather than historians, the workshops did not require a written text to be brought to them, in contrast to most meetings of historians, where this is the norm; also little thought was given initially to the possibilities for publication.

With hindsight this lack of publication of individual workshops is a pity; looking through the abstracts, and in many cases the full transcripts fortunately preserved on the *Genmedhist* website,[1] as well as in the more extensive electronic archive of the project as a whole[2] one can see that many, indeed perhaps most of the contributions, deserve to have been more widely disseminated in full and finalised form. However, there is little doubt that at this early stage of bringing two the hitherto separate

Table 1 The genetics, medicine and history workshops

Year	Place	Principal theme
2003	Birmingham, UK	Launching the *Genmedhist* network
2005	Brno, Czech Republic	Preserving the history of medical genetics
2008	Barcelona, Spain	Genetics, history and public understanding
2010	Gothenburg, Sweden	Early history of human molecular genetics
2012	Nuremberg, Germany	The biological future of man: continuities and breaks in the history of human genetics, before and after 1945
2015	Glasgow, UK	Human gene mapping—oral history of human genetics

[1] www.genmedhist.org/workshops

[2] The archive of the History of Human Genetics Project, which includes material on the International Workshop Series, is held by *Cardiff University Special Collections and Archives* (SCOLAR), in association with other archival material of Professor Peter Harper.

communities of geneticists and historians together, the necessity for publication might have inhibited the informal interactions that have been one of the workshops' characteristic features.

Actually many of the individual papers have since been published, and an attempt has been made here to cite these linked publications, since they are somewhat scattered. However, no connected published account has been attempted until now of the actual workshop series as a whole, and it is hoped that this chapter will give an idea of the wider context in which many of the other chapters were originally presented.

It is fortunate that the *Genmedhist* website and the wider project archive containing both electronic and paper documents preserve a considerable amount of background material. This includes workshop programmes and participant lists, applications and post-workshop reports to *Wellcome Trust* and to other bodies, recordings and transcripts (mostly uncorrected) of individual presentations, as well as numerous photographs. We have drawn on and quoted from this material extensively for this chapter, rather than attempt to rewrite it a decade or more later, since these archived reports have the advantage of being contemporary to the events described.

2 First Workshop on Genetics, Medicine and History (2003): History of Human Genetics

This was an informal, exploratory half-day workshop, held immediately before the start of the main *European Society of Human Genetics* (ESHG) conference in Birmingham UK. Its principal purpose was to launch and publicise the *Genetics and Medicine Historical Network* (*Genmedhist*), initiated less than a year earlier, together with the proposed workshop series on "Genetics, Medicine and History". The programme consisted of talks by three invited speakers, followed by a demonstration of the newly created *Genmedhist* website (www.genmedhist.org) and an extended discussion on what the nature and main aims of the new network should be.

The speakers were not asked to provide a text, nor have their talks been published, but fortunately some rather full background material exists in the form of a report to *Wellcome Trust*, who supported the workshop, which is given here, since it provides a better summary than anything that could be reconstructed now, 14 years after the event. In addition, the individual talks and discussions were all recorded in full, and a transcript (unedited but clear) is available in the project archive for those who wish to consult it.[3]

> A total of 53 people attended the Workshop, organised by the Genetics and Medicine Historical Network as part of its Wellcome Trust supported activities, with a specific grant

[3] See footnote 3.

from the Trust supporting the Workshop itself. Those attending came from Genetics, History of Medicine and from those involved in Public Understanding of Science; most, but not all were attending the main European Human Genetics Congress and the holding of the Workshop in the same venue enhanced the contact.

The programme consisted of two parts: in the morning section there were three invited presentations, while the afternoon was devoted to the Historical Network itself.

The initial presentation was given by Professor Maj Hulten (University of Warwick), who described the original discovery of the correct human chromosome number (46) by Tjio and Levan in December 1955. As a person working as a student at the time in the Institute of Genetics, in Lund, Sweden, where the discovery occurred, Professor Hulten was able to give a vivid description of the event and its background. She also reviewed the technological advances in cytogenetics that made the discovery possible and discussed previous "near misses" that could have made it happen earlier. Of considerable interest was that some of these earlier studies had actually shown 46 to be the correct number but had felt constrained to interpret their data differently as a result of the general acceptance of 48 being correct.

Professor John Edwards (Oxford) gave a stimulating account of "200 years of genetics in Birmingham", tracing its origins from Erasmus Darwin and the Lunar Society during the 18th-century Enlightenment period, up to more recent contributions in the mid-20th century of Lancelot Hogben, McKeown and others.

Professor Peter Harper (Cardiff) spoke on "Julia Bell and the Treasury of Human Inheritance", outlining her life and career, and relating this to the monumental early human genetics work, The Treasury of Human Inheritance, founded by Francis Galton and Karl Pearson. He showed how Julia Bell became the key person in this work and how her rigorous mathematical and clinical training made it a source of major original findings in the field, still relevant today.

In the afternoon session, the Genetics and Medical Historical Network was introduced by Peter Harper, after which Dr Anita Shaw (Techniquest, Cardiff) gave a demonstration of the newly established website, developed by Jeff Alderman, showing how those interested could contribute material to develop this further. A valuable discussion followed, which allowed a series of key points to emerge, including

- The international nature and value of the Network
- The need to avoid editing or alteration of contributed material as far as possible
- To concentrate initially on documenting and raising awareness of existing material before moving to more ambitious projects
- To remain an informal grouping at present, not formally affiliated to any specific professional body

Finally, strong support was given to holding further workshops on historical aspects of genetics and medicine, with the next venue proposed as Toronto, in the form of a satellite meeting of the American Society for Human Genetics in October 2004. It is hoped that this will help to stimulate American interest in both the Historical Network and historical aspects of the field generally.[4]

The positive nature of the discussion and the enthusiastic reception of the talks thus led to a decision to hold further workshops, as well as to pursue the broader aims of the Network. It should be noted, though, that this initial workshop involved mainly scientists and clinical workers in human and medical genetics, rather than

[4] www.genmedhist.org

historians and others in the humanities, something that it was hoped to correct in future.

Another feature of this first workshop, which was continued in some future events, was to give a "local" slant to some presentations, as seen in this case by the talk of John Edwards on the history of genetics in Birmingham, while Maj Hulten was also a locally based cytogeneticist, even though her talk related to earlier years in Sweden.

3 The Toronto American Society of Human Genetics Meeting (2004): Historical Session

While not a "Workshop" as originally envisaged in the discussions held the previous year in Birmingham, this session, at the beginning of the ASHG annual conference on October 27th, 2004, was valuable not only in encouraging American and Canadian human geneticists to preserve and document the history of the field, but it also alerted historians of science and medicine in these countries to the rich body of material potentially available for study, which only few of them had so far been aware of. The well-attended session (over 300 people) was greatly enhanced by the active involvement of two workers, Victor McKusick and James Crow (both from the USA), who had been prominent in historical studies in addition to their distinguished scientific contributions. For the first time also, a science historian, Nathaniel Comfort, was able to contribute, while Peter Harper was able to report on the initiatives developing in Europe, including the previous year's Birmingham workshop.

The content of the session can best be seen in the summary given in the sixth Genmedhist Newsletter (February, 2005):[5]

> This was the first historical session to form part of an ASHG meeting and was highly successful. Moderated by Dr Victor McKusick and organised by the Genetics and Medicine Historical Network, it attracted an audience of around 300, despite an 8.00 am start on the opening day of the Congress.
>
> Dr James Crow (Madison, Wisconsin) was the opening speaker. With memory and experience dating back to the beginnings of human genetics, he gave a vivid account of the founding of the American Society of Human Genetics, its journal and the first editors, notably the brilliant and highly individualistic Charles Cotterman, who subsequently joined the Madison human genetics department.
>
> Dr Nathaniel Comfort (Johns Hopkins and UCLA) presented material from the ongoing American oral history project, comparing three of the founding medical genetics departments—Johns Hopkins, Baltimore (Victor McKusick); University of Washington, Seattle (Arno Motulsky); and University of Madison, Wisconsin (James Crow). He showed how the nature of the institutions and the skills and interest of the founders have had a crucial and lasting influence on how medical genetics developed in these three centres.

[5] www.genmedhist.org/newsletters

> The final speaker was Peter Harper (Cardiff, UK), who outlined some of the current and developing activities of the Genetics and Medicine Historical Network, including the archiving of Society and individual records, the newly formed Human Genetics Historical Library, a structure for historical interest groups in human genetics across Europe, based on The European Society of Human Genetics, and forthcoming events, notably the Brno workshop in Mendel's Abbey in May 2005.
>
> The session received very positive feedback from the audience and a number of people suggested that such sessions might form a regular part of the annual programme. It is also hoped that a coordinated American historical initiative, based around the considerable existing interest and activities, can be developed alongside and linking with that in progress across Europe.

It is worth noting that comparable "Historical Sessions", not discussed here, were also held at the 11th and 13th International Human Genetics Congresses, held in Brisbane (2006) and Kyoto (2016) respectively.

4 Second Workshop on the History of Human Genetics (2005): Preserving the History of Human Genetics

By the end of 2004, the activities of the Genetics and Medicine Historical Workshop, and in particular the international workshop series, had begun to attract the attention not only of human geneticists on both sides of the Atlantic, but of historians of science and medicine, as well as archivists. Despite uncertainty over funding support (the previous 1 year grant from *Wellcome Trust,* which had allowed *Genmedhist* to be set up, had ended and not been renewed), it was decided to hold a 2-day workshop in conjunction with the 2005 ESHG annual meeting in Prague. The possibility also arose of holding the meeting in Mendel's Abbey St Thomas in Brno, 200 km from Prague, which had recently been reopened as a historical and conference centre. The principal theme of the workshop was *Preserving the History of Medical Genetics*.

Thanks to the efforts of Milan Macek Jr. (CZ) and others on the programme committee, the logistical difficulties of transport, technology and accommodation were all successfully overcome, and there can be no doubt that the ambience of Mendel's Abbey was a powerful factor in the workshop's success, giving participants a sense of historical continuity with the beginnings of genetics. Fortunately also, a detailed photographic record of the event, including social activities, was made by Flo Ticehurst (Cardiff, UK), which can be followed on the *Genmedhist* website.[6]

However, the greatest factor in the success of the workshop was the quality of the presentations and the variety and interdisciplinary nature of the participants, as can be judged from both the programme and the abstracts, which are again given on the *Genmedhist* website, as is the list of participants.

[6] www.genmedhist.org/workshops

With hindsight, this workshop alone could have formed the basis for a valuable publication, but in fact a number of the contributions were published in journals or as part of books subsequently. Also, the talks and discussions were recorded and an uncorrected transcript of them, as well as the original recordings, is preserved in the archive of the overall project (as for workshop 1). Re-reading these more than a decade later is an interesting experience.

The *Wellcome Trust* had fortunately given some funding support in time to allow three guest speakers to be invited, and the best account of the workshop itself is the report made to the Trust soon afterwards.

> A total of 52 people from 20 countries took part in this workshop, held in the historic surroundings of Gregor Mendel's St Thomas Abbey in Brno.
>
> The theme of the meeting was 'preserving the history of human genetics' and the first day was devoted to this topic. Julia Sheppard (head of Special Collections, *Wellcome Trust*) opened the workshop with a highly informative talk on 'saving the archives of genetics', in which she outlined the key issues to be addressed and the steps to be taken if comprehensive and effective archives are to be achieved for human genetics. She was followed by Tim Powell, senior archivist at the Bath University National Cataloguing Unit for the Archives of Contemporary Scientists, who discussed the archives of human geneticists already involving this unit, notably the recently acquired and extensive records of Professor James Renwick. The papers were followed by discussion of other countries' experience and how international cooperation might help to ensure a more comprehensive archive.
>
> A brief presentation by Alan Bittles (AU) showed how human genetics research could utilise wider archives, in this case Swedish Lutheran church records for studies of inter-marriages. Finally, Peter Harper (UK) with Steve Pritchard mentioned the recently established Human Genetics Historical Library, involving Cardiff University Library Special Collections, and based on donations and collections from genetics units that would otherwise been lost.
>
> The second session moved to the field of oral history. In a discussion led by Soraya de Chadarevian (D; UK), both the importance of oral history and its potential pitfalls were outlined, as well as the urgent need for undertaking this in the case of human genetics, where many of the founding workers are still living, though now very elderly.
>
> Ludmila Pollock (USA) presented the extensive range of interviews based on workers visiting this laboratory, one of the key world centres for genetics research, where a systematic programme is under way for both an oral and written record of the field. From the UK, Marcus Pembrey gave his experience of chairing the witness seminar on genetic testing, organised by the *Wellcome Trust* History of 20th Century Medicine Group, while Peter Harper described a pilot series of interviews with early human cytogeneticists, hopefully to form part of a more extensive study.
>
> At the end of this session, Tayfun and Iclal Ozcelik (TR) presented a series of remarkable artistic creations where gene structures were translated into the forms of classical Turkish art.
>
> Day Two opened with a session on early pioneers and early concepts in human genetics. Alan Rushton (USA) showed how William Bateson, the founder of British genetics, interacted extensively with clinicians in developing his ideas, so that human genetics formed a key part of the new field from the beginning. Søren Nørby (DK) gave an account of the life and links to human genetics of Wilhelm Johannsen, the founder of genetics in Denmark, while Bent-Olle Bengtsson (SE) described a previously unrecognised Swedish book on heredity and medicine from 1879. In presentations from the Netherlands, Toine Pieters showed how ideas on heredity and cancer had fluctuated over the past 200 years, with phases where first heredity and then environment were regarded as predominant, while Stephen Snelders showed how comparable changes had occurred in the field of alcoholism.

> Session 4, on human genetics, eugenics and Lysenkoism, with contributions from Finland (Jaakko Ignatius), Austria (Tomas Meyer) and the Czech Republic, produced, as expected, a lively discussion and could have filled considerably more time. The session was especially interesting in the local context with presentations from the Czech Republic by Michal Simunek on eugenics and by Milan Macek Sr, Jiri Santavy and colleagues on the effects of the Lysenko period. It was clear that the legacy of these momentous episodes remains painfully real and that the history of Lysenkoism in relation to human and medical genetics has so far been largely undocumented. No Russian workers were able to attend the workshop, but contacts with both historians and geneticists in Russia have been made which should allow this area to be explored further.
>
> In the final session on historical aspects of medical genetics, Susan Lindee (USA) used the genetic research of Victor McKusick on the Pennsylvania Amish to explore how the beliefs and society of this unique population interacted with modern medical genetics, often in unexpected ways. Presentations by William Leeming and Patrick Macleod (both CN) illustrated the evolution and particular features of medical genetics services in Canada and the key role of some of its pioneers over the past 60 years.
>
> The closing discussion looked ahead to possibilities for future workshops, their location and funding support and also debated how wider historical activities in the field might best be encouraged and coordinated. There was general support and enthusiasm expressed for such developments to continue on both sides of the Atlantic, the most likely scenario being a further workshop in two years, with specific shorter sessions forming part of other meetings.
>
> In conclusion, this proved a most valuable and stimulating meeting, which brought together historians and interested geneticists from numerous countries, many of whom would not have had the chance to meet otherwise. There was notable enthusiasm for future collaboration and recognition that both historians and geneticists have key roles to play in documenting the history of this important field. The interactive nature of the workshop was greatly helped by the unique setting of Mendel's beautifully restored abbey and by the informative tours of the Abbey, its library and the Mendel exhibition that were arranged by the expert staff of the *Mendel Centre*. The Programme Committee would like to thank all those whose hard work made possible such a worthwhile and pleasurable workshop.[7]

Several points deserve to be emphasised alongside this report. First, the workshop was not only interdisciplinary in the sense that historians, archivists and both laboratory based and clinical geneticists were present, but there was a real 'meeting of minds' so that each discipline could become aware of the possibilities offered by the others, in ideas, approaches and potential research material. The geneticists and their records could offer abundant experience from their day-to-day work; for the clinical geneticists, this might be data on patient and family-centred aspects, while research and diagnostic laboratories could illustrate the development and impact of new technologies and the effects of genetic testing at both an individual and societal level. Equally, those working in the humanities could avail themselves of practical examples of the ethical and philosophical principles that they were analysing, often with few "real-life" data. Collaborations could be of the greatest value and a number were initiated at this and subsequent workshops.

At an immediate practical level, the possibilities of archiving and preservation of written records and of memories through recorded interviews described by historians and archivists gave encouragement and direction to those who could see the

[7] www.genmedhist.org/workshops: Second workshop.

urgent need for this but had little idea of how to proceed. Thus, the talks of archivists Julia Sheppard and Tim Powell, now published as a joint paper, were of great help in stimulating such preservation across Europe.

The need for efforts to increase understanding among historians as to the nature of medical genetics and its practice was illustrated by the following example recounted by social historian William Leeming (UK, CN) in his talk, of which a draft transcript is in the Project archive:

> Last Fall I had a paper come out on the early history of medical genetics in Canada. When I first tried to get this published, it was four years ago, this was to a reputable historical journal and the response I got back to it was from one anonymous referee, and the referee said "These medical geneticists the author is writing about appear to be a very busy lot. Where do they get time to do their cloning?" Well, I e-mailed the editors and I said "Is this a joke?" And they said "No." and I e-mailed them back and I said "Well they don't do cloning. I'm talking about medical geneticists. Have you read the paper? They have a very specific specialty and I'm talking about a specialty formation in the paper"; and they wrote back to me—(very telling, very telling). They said, "Well a geneticist is a geneticist so please deal with the cloning issue in your paper." Needless to say I moved on to other journals... On the negative side what this is really showing is that historians, people outside of genetics don't know what you do. They have no clear idea what you do.

Although such unawareness and disregard of a major field of science and medicine might seem incredible to objective observers, it was widely echoed in the experience of others, including one of the present authors (PSH). Fortunately, the situation has changed significantly over the past decade, thanks in large measure to the contributions of those historians present at the Brno workshop and its successors.

A further theme deserving particular note is the final session of the workshop, devoted to the effects on human and medical genetics of the Lysenkoist doctrines politically imposed on workers in the Soviet Union and in communist-dominated East European countries. This is an area very little researched by comparison with the situation in agriculture, and it came as a surprise to those participants from Western Europe and North America how painful and raw the topic still was, not surprising with hindsight, since it had blighted the scientific careers of a number of those present from these countries.

5 Third International Workshop (2008): Genetics, History and Public Understanding

This workshop, organised by Toine Pieters (NL), took advantage of the fact the *European Meeting on Philosophy and Genetics* (EMPAG) was being held jointly with that of ESHG in Barcelona at this time, giving greater possibilities for social scientists to take part. The programme, shown here, shows how social scientists, considerably more than historians at this point, had recognised the implications of human genetics for their own fields.

The summary report for the *Genmedhist* website emphasised some of the more historical contributions:

> This highly successful workshop had a different focus from its predecessor in Brno, utilising the opportunity of social scientists being in Barcelona for the joint ESHG/EMPAG meeting immediately following the workshop. Around 60 people attended, from a wide range of backgrounds; the participants list is given on the genmedhist website, along with the programme.
>
> Many of the presentations were primarily of a social science nature, but often with a historical approach and a 'time dimension'. A notable example of this was the presentation by Dr Zhai Xiaomei, leader of the Bioethics Unit at the Peking Union Medical College Department of Social Sciences and Humanities, who outlined the background to the Chinese Maternal and Health Law and the later abandonment of its eugenic aspects. Other important issues addressed included critical bioethical and social analyses of different aspects of reproductive genetics and of pharmacogenetics and technological developments such as microarrays.
>
> A number of the other presentations involved more strictly historical research, including accounts of how population chromosome studies developed in the Edinburgh Medical Research Council Unit in the 1950s and 1960s under Michael Court Brown, and also an overview of how medical genetics and genetic counselling developed in relation to Public Health in the 1930s to 1960s.[8]

6 Fourth International Workshop (2010): The Early History of Human Molecular Genetics

Innovations in technology, from cytogenetics to molecular biology, have undoubtedly played a key factor in many, possibly most of the major advances in human genetics over the past 50 years, even though the actual discoveries may have come originally from simpler organisms. Developments in molecular techniques have totally transformed the face of medical genetics research and applications, while the impact of the Human Genome Project has been and continues to be pervasive.

In the early *Genmedhist* workshops, technology perhaps received less attention than its due; in addition, the relatively recent advent of human molecular genetics, together with the increasingly electronic basis of most research, including correspondence and records, makes the history of this work especially vulnerable to loss or destruction. Thus, it was decided to give this fourth workshop, held in Gothenburg in June 2010, organiser Christos Yapijakis, Athens, a focus on the early history of human molecular genetics. We were fortunate in having as speakers a number of scientists who had been key players in developing this field, in addition to historians actively involved in its preservation and analysis.

Publication of this workshop was actually planned at the time, but sadly negotiations with publishers fell through. However, the project archive and *Genmedhist* website contain much valuable background material, including abstracts,

[8] www.genmedhist.org

participants list, photos and a summary report to *Wellcome* Trust, part of which is given below.

> The principal reason for choosing the theme was the recognition that, while human molecular genetics is a relatively recent area by comparison with other aspects of human and medical genetics, or classical molecular biology, it has had an immense impact during this short time of around 25 years. Also rapid changes in laboratory technology and the predominant use of email, the Web and other electronic techniques make it especially vulnerable to loss of the essential primary sources.
>
> It was recognised that a small workshop of this type could only be a beginning in the process of historical documentation and archiving, but it was felt that a combination of scientists from the field with historians and archivists could make a helpful start and identify some of the key issues.
>
> In the first session, some of the beginnings of human molecular genetics were identified. Christos Yapijakis (Athens), in his opening introductory talk, showed that molecular concepts of inheritance were already being proposed in Hellenistic and Roman times. Soraya de Chadarevian (UCLA) showed how early work on haemoglobin and its structure, by Max Perutz, Hermann Lehmann and others in the 1960s, provided a foundation for the subsequent research that allowed the specific analysis of human genes. Correspondingly, Jan Witkowski (Banbury Center, Cold Spring Harbor) illustrated the impact of the Cold Spring Harbor Symposia in developing both the ideas and technologies that would underpin the future human molecular genetics. The value of the extensive documentation and archiving of all aspects sets an example to others.
>
> In the second session, Tom Maniatis (New York), himself one of the key players in the development of human molecular genetics, described the principal discoveries in terms of advances in technology, such as the construction of DNA libraries, and DNA hybridisation and amplification. This was balanced by the presentation of Andrew Read (Manchester), who showed how the new research techniques and findings were first introduced into medical genetics services for important inherited disorders.
>
> The next talks (Judith Friedman, Max Planck Institute, Berlin; Patrick Lestienne, Bordeaux) illustrated how the application of molecular approaches had resolved two important 'problem' areas in human genetics, that of genetic anticipation, with the apparent deterioration across generations explained by DNA instability, and the analysis of the mitochondrial genome and its maternally inherited disorders.
>
> The history of the Human Genome Project was the focus of the next talk, by Ludmila Pollock (Cold Spring Harbor Library and Archives), who described an exciting international initiative (involving Wellcome Trust) to archive digitally as many documents relating to the project as possible.
>
> The day finished with a general discussion on which were the priority targets for ensuring the preservation of the history of human molecular genetics. Liz Shaw, one of three Wellcome Trust staff at the workshop, described the Trust's current initiative involving cataloguing and digitisation of genetics records; the importance of interviews was also emphasised, and the problems associated with archiving of electronic records and correspondence discussed. The discussions continued over an excellent dinner!
>
> Day two of the workshop began with two presentations from Mediterranean countries (Dimitris Loukopulos, Athens and Constantinos Deltas, Cyprus) on the applications of molecular techniques to carrier testing and prenatal diagnosis of haemoglobin disorders, showing the profound impact on the frequency of the disorder and the social acceptance of the approach in these populations. Describing the extensive restrictions placed on genetic applications as a consequence of the Nazi abuses, Heike Peterman (Nuremberg) placed these developments in a very different perspective. Peter Harper (Cardiff) then discussed the particular lessons to be learned from Huntington's disease both in terms of understanding its molecular basis and in molecular applications.

> Returning to the theme of the Human Genome Project and its predecessors, Sue Povey (London) described the series of Human Gene Mapping Workshops held between 1973 and 1990, which set the stage for the Human Genome Project; everyone agreed that this initiative was important to archive and document historically. Likewise the account by Mary-Claire King (Seattle) of the research in the laboratory of Allan Wilson on human evolutionary genetics, largely based on the new molecular techniques, illustrated another area of human molecular genetics with major impact.
>
> In the afternoon, Bengt-Olle Bengtsson (Lund, Sweden) showed a film that he had edited, from the 1948 8th International Genetics Congress in Sweden, giving valuable images of many important geneticists involved and showing the importance of the prolonged interactions allowed by such congresses in those years. This film will soon be available on the Web.
>
> The final presentation came from Walter Bodmer (Oxford) on the history of cancer genetics, showing how molecular approaches allowed both the isolation of key underlying genes and also the detection of those at high risk and the prevention of death by early intervention.[9]

From the topics covered in this workshop, it can be seen that not only was the molecular basis of specific areas of key scientific importance, such as haemoglobin and its disorders, addressed, but that broader aspects involving the preservation of the early history of human molecular genetics were beginning to be recognised, notably the major initiative by *Wellcome Trust* to preserve and digitise important records, the importance of the Cold Spring Harbor archival resources and the plans for ensuring that the history of the Human Genome Project is fully and impartially preserved. While it would be presumptuous to claim that the *Genmedhist* initiative and its workshops had been a direct factor in these, they have certainly played a role in making both the genetics and historian communities aware of the urgent need to capture this recent but exceptionally important area.

7 Fifth International Workshop (2012): The Biological Future of Man: Continuities and Breaks in the History of Human Genetics Before and After 1945

Human Genetics is a science with two sides: on one side concepts of human genetics have often influenced social and political events, and on the other side the development of human genetics has been influenced by various political forces. There were three main topics at this workshop:

- Eugenic ideas and human genetics before 1945: Concepts of heredity and research on genetic diseases
- Changing approaches after 1945: From molecular biology to molecular genetics.
- The shadow of eugenics on today's human genetics: Scientific, social, ethical, legal and political aspects

[9] Quote from report to Wellcome Trust, June 2010.

The meeting began at the Nuremberg Documentation Centre in the former Congress Hall at the Nazi Party Rally Grounds. After an introduction, participants could visit the exhibition "Fascination and Terror" on various aspects of the history of the Third Reich.

The workshop started with an introductory lecture of Nils Roll-Hansen (University of Oslo, NOR) on "Eugenics and the Science of Genetics". He pointed out that human genetics remained underdeveloped and backward until the 1960s.

The following talks had various approaches to "Human Genetics before 1945" and covered a number of geographic areas. Alan Rushton (USA) gave a talk on Charles Eduard of Saxe-Coburg and the Nazi politics, Yuriditzi Pascacio-Montijo (MX) challenged the use of intelligence tests and Philip Wilson (USA) discussed the collection and use of human pedigrees by the US Eugenics Record Office. Finally, Judith Friedman (USA) explored the different approaches in the study of hereditary disease, and Pim Huijnen (NL) introduced a tool to analyse newspapers by key words.

Afterwards, the topic "Continuities in the History of Human Genetics" was discussed regarding eugenic categories in Switzerland by Pascal Germann (CH) and the different meanings of the term "genome" by Ricardo Noguera-Solano (MX) and Juan M. Rodriguez-Caso (UK). Then Paul Weindling (UK) gave an invited talk titled "The Nuremberg Trials and Their Implications for Human Genetics". The medical trial included the testimony of victims and witnesses and at the end the foundation for the idea of informed consent was laid.

For Dinner the participants met on a historical site, a former brewery, and enjoyed a buffet with Franconian and Mediterranean food.

The second day started with "Informed consent—an Essential of Medicine. Consequences of the Nuremberg Doctor's Trial" by Stephan Kolb (FRG). The talk was on the importance of the verdict.

"Human Genetics after 1945" was the topic of the third session. There were various approaches that looked at the situation in the two parts of Germany: Susanne Doetz (FRG) focused on the situation in the *German Democratic Republic* (GDR) and Christine Scholz on that in the *Federal Republic of Germany* (FRG).

Then Richard Aspin introduced an online resource of the Wellcome Library that will serve as digital archive.

The closing remarks were made by Heike Petermann (FRG) on the continuities and breaks in the development of human genetics in the FRG. This closed the circle from the starting point in the documentation centre to the situation in human genetics today.

The question whether there are continuities or breaks in the history of human genetics is important from today's point of view.

At the end, the participants agreed to continue with workshops on the history of human genetics.

In all, there were 35 participants and the workshop was supported by the German Research Foundation (*Deutsche Forschungsgemeinschaft,* DFG) and *European Society of Human* Genetics (ESHG). The workshop was a satellite meeting of the

European Human Genetics Conference in Nuremberg in 2012 and organised by Heike Petermann (Germany).

At the workshop in Nuremberg, it was said that the next workshop would take place in Milan 2014. During the discussion in preparing it, the decision was made to shift it to 2015 because of the 50th anniversary of the ESHG in 2017.

8 Sixth International Workshop Glasgow (2015): Human Gene Mapping and Oral History of Human Genetics

Gene mapping in Drosophila began over a century ago, but human gene mapping is more recent, beginning with the linkage between haemophilia and colour blindness on the X chromosome in 1937 and the first autosomal linkage in 1951. In 1973, the first Workshop on Human Gene Mapping took place at Yale University. Guido Pontecorvo (1907–1999) had in Glasgow demonstrated that genes could be mapped in somatic cells. Therefore, the topic Human Gene Mapping is related to Glasgow.

"Human gene mapping" was chosen as one of the workshop themes partly to allow a more international coverage of this topic following an important UK Witness Seminar organised 2014 by Tilli Tansey.[10] Also because with initiatives under way to document the history of the Human Genome Project, focusing mainly on the later sequencing efforts, it was felt important to ensure that the earlier but essential work on mapping of human genes was not ignored.

On the other hand, "Oral History" is an important method in the history of human genetics providing an invaluable source of information. Conducting an interview requires careful planning before and after the interview itself, regarding also ethical and legal aspects.

This time the workshop started at the Archive of the University of Glasgow: showcase display and behind the scenes tour. This gave an interesting insight into the archive itself, but also for the collections of modern genetics including items from the papers of Guido Pontecorvo and James Renwick. It was a pleasure to be invited to the archive.

The first session was on "Human Genetics in Glasgow" illustrated by the talks of Malcom Ferguson-Smith (UK) on its contributions to the human gene mapping project. Paula Blair (UK) talked about the legacy of Guido Pontecorvo and Kevin O'Dell (UK) on James Renwick and the first human genetic maps. Closing remarks were made by Darren Monckton on the situation in 2015 and beyond. This session gave interesting insight into the situation of human genetics in Glasgow.

The next session was on "Human Gene Mapping". Alan Rushton (USA) made remarks on the first human genetic map in 1936, and Michael McGovern (USA) added more on the link between London and Baltimore including the

[10]Jones and Tansey 2015.

computerisation of genetics. Andrew Hogan (USA) was bridging the gap between cytogenetics and molecular biology.

The following session focused on the history of human genetics in general. Mauro Capocci (IT) illustrated the history of histocompatibility and Judith Friedman (CA) those of Leber's hereditary optic neuropathy. Reed Pyeritz made some remarks on the history of uncertainty in genomic medicine. Karen Birmingham (UK) introduced Marcus Pembrey and the International Federation to the audience. Then Aaro Tupasela (DK) talked on research into rare diseases in Finland. Elisa Houwink (NL) gave insights into genetic competence of primary and secondary care. The session was closed by Michael Simunek's (CZ) talk on the development of medical genetics in Czechoslovakia after 1945.

The workshop dinner took place in an Italian restaurant of Jamie Oliver in the city of Glasgow. This was accompanied by an animated conversation of the participants.

The sessions on oral history started with an introductory talk by Tilli Tansey (UK) on "You're all history now: recording the voices of modern genetics". This was followed by information on various projects of oral history: Peter Harper (UK) informed participants about his interviews with human geneticists, primarily across Europe. Heike Petermann (FRG) and Susanne Doetz (FRG) talked about their experiences with interviews in the Western and Eastern part of FRG. The last talk was on the combination of archival sources and oral histories by Miguel Garcia-Sancho (UK). It was the general opinion that oral history is a good method to explore the newer and recent history of human genetics.

In the area for the breaks, there was a exhibition of eight posters. Those interesting aspects especially relevant to the workshop ranged from medical genetics in Mexico (Barahona) to the human genome project in Greek newspapers (Morfakis), the history of human genetics in FRG (Petermann), the genomic collection at Wellcome library (Sloyan), two contributions on witnesses to medical genetics (Tansey, Jones) and neonatal screening in the Netherlands (Van El).

The closing remarks were the invitation to the next workshop on the history of human genetics on behalf of the 50th anniversary of ESHG.

The workshop was organised by Heike Petermann (FRG) and Judith Friedman (CA) and supported by the ESHG. In all we had 38 participants at the workshop.

9 Conclusions

The series of six international workshops (with a seventh planned for 2017) on the broad topic of the history of human and medical genetics has provided the opportunity to explore a range of major themes within this important area of science and medicine. Over this period of 14 years, the principal topics of focus have naturally evolved, but the key principles of the workshops and of the *Genetics and Medicine Historical Network* that has underpinned them have remained rather constant.

The first of these principles has been a pragmatic and practical one—the need to ensure that the primary material underlying the history of human genetics is preserved, whether this be written or oral, that this occurs on a worldwide basis and that it continues into the present (and future) digital era. It is easy now to forget that at the time of the first workshop and of the founding of *Genmedhist* the previous year (2002) how little was being done in this respect; certainly, there were almost no planned or funded initiatives, and there seemed to be little sense of urgency that action needed to be taken at once if a considerable part of the history of human and medical genetics might be irretrievably lost. Fortunately, this has now been overcome to a considerable extent, thanks in large measure to the involvement of major funding bodies such as *Wellcome Trust*, but with the growth of material to be preserved, the fragility of electronic information and the generally unfavourable financial climate for archives and archivists, this will inevitably be a continuing struggle.

The second principle, for which the workshop series can undoubtedly claim much credit, is the bringing together of two very different communities, historians and geneticists, both equally necessary for the full documentation and analysis of the history of human and medical genetics. As was seen by the experience of William Leeming quoted above in Workshop 2, most historians, at the time that the workshop series began, simply had no idea what the field of human and medical genetics was or did; or indeed that it existed at all! One of us (PSH) when beginning work in this field and consulting experts to find out what was going on already was repeatedly met by blank looks and by puzzlement that the basic material of the field was actually worth preserving! It seemed that there was a vacuum between the period of eugenics and the very recent Human Genome Project. Fortunately, a small number of historians felt otherwise, and the combined efforts of them and of similarly minded geneticists in the initial workshops soon helped workers from both disciplines to realise how productive a greater mutual understanding might be.

The third workshop helped to promote links with workers in the wider social sciences, who had already begun to form links with geneticists and to help in the analysis of the numerous and rapidly growing issues around genetic testing and broader human genetics. Likewise, the fourth workshop started to address historical aspects of new genetic technologies and how these had migrated from basic microorganisms to human genetics.

It rapidly became clear that each workshop was helping to open up new fields for detailed study and that there was an abundance—in fact a superabundance given the limitations of funding and available personnel—of important material and issues available for detailed historical studies. Also that there were numerous workers in genetics happy to collaborate, both in terms of being interviewed and making their records available and in tracing the development of a specific field.

This volume contains a number of different contributions related to the workshop series, some directly presented at one of the workshops, others building on such presentations. The general account of the workshops given in this chapter should hopefully help to place these varied contributions in the context of other

related presentations given at the specific workshops, some published elsewhere, others so far unpublished but nonetheless valuable.

By the year 2017, when this volume will have been published, the seventh workshop in this series will have been held, in Copenhagen, where the European Society of Human Genetics is holding its 50th anniversary meeting, and partly in neighbouring Lund. Since the field of human genetics continues to develop rapidly, with its applications in medicine and to Society as a whole increasingly widening, there will be a continuing need for historians and other workers in the humanities to link closely with geneticists to ensure that future developments are recorded fully and their history preserved and disseminated widely, so that we can all learn from them and ensure that they are used wisely for the benefit of humanity generally.

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Appendix

1st International Workshop on the History of Human Genetics.

Genetics, Medicine and History

Date: May 3rd, 2003.
Place: Birmingham, UK.
Support: Wellcome Trust, London; ESHG, Wien.

PROGRAMME

10.00 a.m.	Coffee and registration
10.30 a.m.	Hulten, Maj (Warwick, UK): Cytogenetic Milestones in Human Genetics. How to count to 46.
11.10 a.m.	Edwards, John (Oxford, UK): 200 years of genetics in Birmingham
11.45 a.m.	Harper, Peter (Cardiff, UK): Julia Bell and the 'Treasury of Human Inheritance'.
12.15	Lunch
1.00 – 2.30 p.m.	The Genetics and Medicine Historical Network - Introduction - Presentation of website - Further development - General discussion

2nd International Workshop on Genetics, Medicine and History.

Preserving the History of Human Genetics

Date: May 11-12, 2005.
Place: Brno, Czech Republic.
Support: *Wellcome Trust*, London; *European Society for Human Genetics* (ESHG), Wien; British Society for Human Genetics; Wales Gene Park, Cardiff.

Programme

Wednesday, May 11

9:00 a.m.	Coach leaves Prague Congress Centre for Brno (for those attending ESHG Congress)
1:15 p.m.	Tour of Abbey St. Thomas, Brno
2:00 p.m.: Session 1	***Preserving the Records of Human Genetics***
Sheppard, Julia (London, UK)	The Future of the History of Human Genetics; the role of Archives
Short contributions:	
Powell, Tim (Bath, UK)	Human Geneticists and the UK National Cataloguing Unit for the Archives of Contemporary Scientists.
Bittles, Alan (Perth, AUS)	Historical Patterns of Consanguineous Marriage in Northern Sweden.
Harper, Peter (Cardiff, UK)	The Human Genetics Historical Library
General discussion:	*archiving and records*
3:30 p.m. – 4.00 p.m.	Tea and coffee Break
4:00 p.m. Session 2:	***Oral History and Human Genetics – a discussion session***
Chadarevian, Soraya de (Berlin, D; Cambridge, UK)	The Value of Oral History
Short contributions:	
Pembrey, Marcus (London and Bristol, UK)	Witness seminars and Human Genetics
Pollock, Ludmila (Cold Spring Harbor, USA)	Talking Genomics
Harper, Peter (Cardiff, UK)	Interviews with early Human Cytogeneticists
Özçelik , Tayfun (Ankara, TR)	DNArt: A contemporary Sci-Art movement inspired by genetics.
5:30 p.m.	Informal reception, Mendel Museum (in Abbey) with introduction to exhibition by one of *Mendel Center* staff
8:00 p.m.	**Workshop Dinner**

Thursday, May 12

9:00 a.m.: Session 3	***Early Pioneers and Concepts of Human Genetics***
Rushton, Alan (Flemington, USA)	William Bateson and Human Genetics
Nørby, Søren (Copenhagen, DK)	Wilhelm Johannsen and the development of Danish Human Genetics
Pieters, Toine (Amsterdam, NL)	Two Centuries of Medical Thought about Heredity and Cancer
Short contributions;	
Bengtsson, Bengt-Olle (Lund, SWE)	Clinical Genetics Before Mendel
Snelders, Stephen (Amsterdam, NL)	Heredity, Genetics and Alcoholism in the Netherlands
DeArce, Miguel (Dublin, IRL)	Brno Revisited
10:30 a.m.	Tea and Coffee Break
11:00 a.m.: Session 4	***Human Genetics, Eugenics and Lysenkoism***
Simunek, Michal (Prague, CZ)	Eugenics in the Czech Lands
Mayer, Thomas (Vienna, AT)	Brief comments
Jaakko, Ignatius (Oulu, FIN)	How Eugenics Reached Finland
Discussion:	Lysenkoism and Eastern Europe
12:30 p.m.	Lunch
1:30 p.m.: Session 5:	***Historical Aspects of Medical Genetics***
Lindee, Susan (Philadelphia, USA)	Provenance and the Pedigree: Victor McKusick's Work with the Amish
Macleod, Patrick (Victoria, CAN)	F. Clarke Fraser and the Birth of Medical Genetics in Canada
Leeming, William (London, UK; Ontario, CAN)	Development of Medical Genetic Services in Canada and Britain.
Posters and discussion	
Soltan, Hubert (London Ontario, CAN)	The Early History of Medical Genetics in Canada
Jiri Santavy (Olomouc, CZ)	History of Medical Genetics in the Czech Republic
3:15 p.m.	Tea and Coffee Break
3:45 p.m.	Discussion on future workshops
4:15 p.m.	**Close of Workshop**
4:30 p.m.	Coach leaves for Prague

3rd International Workshop on

Genetics, History and Public Understanding

Date: May 30-31, 2008
Place: Barcelona
Support: *Wellcome Trust*, London; *European Society of Human Genetics* (ESHG), Wien

Programme

Friday, May 30th

10.00 a.m.	**Morning Plenary Session** **Welcome: Toine Pieters**
Condit, Celeste (Atlanta, USA)	From the "Central Dogma" to Gene "Expression": Materialist Understandings of Human Being
11.00 a.m.: Session 1a	***Organizing public communication and genetic literacy***
Patel, Heena (Leisester, UK)	The Sickness of Genes? Exploring the attitudes towards genetics in the community
González-Silva, Matiana (Barcelona, ESP)	The rhetoric of hope: The promises of the HGP in the Spanish Daily Press
Mathieson, Kate (Univ. Manchester, UK)	Dialogue events on genetic medicine
11.00 a.m.: Session 1b	***Organizing communication in specific cultural contexts***
Petermann, Heike (Muenster, FRG)	Brave New World? Reflections on the role of utopias and their public understanding in the history of human genetics.
Needs, Jaqueline (Cardiff, UK)	Participation in Huntington's disease research: hoping, coping and a nice day out
Wilde, Alex (Sydney, AUS)	The impact of news coverage of the genetics of major depression, bipolar disorder and schizophrenia
12.30 p.m.	Lunch
Topic 2	**Public challenges: data sharing, risk and stigma**
1.30 p.m.: Session 2a	***Public challenges: the question of ownership***
Exteberria Agiriano, Arantza (San	Ethical challenges of genetic biobanks

Sebastián, ESP)	
Rodriguez, Victor (Delft, NL)	Is there a failure of concern on modes and impact of material transfer? A review of the empirical evidence
Pavone, Vincenzo (Madrid, ESP)	Genetic testing, geneticization and social change
1.30 p.m.: Session 2b	***Public challenges; the question of race and ethnicity***
Michelsen, Øivind (Oslo, NOR)	Morality of inclusion: reflections on the legitimacy of population structuring by 'race' in contemporary medical genetics
Bonham, Vence L. (Bethesda, USA)	US Physicians' attitudes towards race, genetics and clinical medicine
Snelders, Stephen (Amsterdam, NL)	Genetics in the doctoring of cancer and alcoholism - Historical lessons on racial and ethnic discrimination
3.00 p.m.	Tea and coffee break
3.30 p.m.: Session 2c:	***Public challenges: the question of ethnicity and cultural differences***
Olarte Sierra, Maria Fernanda (Amsterdam, NL)	Amniocentesis: A Troubled Hope (The Columbian experience)
Baig, Shahid Mahmood (Faisalabad, Pakistan)	Controlling monogenic disorders through cascade testing, prenatal diagnosis and genetic counselling in a highly consanguineous population
Topic 3	**Understanding genetics as a technological and social project.**
3.30 p.m.: Session 3a:	***Testing the Person and Society***
Cornel, Marina (Amsterdam, NL)	Governing the balance between 'duty to protect' and 'right to test'
Löwy, Ilana (Paris, FR)	Preimplanatory diagnosis and cancer risk
de Chadarevian, Soraya (Los Angeles, USA)	Karyotyping human populations in the 1960s
Santesmases, María Jesús (Madrid, ESP)	Establishing karyotypes: from children to foetus, 1966 -1976
8.00 p.m.	**Workshop Dinner**

Saturday 31 May

10.15 a.m.: Plenary Session	
Hedgcoe, Adam (Sussex, UK)	Genomic Expectations, Bioethics and the Social Sciences.
Topic 3:	**Understanding genetics as a technological and social project**
11.00 a.m.: Session 3b	***Taming microarrays***
Fujimura, Joan H. (Madison, USA)	Fishin' Chips: Microarrays as predictive technology in biomedical genomics

Jordan, Bertrand (Marseille, FRA)	Why is the adoption of DNA microarrays for clinical diagnostics so slow?
Rogers, Susan (Montreal, CAN)	Collaborating on comparability: How standards and regulation sustain microarray experimentation
11.00 p.m.: Session 3c	***Promise and hope***
Moors, Ellen (Utrecht, NL)	Genomics based expectations of personalised health. An innovation systems perspective
Boenink, Marianne (Twente, NL)	An ethics of promising
Wieser, Bernhard (Graz, AT)	Guiding public accounts of genetics: a strategic enterprise
12.30 p.m.	Lunch
1.30 p.m.: Plenary Session	
Zhai, Xiaomei (Peking, CHIN)	Public understanding of genetics in China
2.15 p.m.: Session 3b	***Taming microarrays***
Mustar, Philippe (Paris, FRA)	Microarrays as collective innovation
Bourret, Pascale (Marseille, FRA)	Governing Microarrays
Söderqvist, Thomas (Copenhagen, DK)	Microarrays and the Empire/Multitude-distinction: A 'presence'-approach to the public engagement with microarray technology and SNP genotyping
2.15 p.m.: Session 3d	***The challenge of integrating preventive and genetic medicine***
van El, Carla (Amsterdam, NL)	An unhappy wedding between genetic screening and prevention
Holton, Neal (Minneapolis, USA)	Public health genetics in mid-twentieth century Minnesota
3.45 p.m.	Tea and Coffee Break
4.15 p.m.: **Plenary Session**	The future of the history of human genetics: the role of archives
Harper, Peter (Cardiff, UK)	
4.45 p.m.: Closing remarks	Toine Pieters

4th International Workshop on History, Medicine and Genetics

The Early History of Human Molecular Genetics

Date: June 11-12, 2008
Place: Gothenburg, Sweden
Support: Wellcome Trust, London; ESHG, Wien

Programme

Friday, June 11th

9.30 a.m.	Coffee and registration
10.30 a.m.: Session 1	***From basic molecular biology to human molecular genetics***
Yapijakis, Christos (Athens, Greece)	Ancestral concepts of human genetics and molecular medicine in Epicurean philosophy.
Chadarevian, Soraya de (Los Angeles, USA):	Hemoglobin and human molecular genetics.
Witkowski, Jan (Cold Spring Harbor, USA):	Cold Spring Harbor and the beginnings of human molecular genetics.
12.30 - 1.30 p.m.	Lunch
1.30 p.m.: Session 2	***From DNA analysis to human genetic disease***
Maniatis, Tom (New York City, USA):	Recombinant DNA technology and human molecular genetics.
Read, Andrew (Manchester, UK):	Technology and the development of clinical molecular genetics.
Lestienne, Patrick (Bordeaux, France):	The mitochondrial genome: historical aspects.
3.30 - 4.00 p.m.	Tea and Coffee Break
4.00 p.m.: Session 3	**From pedigrees to the human genome**
Friedman, Judith (Victoria, Canada):	A brief history of the theory of anticipation in hereditary disease.
Pollock, Ludmila (Cold Spring Harbor, USA):	Documenting the history of the Human Genome Project. An international data repository.
5.30-6.15 p.m.: Discussion	How can we best preserve the history of human

	molecular genetics?
7.30 p.m.	**Workshop Dinner**

Saturday June 12th

9:00: Session 4	***Genetic testing and prenatal diagnosis***
Loukopoulos, Dimitris (Athens, Greece):	Thalassaemia: genetic testing and prenatal diagnosis
Deltas, Constantinos (Nicosia, Cyprus):	Founder mutations, heterozygous advantage and thalassaemia in Cyprus.
Petermann, Heike (Muenster, Germany):	The 'special' situation of genetic testing and prenatal diagnosis in Germany. The influence of history.
10.30-11.00 a.m.	Tea and Coffee Break
11.00: Session 5	***Gene mapping and isol*ation**
Povey, Sue (London, UK):	The Human Gene Mapping Workshops 1973-1991.
King, Mary-Claire (Seattle, USA):	The revolution in understanding human molecular evolution: Work in the laboratory of Allan Wilson, 1964-1991.
Harper, Peter (Cardiff, UK):	Historical lessons from Huntington's disease.
1.00-1.45 p.m.	Lunch
1.45 p.m.: Session 6	***From human genetics to genetic medic*ine**
Bentsson, Bengt-Olle (Lund, Sweden):	Film from the 1956 First International Human Genetics Congress, Copenhagen.
Bodmer, Walter (Oxford, UK):	The beginnings of clinical cancer genetics.
Harper, Peter (Cardiff, UK)	Planning of future workshops, and conclusion of Workshop

5th International Workshop on the History of Human Genetics.

The Biological Future of Man. Continuities and Breaks in the History of HumanGenetics before and after 1945

Date: June 21-23, 2012
Place: Nuremberg, FRG
Support: German Research Foundation (DFG); European Society of Human Genetics (ESHG)

Programme

Thursday, June 21

6.30 —8.00 p.m.	**Introduction to Documentation Centre:** Hans-Christian Taeubrich, Director Documentation Centre **Visit of the Exhibition**

Friday, June 22

9.00 a.m.: **Lecture 1**	
Roll-Hansen, Nils Oslo, NOR)	Eugenics and the Science of Genetics.
10.15 a.m.: Session 1	***Human Genetics before 1945***
Rushton, Alan (Flemington, USA)	Charles Eduard of Saxe-Coburg: The Nobility, the Red Cross and the Nazi Eugenic Program
10.45 – 11.15	Tea and Coffee Break
11.15 a.m.: Session 2	
Pascacio-Montijo, Yuriditzi (Bielefeld, FRG)	The IQ test and its hereditary explanation: a systematized measure that transits from the scientific to the social.
Wilson, Philipp (Hummelstown,	"Pedigrees and Prejudices: Pre-WWII Inherited Disease

USA)	Classification at the U.S. Eugenics Record Office"
12.30 p.m.	Tea and Coffee Break
2.00 p.m.: Session 3	
Friedman, Judith (Rockville, USA)	The increasing divide between clinical and theoretical approaches to the study of hereditary disease before the Second World War
Huijnen, Pim (Utrecht, NL)	Genetic and eugenic thinking in public discourse in the Netherlands and Germany, 1900-1945
3.00 p.m.	Tea and Coffee Break
3.30 p.m.: Session 4	***Continuites in the History of Human Genetics***
Germann, Pascal (Zürich, CH)	„Nature's Laboratories of Human Genetics". Alpine Isolates, Hereditary Diseases and the Swiss Trajectory of Medical Mendelism, 1920-1970.
Noguera-Solano, Ricardo (Mexiko, Mx)	Genome: Twisting stories with DNA
4.30 p.m.: Lecture 2	Weindling, Paul: The Nuremberg Trials and Their implications for human Genetics.
7.30 p.m.	**Workshop Dinner**

Saturday, June 23

9.00 a.m.: Lecture 3	
Kolb, Stephan (Nuremberg, FRG)	Informed consent – an Essential of Medicine. Consequences of the Nuremberg Doctor´s Trail.
9.45 a.m.: Session 5	***Human Genetics after 1945***
Doetz, Susanne (Berlin, FRG)	Human genetics in a socialist society.
Scholz, Christine (Muenchen, FRG)	Human Genetics in transition. Professionalization of Human Genetics under Changing Structural Conditions in Germany during the last 50 years.
10.45 a.m.	Tea and Coffee Break
11.15 a.m.: Session 6	
Aspin, Richard (London, UK)	'Foundations of Modern Genetics': a new on-line resource for advanced historical research in the field
11.45 a.m.: Session 7	
Petermann, Heike (Muenster, FRG)	The Biological Future of Man: Continuities and Breaks in the History of Human Genetics in Germany, Before and After 1945. The Future of the Workshop?
12.30 – 14.00	**Lunch**

6th International Workshop on the History of Human Genetics

HUMANE GENE MAPPING. - ORAL HISTORY OF HUMAN GENETICS

Date: June 4-6, 2015
Place: Glasgow (UK)
Support: *European Society of Human Genetics* (ESHG), Wien

Programme

Thursday, June 4

6.00 – 8.00 p.m.	**Showcase Display & Behind the Scenes Tour, University of Glasgow Archive Services** **LOCATION:** 13 THURSO STREET, GLASGOW, G11 6PE

Friday, June 5

9.00 a.m.	Opening
9.15 a.m.: Session 1	***Human Genetics in Glasgow***
Ferguson-Smith, Malcolm (Cambridge, UK)	Glasgow contributions to the human gene mapping project, 1959-1987
Blair, Paula (Glagow, UK)	Pontecorvo's Legacy
O'Dell, Kevin (Glasgow, UK)	James Renwick: the First Human Genetic Maps
Monckton, Darren (Glasgow, UK)	Glasgow 2015 and beyond
11.00 a.m.	Tea and Coffee Break: **Poster Viewing**
11.30 a.m.: Session 2	***Human Gene Mapping***
Rushton, Alan (Flemington, USA)	The First Human Genetic Map 1936
McGovern, Michael (Chicago, USA)	'The London / Baltimore link has been severed': Human Linkage Mapping and the Early Computerization of Genetics
Hogan, Andrew (Omaha, USA)	The Thrill of Mapping: Bridging the Gap in Post-war Human Genetics
1.00 – 2.00 p.m.	Lunch: **Poster Viewing**
2.00 p.m.: Session 3	***History of Human Genetics (1)***
Capocci, Mauro (Rome, IT)	Unravelling the Complexity of HLA: Genesis and Success of the International Histocompatibility workshops
Friedman, Judith (Edmonton, CAN)	The Enduring Puzzle of Leber's Hereditary Optic Neuropathy
Pyeritz, Reed (Philadelphia, USA)	A Brief History of Uncertainty in Genomic Medicine

3.30 p.m.	Tea and coffee Break: **Poster Viewing**
3.45 p.m.: Session 4	***History of Human Genetics (2)***
Birmingham, Karen (Bristol, UK)	Marcus Pembrey Recalls the Catalyst for the Establishment of the International Federation of Human Genetic Societies
Tupasela, Aaro M. (Copenhagen, DK)	Critical Inquiry into Rare Disease Research in Finland: Finnish Disease Heritage in a Broader Historical Context
Mahr, Dominik (Lübeck und Bielefeld, FRG)	Narrating 'Geneticization': Living Your Genome in Shifting Scientific Paradigms
5.15 – 5.30 p.m.	Tea and Coffee Break: **Poster Viewing**
5.30 p.m.: Session 5	***History of Human Genetics (3)***
Houwink, Elisa (Maastricht, NL)	The History of Human Gene Mapping: Remembering the Times of PCR and Discovery of the MECP2 Gene Mutation Behind Rett syndrome at UCLA and Translation of Genetic Competences to Primary and Secondary Care PCR/MECP2
Simunek, Michal V. (Prag, CZ)	Project Documenting the Development of Medical Genetics in Czechoslovakia after 1945
7.30 p.m.	**Workshop Dinner**

Saturday, June 6

9.00 a.m.: **Lecture**	
Tansey. Tilli (London, UK)	You're All History Now: Recording the Voices of Modern Genetics
9.45 a.m.: Session 5	***Oral History (1)***
Harper, Peter (Cardiff, UK)	Interviews with Human and Medical Geneticists
Petermann, Heike (Muenster, FRG)	Reflections on Ethical and Theoretical Aspects of Oral History of Human Genetics in Germany
10.45 a.m.	Tea and Coffee Break: **Poster Viewing**
11.00 a.m.: Session 6	***Oral History (2)***
Donohue, Christopher (Bethesda, USA)	The Oral History Initiative at the National Human Genome Research Institute (NHGRI)
Doetz, Susanne (Berlin, FRG)	The Use of Oral History to Explore the Establishment of Genetic Counselling in the GDR during the 1970s and 1980s
Garcia-Sancho, Miguel (Edinburgh, UK)	A Critical Triangulation: the Combination of Archival Sources and Oral Histories in the Investigation of Contemporary Genetics
12.45 – 13.30	Further Projects
	Close of Workshop

Poster:

1	Barahona, Ana (Mexiko, Mx)	Medical Genetics in Mexico: the Origins of Cytogenetics and the Health Care System
2	Morfakis, Constantinos (Athen, GR)	Human Gene Mapping: The Mass Media Iconography of Human Genome Project in the Most Popular Greek newspapers
3	Petermann, Heike (Muenster, FRG)	Changing the Point of View: the History of Human Genetics as an Applied science in the Federal Republic of Germany from 1945 to 1975
4	Sloyan, Victoria (London, UK)	Collecting Genomics at the Wellcome Library
5	Tansey, Tilli, Emma M. Jones (London, UK)	Witnesses to Medical Genetics
6	Tansey, Tilli, Emma M. Jones (London, UK)	Mapping the Gene Mapping Workshops
7	Van El, Carla (Amsterdam, NL)	Neonatal Screening: a Historical-comparative Perspective
8	Petermann Heike (Muenster, FRG); Friedman, Judith (Edmonton, CAN)	*Publication of the History Workshops*

International Workshop: The Establishment of Genetic Counselling in the Second Half of the 20th Century

Date: February 2-3, 2016
Place: Berlin (FRG), Institute of the History of Medicine and Ethics in Medicine
Support: *Deutsche Forschungsgemeinschaft* (DFG)

Programme

Tuesday, February 2

8:45-9:00	Come together
9:00 -9:15	Welcome Volker Hess and Susanne Doetz
9.15 a.m.: **Session 1**	***Genetic Counseling in Europe and the USA: International Case Studies*** **Panel I: Genetic Counseling in the Mediterranean Area**
Luc Berlivet (France)	Genetic Counseling as a Eugenic Device. The "Fight Against Thalassemia" in 1950s Italy and Afterwards
Alexandra Barmpouti (UK)	Genetic Counseling for Mediterranean Anaemia in Post-War Greece (1950-1980)
10.15 a.m.	Tea and Coffee Break
10.30 a.m.	**Panel II: Country Case Studies**
Maria Björkman/Anna Tunlid (Sweden)	Development of Genetic Counseling in Sweden 1950-1980
Joris Vandenriessche (Belgium)	Genetic Counseling in Belgian Academic Hospitals, 1960-1980
Katja Geiger/Thomas Mayer (Austria)	The Establishment of Human Genetic Counseling in Austria during the 1970s in between the Formation of Human Genetics and the Eugenic Indication of Abortion
Heike Petermann (FRG)	Genetic Counseling in the United States of America and the Federal Republic of Germany (1945 to 1974). A Comparative Perspective.
01.00 p.m.	Lunch
02.00 p.m.	**Panel III: Shaping the Development of Genetic Counseling in the US: Crosscurrents of Professionalization, Uncertainty, and Disability**
Robert Resta (USA)	Colleagues, Conflicts, and Conciliations: Genetic Counsellors, Medical Geneticists, and the Historical Arc of The Genetic Counseling Profession
Marion Schmidt (USA)	From Preventing Defect to Serving a Disadvantaged Minority: Genetic Counseling for Deaf People
Andrew J. Hogan (USA)	Managing a Marginal Diagnosis: Genetic Counseling and the Expansion of Prenatal Testing
Adam Turner (USA)	Genetic Counsellors and Parent Advocates on Abortion and Disability, 1950-1990

04.30 p.m.	Tea and Coffee Break
05.00 p.m.	**Panel IV: Genetic Counseling behind the Iron Curtain**
Michal Simunek (Czech Republic)	Genetic Counseling in the CSSR
Susanne Doetz (FRG)	"The Happiness of the Individual is of Primary Importance" - Genetic Counseling in the GDR
Jean Paul Gaudillière (France)	Comment
08.30 p.m.	**Dinner**

Wednesday, February 3

09.30 a.m.: **Session 2**	***Genetic Counseling: Actors, Practice, and Methods***
	Panel I: Actors
Yechiel Michael Barilan/Margherita Brusa (Israel)	Expanded Newborn Screening: Genetic Counseling at the Level of Public Health through the Prism of three Historical Case Studies
Mauro Capocci (Italy)	Catholic Counseling. Medical Genetics and the Church Approach
Birgit Nemec (FRG)	Risk, Prevention and Counseling in Human Genetic Screenings - Western Germany 1945-1980
Gabriele Moser (FRG)	Abortion and Sterilisation in Mecklenburg after WWII: Family Planning between Social Needs and Eugenics
Jörg Pittelkow (FRG)	Herbert Bach (1926 – 1996) – A Pioneer of Human Genetics in East Germany (GDR)
12.00 – 01.00 p.m.	Lunch
01.00 p.m.	**Panel II: Practice and Methods**
Ana Barahona (Mexico)	Karyotyping and Genetic Counseling in Mexico in the 1960s
Shachar Zuckerman (Israel)	Challenging the Feminist Criticism of Genetic Counseling
Angus John Clarke (UK)	Evolving Ideas around 'non-directiveness' in Genetic Counseling
02.50 – 03.15 p.m.	Tea and Coffee Break
Von Schwerin, Alexander (FRG)	Final Comment
03.45 – 04.00 p.m.	**Final Discussion**

Reference

Jones, Emma M., E M Tansey (eds): (2015): Human Gene Mapping Workshops, ca. 1973 – c. 1991. London: Queen Mary University London. (Wellcome Witnesses to Contemporary Medicine. 54)

Part II
Beginning of Human Genetics

Ancestral Concepts of Human Genetics and Molecular Medicine in Epicurean Philosophy

Christos Yapijakis

Abstract Human genetics and molecular medicine are scientific fields that evolved during the last century. Nevertheless, less known is the fact that over two millennia ago mankind had grasped the concepts of the molecular basis of life in health and disease, in addition to the basic laws of heredity. It was the influence of the Epicurean philosophy that led some exceptional people of the ancient Hellenistic and Roman eras in observing human nature and proposing some notions that were discovered as scientific facts only recently. The founder of this humanistic philosophy was Epicurus of Athens (341–270 BC) who combined the atomic physics of Democritus with the naturalistic ethics of Aristotle. Epicurus suggested that eternal atoms ("atomoi") continuously combine by necessity and chance forming "bodies" ("onkoi") or molecules which produce worlds, mountains and evolving living organisms. He proposed that any given arrangement of atoms within a molecular structure confers new qualities to the molecule. Unlike Aristotle who believed that only males contributed in heredity, from observation of families Epicurus inferred that males and females equally contributed hereditary material to their progeny. According to the Roman Epicurean philosopher Lucretius (95–45 BC), Epicurus described dominant, recessive and co-dominant types of inheritance. The Epicurean physician Asclepiades of Bithynia (124–40 BC) suggested that diseases are caused by alteration of form or position of a patient's molecules. He introduced the psychological support of all patients as well as the distinction of diseases into acute and chronic ones, based on an observation of Epicurus regarding acute and chronic pains. One of the followers of Asclepiades' molecular medicine was the Greek physician Soranus of Ephesus (first to second century AD), known as the father of gynaecology and paediatrics. He described congenital malformations as

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well as hereditary conditions such as mental disorders in materialistic terms without any supernatural prejudice.

Keywords Empiricism • Naturalism • Materialism • Epicurus • Lucretius • Asclepiades of Bithynia • Soranus

1 Introduction

It is well known that human genetics and molecular medicine are scientific fields that evolved rapidly during the twentieth century.[1] Nevertheless, less known is the fact that over two millennia ago humankind had grasped some of the basic concepts of the molecular basis of life in health and disease, in addition to the basic laws of heredity. It was the influence of Epicurean philosophy that led some exceptional people of the ancient Hellenistic and Roman eras to observe the human nature and suggest some notions that were established as facts only recently (Fig. 1). In the extant texts of Epicurus of Athens (341–270 BC), Lucretius Carus (99–55 BC), Asclepiades of Bithynia (124–40 BC) and Soranus of Ephesus (70–140 AD), there are several ancestral concepts of chemistry, molecular biology, physiology,

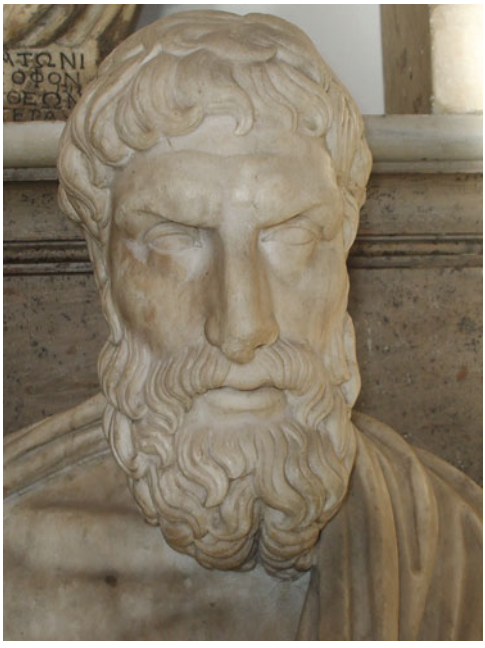

Fig. 1 Epicurus of Athens (Room of Philosophers at the Capitoline Museum, Rome, Italy). Source: Author

[1] Harper 2008.

microbiology, molecular medicine, human genetics and cognitive psychology. These matters will be discussed in the following text.

2 Origin of the Scientific Naturalistic Methodology

Science originates from philosophy (φιλοσοφία, "love for wisdom" in Greek). Religious and magical beliefs dominated human minds for millennia, until philosophy started to flourish in the Greek world in the sixth century BC.[2] Thales of Miletus started a long tradition of attempts to explain nature using experience based on senses, as well as imagination based on empirical analogies and rational assumptions.[3]

Eventually, two main philosophical lines emerged, usually referred as idealism and materialism. In his work "Critique of pure common sense (Kritik der reinen Vernunft)" (1781), the German philosopher Immanuel Kant discussed the two main philosophical lines, naming them Intellectualismus (intellectionalism, rationalism) exemplified by philosophers Plato of Athens and German Gottfried Wilhelm von Leibnitz, and Sensualismus (empiricism, sensationalism) exemplified by philosophers Epicurus of Athens and Englishman John Locke. The methods of acquiring true knowledge (gnosiology) used by the two philosophical paradigms were entirely different. Platonic intellectionalism used logical thinking or discussion (under the name "science") which claimed to be involved with universal concepts, while Epicurean empiricism used naturalism, namely empirical observation of nature. Kant remarked that Aristotle of Stagira had used the empirical observation method before Epicurus, but the Stagirite philosopher chose not to use it always. For example, Aristotle was "rationally" convinced that females were inferior to males of all animal species, so he wrote in his work "On animal histories"[4] that "males have more teeth than females in humans, sheep and pigs", a mistake that could have easily avoided by unbiased observation.[5]

The platonically derived distinction of "science" versus naturalism may surprise a modern scientist, but it should be mentioned that Kant wrote his essay over two decades before the Englishman John Dalton measured the weight of "atoms" of elements like hydrogen, oxygen and carbon in 1803 proving that Epicurus' atomic physics was correct. It is well known that since then, in the last two centuries science has advanced through empirical and experimental observation.

In addition, the Kantian approach to "science" by logic alone has influenced most modern philosophy scholars who treated Epicurus as a rational intuitionalist and failed to view him as he truly was, namely an objective naturalist.[6] Before

[2]Farrington 2000.

[3]Farrington 2000.

[4]Aristotle. Τῶν περί τά ζῶα ἱστοριῶν (On animal histories) A 501b.

[5]Mayhew 2004.

[6]Yapijakis 2016.

further mentioning Epicurean naturalism, it would be interesting to discuss Aristotle's contributions in naturalism.

3 Aristotle and Biology

Aristotle of Stagira (384–322 BC) is considered to be one of the most influential philosophers in history. He was interested in a variety of subjects and composed extensive works in philosophy, ethics, politics, logic, art, biology, psychology, physics and metaphysics. Aristotle studied philosophy in Academy, the School of the idealist philosopher Plato of Athens, but he had previously studied medicine with his father Nicomachus, who was a Hippocratic physician. Thus, the medical training in observation of patients' symptoms enabled Aristotle to disregard most Platonic theories about the invisible world of "immaterial ideas", since the Stagirite wrote about Plato and truth: "Where both are friends, it is right to prefer truth".[7]

Aristotle is considered as the father of empirical observation of nature as well as the father of several scientific fields, including zoology and systematic biology. He described the concept of species (εἶδος, "eidos" in Greek) and established it as a basic unit of classification and taxonomy. He described a variety of animals and established the biological basis of ethics and psychology. His School, named Lyceum after the Athenian gymnasium in which it was located, continued to exist for several centuries after him.

Despite his many advances, Aristotle made several mistakes, because he relied more on his common sense beliefs and more on logic as method of inquiry than observation.[8] He believed that the metamorphosis and decomposition phenomena resulted from a "vital force" existing within organic substances. Aristotle believed that living organisms had a timeless, unchanging and absolute essence; therefore, he did not accept evolution. Instead, he believed that everything in nature exists for a purpose advocating teleology (from Greek τέλος, "telos" meaning end or purpose). Aristotle thought that all living organisms were animals; therefore, for him plants were just degenerate animals. His disciple and successor as head of Lyceum Theophrastus of Eressus (c.371–287 BC) studied plants extensively and correctly mentioned that they constituted a different group of living organisms; therefore, he became known as the father of botany. Nevertheless, even the Eressian philosopher made curious mistakes, under the influence of Aristotle's "vital force" theory. For example, in his work "On stones", Theophrastus mentioned that "certain mineral crystals are possibly living organisms because they displayed growth", "they could be distinguished in males or females" and "they gave birth to offspring".[9]

[7] Aristotle. Ἠθικά Νικομάχεια (Nicomachean Ethics) 1096a15.
[8] Mayhew 2004.
[9] Duffin 2005.

4 Epicurus and Naturalism

According to his biographer Diogenes Laertius, Epicurus of Athens (341–270 BC) studied philosophy with the Aristotelian Praxiphanes and with the Democritean Nausiphanes,[10] before he established his own School in Athens, named Kepos (Κῆπος, in Greek meaning Garden). He combined Democritus' atomic physics ("everything is consisted of atoms and void") and Aristotle's biological ethics (based on knowledge acquired by senses). Epicurus corrected the mistakes of the two previous philosophies based on observations and he managed to form a humanistic philosophy that spread in the Hellenistic and Roman worlds influencing many thousands of men and women regardless of age, nationality or social class, including free people and slaves.[11]

Epicurus taught that the study of nature (φυσιολογία, "physiology", science in modern terminology) is an important means for happiness (εὐδαιμονία, eudaimonia) of all people. The Athenian philosopher favoured objective and unbiased observation of nature claiming that only in this way the human mind could be enlightened and free of illogical fears, so as to facilitate tranquility and happiness.[12] He insisted that "we should not study nature with empty axioms and arbitrary laws but as phenomena require. Because our life does not need illogical and foolish opinions, but it needs tranquillity".[13] Several Epicurean texts attest that Epicurus did not trust the myths ("Only the myth must stay away from us"[14]), nor the dialectic method ("The Epicureans reject dialectic. Because it suffices for physicists to promote their thoughts according to words that correspond to natural things"[15]), nor the rhetoric method ("It is useless, because it is so obvious, to continuously demonstrate that sciences do not change in various locations while rhetoric seems altered in different countries and cities"[16]).

Epicurus was very much interested in observing and understanding nature, as he attests in his writings: "I recommend constant activity in the study of nature and this way more than any other I bring calm to my life".[17] For this purpose, he invented the Canon (Κανών, meaning "ruler" or "measuring stick" in Greek), an empirical methodology of inquiry consisting of observation by the senses (following Aristotle) and drawing inferences about the unknown based on analogies with the observed. This approach made Epicurean philosophy very comprehensive and among all ancient philosophies by far the most compatible with modern scientific findings.

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[10]Diogenes Laertius. Βίοι καὶ γνῶμαι τῶν ἐν φιλοσοφίᾳ εὐδοκιμησάντων (Lives and Opinions of Eminent Philosophers) 10.13.

[11]Warren 2009.

[12]Warren 2009.

[13]Epicurus. Letter to Pythocles: Diogenes Laertius 10.87.

[14]Epicurus. Letter to Pythocles: Diogenes Laertius 10.103.

[15]Diogenes Laertius 10.31.

[16]Philodemus. Περί ῥητορικῆς (On rhetoric) II 105.

[17]Epicurus. Letter to Herodotus: Diogenes Laertius 10.37.

5 Epicurean Gnosiology

The Canon comprises two basic principles and four criteria of truth all stemming from experience.[18] The basic principle of creation of everything from seeds ("nothing comes from nothing") is supported by the observation that certain plants derive from certain seeds. The basic principle of analogy is deducted from the observation that the same natural laws apply for every material body.

The four criteria of truth include (a) the sensations, the most reliable basis of our knowledge of the world according to Epicurus, (b) the preconceptions, the mind-saved concepts that always derive from sensory experience, (c) the emotions, pleasant or painful, which inform us respectively about what is good or bad for our nature, and (d) the focusing thoughts into impressions, which correspond to mental pictures.

Epicurus was an empiricist and he maintained that the best way to understand nature was through our senses, since we developed them during evolution and we are adapted to live in this world. Every theory should be verified or falsified by witnessing of senses (by analogy to universal physical laws). Otherwise, it will be just probable until more observable evidence comes up. Every sensory impression is always correct according to Epicurus because it is spontaneously generated by environmental stimuli, it is not affected by previous similar ones or contemporaneous dissimilar ones, and only mental bias may distort it.

Epicurus was the advocate for multiple theoretical explanations of a phenomenon, if enough observational data were not available: "When someone accepts an explanation and dismisses another one, while they both explain a phenomenon, it is obvious that one both distances him/herself from naturalistic/scientific approach and retreats to myths".[19] Thus, he set the basis for experimentation to empirically distinguish between different theories, like modern scientists do.

It is very likely that Epicurus and his followers in the Garden conducted actual experiments, but their works have been lost. Nevertheless, it is known that Strato of Lampsacus, the third leader of Lyceum and Epicurus' contemporary in Athens for 20 years, definitively performed experiments.[20] As a result, Strato not only agreed with Epicurus that everything consisted of small bodies and void, but he also renounced Aristotle's teleological theories. The Hellenistic era saw the development of a scientific tradition that was based on empirical observations. The advancement of science and technology led to highly sophisticated instruments like the calculator mechanism of Antikythera[21] and the steam engine precursor mechanism of Heron of Alexandria that converted vapour pressure into circular power, almost 2000 years before the Industrial Revolution.[22]

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[18] Warren 2009.

[19] Epicurus. Letter to Pythocles: Diogenes Laertius 10.87.

[20] Yapijakis 2009.

[21] Charette 2006.

[22] Tassios 2002.

Using the Canon, observation, analogical thinking and possible experimentation, Epicurus was able to confirm that nature is composed from atoms and void. He taught that eternal atoms continuously combine by necessity and chance forming worlds, mountains and evolving living organisms, earth is a spherical world, sun and stars are spheres of fire, and there are infinite number of worlds (κόσμοι, "cosmoi") similar or unsimilar to earth.[23] The Epicurean philosophy introduced several notions that were reaffirmed by scientific inquiry in the last four centuries: the weight of atoms[24] (ἄτομοι, "atomoi" in Greek meaning indivisible particles), the emerging new properties of chemical substances based on their atomic structure,[25] the multitudes of worlds in the universe,[26] the atomic nature of our sense perception,[27] the evolution of species based on natural selection,[28] the molecular basis of disease,[29] free will,[30] justice as a social contract,[31] progress of civilization[32] and many other notions, including the existence of extraterrestrial life,[33] which science is still investigating.

6 Epicurean Chemistry

Epicurus managed to combine the reality of Democritus' atomic physics with the reality of Aristotle's observational biology, correcting each other with the empirical method he invented, Canon. Thus, the Athenian philosopher was able to create the bridge of composite bodies that have new properties not found in simple atoms, in other words the reality of molecules, the reality of Chemistry. Epicurus addressed the old philosophical problem of "being" and "becoming", with the notion that eternal atoms always exist, while they give form to composite bodies (molecules) with a limited life span. According to Epicurean philosophy, eternal nature consists of atoms and void space, the sum of all matter is conserved, but atoms are perpetually used by necessity and chance in an endless process of construction and decay of composite material bodies.[34]

[23]Epicurus. Letter to Herodotus: Diogenes Laertius 10.35-83.

[24]Epicurus. Letter to Herodotus: Diogenes Laertius 10.54.

[25]Epicurus. Letter to Herodotus: Diogenes Laertius 10.54-55.

[26]Epicurus. Letter to Herodotus: Diogenes Laertius 10.45.

[27]Epicurus. Letter to Herodotus: Diogenes Laertius 10.49,52,53; Lucretius. De rerum natura IV615-628, 642-662.

[28]Lucretius. De rerum natura V828-834, 855-859.

[29]Yapijakis 2009.

[30]Epicurus. Letter to Menoeceus: Diogenes Laertius 10.133; Lucretius. De rerum natura II251-293.

[31]Kyriai Doxai XXXI,XXXII,XXXIII: Diogenes Laertius 10.150.

[32]Lucretius. De rerum natura V1011-1457.

[33]Lucretius. De rerum natura II1072-1076.

[34]Warren 2009.

Epicurus proposed that any given arrangement of atoms within a molecular structure confers new qualities to the molecule: "We have to accept that atoms do not have any other quality than shape, size and weight as the phenomena attest. Because indeed while the atoms do not change at all, the complex bodies that they form have different qualities than the atoms. The quality of a complex body changes when its atomic structure changes".[35] The Epicurean concepts of matter conservation as a basic law of nature, as well as the emersion of new properties in a molecule due to its atomic structure, were duly recognized by the first chemist Robert Boyle in his work "Atomicall Philosophy",[36] although he later made efforts in his works "The Skeptical Chymist" and "Christian Virtuoso" to renounce Epicurean philosophy.[37]

Epicurus discussed about "bonds of atoms within a molecule" (chemical bonds). Following in his steps, the Epicurean Roman Lucretius (95–55 BC), author of the philosophical poem "De Rerum Natura" ("On the nature of things"), described atoms as tiny spheres attached to each other by fishhook-like arms.[38] Lucretius wrote that two atoms combine with each other, when their hooked arms become entangled. Lucretius refers to solid, liquid and gas bodies attributing their compact or fluid nature in their atomic structure. He mentions characteristically: "The bodies that appear to us hard and compact probably consist of atoms more hooked among them, composing complex branches".[39] The Epicurean theory of atoms with hooked arms was practically how chemists viewed atoms until the beginning of the twentieth century.

7 Epicurean Biochemistry

Epicurus was the first philosopher who discussed concepts of biochemistry and molecular biology. Since the arrangement of atoms in molecules allows the emergence of new qualities, he inferred that life is a quality manifested by certain composites of atoms.[40] According to the analogy principle of the Epicurean Canon, the same natural laws of necessity and chance that characterize chemistry of lifeless inorganic matter, the same apply for chemistry of living organic matter, biochemistry, as well.[41] In the middle of the twentieth century, Watson and Crick described DNA, the life molecule that contains all genetic information for structure

[35]Epicurus. Letter to Herodotus: Diogenes Laertius 10.54.
[36]Boyle 2000: Vol. 13, 227.
[37]Wilson 2008.
[38]Gillespie and Hardie 2007; Lezra and Blake 2016.
[39]Lucretius. De rerum natura II 444-446.
[40]Epicurus. Letter to Herodotus: Diogenes Laertius 10.63-67.
[41]Wilson 2008.

and function of a living organism, in accordance with Epicurus' notion of the atomic/chemical origin of life.

8 Epicurean Biology

Aristotle laid the basics of biology by the description of numerous animal species, but he believed in natural end goal of everything (teleology); therefore, he thought that the animal forms were fixed. Epicurus suggested in his main scientific book "On nature" that due to ceaseless change in natural conditions everything evolves, including living organisms. Lucretius describes poetically the continuous change in nature: "With the passing of time, the nature of the whole world necessarily changes and nothing stays the same. Everything evolves, nature changes all things and makes them transformed".[42]

The evolution happens through natural selection: "Numerous species of animals must have been extinct, since they were not able to strengthen their kind with proliferation. Because whatever creatures you see now to breathe the life-giving air, they on their own assured their survival by cunning, by bravery, or their speed".[43] It is well known that Charles Darwin knew well Lucretius' evolution theory, since his grandfather Erasmus Darwin was an admirer and imitator of the Roman philosopher.[44]

Since Epicurus supported the perpetual change of nature and the infinity of worlds in the universe, according to the principle of analogy, it would be only natural to propose the existence of extraterrestrial life. Lucretius' testimony is lucid: "If nature has the power to combine atoms everywhere, as it has done on this earth, then it follows that we have to admit that there are more earths somewhere in the universe and various other kinds of humans and animals".[45] Modern science is currently searching for living organisms in other planets, following the Epicurean analogical thinking.[46]

9 Epicurean Neurobiology

Epicurus described the atomic origin of environmental stimuli that senses apprehend and the muscle motion, in a way that is similar to modern molecular neurobiology.[47] The Athenian philosopher taught that microscopic bodies (atoms or

[42]Lucretius. De rerum natura V828-830.
[43]Lucretius. De rerum natura V855-859.
[44]Jackson 2009.
[45]Lucretius. De rerum natura II 1072-1076.
[46]Mayor and Queloz 1996, Brake 2006.
[47]Hyam et al. 2011.

molecules) come from the environment and interact with human sensory organs in order to allow awareness of the outside world. Humans, like all other animals, have evolved to live in this world; therefore, their senses are reliable and the basis of true knowledge about nature.

Epicurus proposed an atomic and molecular model for the senses. He described vision stimulated by "thin atomic images of objects",[48] hearing caused by "flow of molecular waves",[49] smell caused by "flow of molecular substances",[50] taste caused by "molecules of various shapes".[51] Some of these atomic stimuli are pleasant and others unpleasant, respectively, if they concur or not with human nature. Humans, like other animals, have a natural inclination for pleasure and avoidance of pain. Like all other composites of atoms, living organisms including humans follow the same natural laws of birth, development and decay. Human soul is a material composition of atoms, including a central part corresponding to mind and a peripheral part distributed in all body regions.

10 Epicurean Psychotherapy

Epicurus taught that the purpose of philosophy is to increase human happiness; otherwise, it is a useless endeavour. He mentioned that the right philosophy (based on naturalism) cures the anxieties of the soul in a similar manner that the right medicine cures the pains of the body.

Epicurus observed that we are naturally inclined towards pleasure, which is measured by the absence of pain. He defined happiness as a condition in which the body does not feel pain and the soul is not anxious.[52] He tried to free people from superstition and unsubstantiated fears of the unknown. He observed that there is chance in the world and no destiny; thus, the existence of chance atomic movements permits free will in people.[53]

He offered a four-part remedy (τετραφάρμακος, tetrapharmakos) for living a pleasant, virtuous, fearless life. Gods do exist but they are not concerned with people or the celestial bodies, which is why the world is so imperfect. It is absurd and unrespectable to be afraid of gods, instead of admiring them as examples of perfect happiness. Death destroys our soul and senses; therefore, as long as we live we will never experience it. Armed with the right philosophy, prudence and friendship, all necessary good is easy to achieve, while all bad is easy to endure.

The message of Epicurus was that all people (including poor men, women, even slaves) may achieve happiness if their way of living is based on prudence, virtue,

[48]Epicurus. Letter to Herodotus: Diogenes Laertius 10.46-50.

[49]Epicurus. Letter to Herodotus: Diogenes Laertius 10.52.

[50]Epicurus. Letter to Herodotus: Diogenes Laertius 10.52.

[51]Epicurus. Letter to Herodotus: Diogenes Laertius 10.56.

[52]Yapijakis 2013.

[53]Warren 2009; Gillespie and Hardie 2007; Lezra and Blake 2016.

justice, friendship and scientific knowledge. The Epicurean approach of psychotherapy appears to be based on human nature.[54] It reveals the absurdity of unsubstantiated fears by curing mental agitation with facts and aims at a serene blissful state. Modern existential cognitive psychotherapy, which seems to be more effective than other approaches, focuses on the identification of one's specific fears and negative thoughts revealing their absurd character and then proposes a systematic engagement with pleasurable activities.[55] Numerous recent studies have shown that people feel happier when they satisfy their basic needs and have meaningful relationships with relatives and friends regardless of economic or social status.

According to Epicurus, the blissful life includes the conscious realization that death ("the most horrible of all evils"[56]) is nothing to us, since death is the deprivation of sense experience.[57] The philosopher taught that the inevitability of death makes life enjoyable, since the prudent person does not waste time on irrational fears and unnecessary desires preferring a happier instead of a more lasting life, "as we choose the most pleasant food and not the largest amount of food".[58]

Recent studies have shown indeed that people who have not dealt with the fear of death consciously, experience many phases of anxiety and fail to enjoy their life.[59] Although they develop various subconscious defence mechanisms, these are not sufficient because repressed thoughts often surface. It seems that the best psychotherapy approach is the conscious treatment of the fear of death with recognition of the limits of human biological nature, i.e. the Epicurean approach. The message of this therapeutic approach can reach virtually any recipient due to the biological plasticity of the human brain.

11 Epicurean Human Genetics

Aristotle had noticed that "human gives birth to human and the plant gives birth to plant from matter corresponding to each organism".[60] Nevertheless, the Stagirite did not accept genetic contribution of females but instead he thought that only men contributed the hereditary material through sperm while the women conferred the necessary environment for development of the foetus, like the seed and the field, respectively. Unlike Aristotle who believed that only males contributed in heredity, Epicurus observed families and inferred that males and females equally contributed hereditary material to their progeny. Therefore, Epicurus was able to describe

[54] Yapijakis 2013.

[55] Strenger 2008.

[56] Epicurus. Letter to Menoeceus: Diogenes Laertius 10.125.

[57] Warren 2004.

[58] Epicurus. Letter to Menoeceus: Diogenes Laertius 10.126.

[59] Strenger 2008; Wegner 2009; Yalom 2009.

[60] Aristotle. Περὶ ζῴων μορίων (On the parts of animals) 646α 34.

dominant, recessive and co-dominant types of inheritance, most probably in his book "On nature". The relative passage from the Athenian philosopher's work was lost, but his Roman follower Lucretius saved a poetic description of this matter:

> As the male and female seeds combine, if the woman's strength happens to dominate the man's strength, then the children are born looking like their mothers. On the contrary, when the paternal seed dominates they look like their fathers. But those children that look like both their parents and combine their characteristics. . . neither of the two seeds was defeated or won. Sometimes the children take their characteristics from their grandparents, sometimes they look like their grand-grandparents, because in the bodies of the parents atoms hide which have been combined there with thousands of ways, atoms that sprang from ancestors and are inherited from parent to parent in generations.[61]

Over two millennia before Gregor Mendel, the Epicureans knew about the basic Laws of Heredity, including the Law of Segregation of genes ("seeds"), the Law of Independent Assortment and the Law of Dominance. That fact was observed by philosophically educated human geneticists, such as John B.S. Haldane.[62]

12 Epicurean Molecular Medicine

The Epicurean Greek physician Asclepiades of Bithynia (124–40 BC) was the first Greek physician who established medicine in Rome (Fig. 2).[63] Influenced by the Epicurean philosophy, he rejected Hippocrates' doctrines of four humours (based to the dogma of four elements) and the axiom of the "benevolent nature".[64] He adhered to atomic theory, chance and evolution. The Bithynian physician suggested that diseases are caused by alteration of form or position of a patient's molecules, introducing stereopathology.[65] He is considered the ancestral father of molecular medicine. Asclepiades was the first physician who introduced the highly important division distinction of disorders into acute and chronic ones.[66] Freed by the misconception of a benevolent nature and influenced by one of the principal sayings of Epicurus regarding pains ("those that are acute are more intense, while those that are lasting are milder"), Asclepiades recognized that some diseases have a short duration, while others are incurable. He realized that the physician has to act swiftly in order to have an opportunity to cure the acute diseases, while the best thing to do in incurable chronic diseases is to comfort the patients.[67] Asclepiades was the first to study chronic diseases systematically. It was only after him that the cure of

[61] Lucretius. De rerum natura IV: 1209-1222.
[62] Haldane 1954.
[63] Gumpert 1794; Green 1955.
[64] Yapijakis 2009.
[65] Yapijakis 2009; Gumbert 1794; Green 1955.
[66] Yapijakis 2009.
[67] Gumpert 1794; Green 1955.

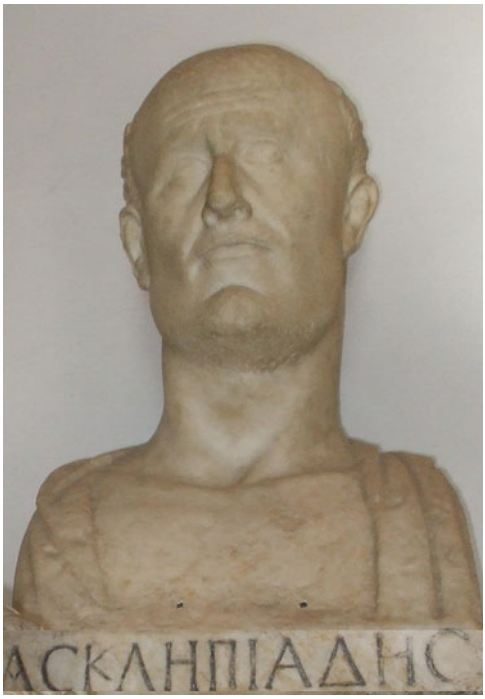

Fig. 2 Asclepiades of Bithynia (Room of Philosophers at the Capitoline Museum, Rome, Italy). Source: Author

chronic diseases rather than the cure of acute ones became the outstanding accomplishment of doctors.

He introduced the psychological support of patients. He was a pioneer in the humane treatment of patients with mental disorders, using labour and music therapy. One of his or his disciple's sayings in "Precepts" was: "Where there is love for Man there is also love for the Art".[68] His humane and naturalistic approach, as well as his medical skills, gave him a great reputation. Asclepiades' legacy continued though Methodic medicine (μέθοδος, "methodos" implies systematic scientific investigation). It was the only medicine of the ancient world that was based on atomic theory and was free of metaphysical ideas. It lasted for five centuries until the beginning of the Middle Ages, when Platonism merged with Christianity by theologians Basil and Augustine.

[68]Yapijakis 2009.

13 Epicurean Microbiology

Thucydides of Athens (c.460–c.400 BC) was the first scientific historian who did not mention supernatural or metaphysic interventions in his History of the war between democratic Athens and oligarchic Sparta. Thucydides described the symptoms of patients infected by the Plague of Athens so that future generations can figure the cause, an accomplishment achieved only a decade ago.[69] Epicurus had learned to respect Thucydides' historical narrations from his Aristotelian teacher Praxiphanes and probably used the Plague of Athens as an example of an epidemic of natural cause in his scientific text "On nature". That may explain the fact that three centuries later Lucretius ended his philosophical poem "On the nature of things" with a lengthy description of the Plague of Athens,[70] almost paraphrasing Thucydides.[71]

During the same period, the Epicurean physician Asclepiades probably suggested that in stagnant waters "invisible tiny animals" (microbes) live which if inhaled may cause disease, as documented by his contemporary Roman writer Terentius Varro.[72] Asclepiades might have grasped that concept using the analogical inference of the Epicurean Canon in order to explain the observations about sick people who had drunk stagnant water even after dilution in freshwater.

Fifteen hundred years later, when the philosophical poem of Lucretius was rediscovered during Renaissance, the Italian Fracastoro wrote about minute living entities ("semina") as the cause of infectious diseases ("pestilitas") using the terminology of the Roman Epicurean.[73] The existence of these small living organisms was verified only after the invention of the microscope by the Dutch Leeuwenhoek in 1674, while the microbes were proven to act as pathogens by experiments of the French Pasteur in 1875.

14 Gynaecology, Paediatrics and Clinical Genetics

One of Asclepiades' followers of the Methodic school of medicine was Soranus of Ephesus (70–140 AD), a Greek physician known as the father of gynaecology, obstetrics and paediatrics.[74] He is the author of famous books regarding these specialties. In his books, he described congenital malformations, including club-feet, dysmorphy, etc. In addition, Soranus is the author of the oldest known biography of Hippocrates.[75]

[69]Papagrigorakis et al. 2006.
[70]Lucretius. De rerum natura V 1138–1286.
[71]Gillespie and Hardie 2007.
[72]Yapijakis 2009.
[73]Fracastoro 1930.
[74]Todman 2008.
[75]Yapijakis 2009.

His medical approach is humane and methodical. He described as hereditary conditions of the "soul atoms" mental disorders such as melancholia (depression), paranoia and mania.[76] A genetic basis for these psychiatric conditions has been established only in the last decades of the twentieth century.

15 Influence of Epicurean ideas in Renaissance, Enlightenment and Modernity

After a millennium of imposed silence, the rediscovery of the philosophical poem of Lucretius during Renaissance made a great impact in disseminating the philosophy of Epicurus.[77] Several early scientists recognized the influence of Epicurean concepts in their work,[78] including Galileo (planets with satellites, 1632), Boyle (early chemistry, 1661), Newton (gravity, 1687) and Dalton (atomic weight, 1803). Several life scientists and physicians discovered biological mechanisms proposed by Epicureans, including Darwin (evolution of species by natural selection, 1852), Pasteur (life derives from life, microbes and disease, 1864), Mendel (laws of genetics, 1866), Freud (psychotherapy, 1899) and Garrod (molecular basis of disease, 1902).

Many philosophers of the Enlightenment were influenced by Epicurus, including Locke, and the French encyclopaedists.[79] One of the major political figures of the Enlightenment, Thomas Jefferson (34), author of the American Declaration of Independence, third president of the USA, and founder of the first public American university (University of Virginia) wrote: "I too am an Epicurean. I consider the genuine doctrines of Epicurus as containing everything rational in philosophy which Greece and Rome have left us".[80]

Several modern philosophers were influenced by Epicurus. Among them, the British Jeremy Bentham and John Stuart Mill should be mentioned. They developed Utilitarianism which advocates happiness for most people and has a firm basis on Epicurean philosophy. The German philosopher Nietzsche wrote: "The wisdom had taken many steps forward with Epicurus, but then it went many thousand steps backward".[81]

The same backward moves happened during the last two centuries. The dualism of Plato was introduced in university curricula and the studies have been divided into Humanities and Natural Sciences, as if the first are "Unnatural Sciences" and the latter "Inhumanities". The result is the production of humanities majors without scientific knowledge and scientists without philosophical training.

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[76] Gerdtz 1994.

[77] Greenblatt 2011.

[78] Warren 2009; Wilson 2008; Gillespie and Hardie 2007; Lezra and Blake 2016; Redondi 1987; Beretta 2008; Albury 1978.

[79] Wilson 2008; Lezra and Blake 2016.

[80] Mapp 1991, 295.

[81] Nietzsche 1884, § 45.

The ideas and methods for eugenic cleansing that Plato described in his "Republic" inspired the Nazi atrocities before and during the Second World War.[82] After the war, the term "eugenics" meaning the science of heredity, based on a word that Plato first used (εὐγονία, eugonia), was so much discredited that the word "genetics" (γενετική) had to replace it, a term first coined by Festetics in 1819.[83] And yet, most young geneticists are not aware of this recent historical fact.

In conclusion, the Epicurean philosophy includes many concepts that recent scientific research has verified. It was Epicurus' empirical methodology, Canon, which enabled him and other Epicurean philosophers and physicians to arrive at such astonishing insights. Epicurus deeply understood human nature and proposed a sound utilitarian ethical system based on prudence (φρόνησις), virtue (ἀρετὴ) and friendship (φιλία). His assertion that our well-being depends on how wisely we understand nature and our place in it seems of fundamental importance in an era characterized by socioeconomic crisis, environmental hazards and religious fanaticism. Humankind should listen to the Athenian sage if it wishes to prevent the worst of catastrophes.

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[82]Chapoutot 2008.

[83]Poczai et al. 2014.

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Bateson and the Doctors: The Introduction of Mendelian Genetics to the British Medical Community 1900–1910

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Abstract William Bateson of Cambridge was the leading proponent of Mendelian genetics in England after 1900. His studies demonstrated segregation of inherited characters in both animal and plant species. Bateson was also asked to evaluate pedigrees of families collected by physicians in which various diseases appeared to be transmitted from one generation to the next. Bateson and Archibald Garrod collaborated on the analysis of alkaptonuria, a rare disorder of metabolism, often found in individuals who had first-cousin parents. This was consistent with recessive inheritance. Other metabolic disorders such as albinism, cystinuria, and pentosuria also followed the Mendelian recessive pattern for inheritance. However, Bateson found that other human diseases did not exhibit such clear examples of Mendelian inheritance. The ophthalmologist Edward Nettleship sent him detailed pedigrees from families with a host of inherited eye diseases. Stationary night blindness, glaucoma, and cataract were dominant. Retinitis pigmentosa demonstrated two forms of inheritance. In certain families, a dominant pattern was evident, while in other examples, a recessive inheritance pattern was observed. Alfred Gossage also sent Bateson family histories of heterochroma irides, exostoses, myotonia congenita, cleidocranial dysostosis, and tylosis palmaris et plantaris; all demonstrated dominant inheritance. George Mudge of the London Hospital Medical College worked with Bateson on the heredity of eye color and then organized a course for the students at his college on inheritance in clinical practice. The two also collaborated to establish *The Mendel Journal* in October 1908 to publicize the importance of Mendel's laws for the understanding of inheritance in all living species. The Scottish physician Harry Drinkwater sent Bateson pedigrees with asthma and brachydactyly; all were consistent with the segregation of dominant traits. Redcliffe Salaman collected data from Jewish families with a rare neurologic

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disorder amaurotic family idiocy (Tay–Sachs disease). The reappearance of the character in an inbred population was consistent with a recessive segregation pattern. The Royal Society of Medicine sponsored a conference titled "Heredity and Disease" in November 1908. Bateson acknowledged the assistance of his medical colleagues in collecting well-documented pedigrees that indeed illustrated the Mendelian segregation of many human characters. He noted chorea, ectopia lentis, distichiasis, ptosis, and brachydactyly as examples of dominant traits. Albinism and alkaptonuria were often evident in consanguineous families as expected for recessive disorders. Hemophilia, muscular dystrophy, and color blindness followed the unusual pattern of affected males with unaffected female carriers that fit the pattern for sex-limited segregation. By the end of the first decade of the twentieth century, Bateson and his medical colleagues had successfully communicated in understandable terms the important role for Mendelian heredity in providing workable solutions to the riddle of human heredity.

Keywords William Bateson • Edward Nettleship • Human genetics • Medical genetics

In 1902, at the age of 57, Edward Nettleship (1845–1913), one of the leading ophthalmologists in England, decided to retire from clinical practice. In his extensive experience at the *Royal London Ophthalmic Hospital*, he had encountered several eye diseases which reappeared in successive generations. He decided to focus his retirement energies on collecting data from such affected families in an attempt to understand the role heredity might play in causing specific eye disorders.[1] Over the next few years, Nettleship did collect family histories showing inheritance of color blindness, cataract, albinism, and retinitis pigmentosa. But he was not well read in the science of heredity and was unable to analyze the data he had in hand. Nettleship received advice on his dilemma from a colleague who suggested: "You had better come and meet Mr. Bateson of Cambridge, who knows more about heredity than anyone".[2]

William Bateson (1861–1926) received training in vertebrate embryology at Cambridge and Johns Hopkins Universities. He was appointed Fellow at St. John's College Cambridge in 1887 and began a study on the origins of variations which acted in natural selection. His research in the late 1890s focused on how such variation could be transmitted from one generation to the next in a series of "systematic experiments in breeding".[3] He gathered a group of young students to assist in his studies on peas, poppies, and poultry.[4]

[1]Lawford 1922, ix–xv.

[2]P 330: Herringham to Nettleship, 1904.

[3]Bateson 1894, 514.

[4]Bateson B 1928.

Although Bateson accumulated a large body of breeding data showing the transmission of characters, he did not have a satisfactory theory of heredity to predict results of specific parental crosses. His need was suddenly filled in the summer of 1900 when Bateson became aware of the theoretical work done 35 years previously by the Austrian monk Gregor Mendel (1822–1884). Three continental plant breeders: Hugo de Vries (1848–1935), Carl Correns (1864–1933), and Erich von Tschermak-Seysenegg (1871–1962) all observed the mathematical precision with which Mendel's theory predicted the results of cross breeding.[5] Bateson immediately understood the potential importance of these ideas in elucidating his own work. Previously he had no "principle" of heredity; now he felt he possessed a "law" that could be validated experimentally.[6] Bateson was soon recognized as the "most ardent advocate of the new view on heredity in England." He believed that Mendel's work was as fundamental to biology as the atomic theory was to chemistry.[7]

Bateson had many friends in the medical profession. His wife Beatrice was the daughter of Arthur Durham (1834–1895), senior surgeon at Guy's Hospital in London. For many years, he had been friendly with Archibald Garrod (1857–1936), a physician at St. Bart's Hospital. Garrod had been studying human blood and urine chemistry, and Bateson noted its relevance to his own work on variation in a December 1901 report when he appended a footnote which marked the beginning of modern medical genetics: the application of heredity to human disease. Bateson used his breeding data to illustrate how a parent might carry a hidden trait, which would then appear in the offspring. He observed that:

> In illustration of such a phenomenon, we may perhaps venture to refer to the extraordinarily interesting evidence collected by Garrod regarding the rare condition known as "Alkaptonuria." In such persons the substance, alkapton, forms a regular constituent of the urine, giving it a deep brown colour, which becomes black on exposure [to air].
>
> The condition is extremely rare and, though met with in several members of the same families, has only once been known to be directly transmitted from parent to offspring. Recently, however, Garrod has noticed that no fewer than five families containing alkaptonuric members, more than a quarter of the recorded cases, are the offspring of unions of *first cousins*. In only two other families is the parentage known, one of these being the case in which the father was alkaptonuric. In the other case, the parents were *not* related. Now there may be other accounts possible, but we note that the mating of first cousins gives exactly the conditions most likely to enable a rare and usually recessive character to show itself. If the bearer of such a gamete mates with individuals not bearing it, the character would hardly ever be seen, but first cousins will frequently be bearers of *similar* gametes, which may in such union meet each other, and thus lead to the manifestation of the peculiar recessive characters in the zygote [fertilized ovum].[8]

Here was a mechanism to explain why inbreeding (consanguinity) seemed to increase the frequency of certain hereditary conditions.

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[5] Olby 1987.

[6] Darden 1977.

[7] Dunn 1991, 64–65.

[8] Bateson 1902.

Bateson and his friend collaborated on a detailed study of the heredity of this chemical variation. Garrod noted early in 1902, "The subject interests me greatly in its bearing upon chemical as distinguished from structural variations, and it seems to me that alkaptonuria, cystinuria and perhaps albinism also are chemical analogues of malformations." He was also very interested in the heredity of such chemical variants. He wondered what offspring might result from the "union of two potentially alkaptonuric strains." He was "afraid that [. . .] a marriage of alkaptonurics is very unlikely to occur, nor do I see any way of introducing any marriageable alkaptonurics to each other with a view of matrimony!"[9] But in more whimsical moments, he may have hoped for just such an event to occur. One of his medical colleagues remembered that Garrod had several young alkaptonuric patients in the London area. He would often admit them to hospital at the same time for demonstration to medical students, and in the hope that they might one day find each other attractive and marry. Such a guided experiment of nature might have produced some interesting results.[10]

Garrod analyzed forty cases of alkaptonuria in his 1902 paper on "Chemical Individuality." He noted that 60% of affected families had first-cousin parents, while the general population at that time had 2–3% such parentage. He reported that the familial pattern for both albinism and alkaptonuria was identical. The trait could remain latent for several generations, but then reappear in the "offspring of the union of two members of a family in which it is transmitted." Garrod observed that Mendel's laws of heredity appeared to offer a "reasonable account of such phenomena."[11] This paper in *Lancet* marked the introduction of Mendelism to the British medical literature (For a general overview of this topic, see Harper 2008 and Rushton 2009.).

Garrod continued his studies of chemical variation and heredity in man and presented the *Croonian Lecture* at the Royal College of Physicians in June 1908. His title "Inborn Errors of Metabolism" summarized his connection of heredity, biochemistry, and medicine to elucidate the mechanism of disease resulting from inherited altered body chemistry. He repeatedly used Mendel's theory of heredity to illustrate the inheritance of altered metabolism in families with alkaptonuria, cystinuria, albinism, and pentosuria.[12]

Bateson continued to struggle to relate Mendel's work to other human characters, but was dismayed with the difficulties he encountered. His relationship with the ophthalmologist Nettleship would prove mutually beneficial to both, as the doctor learned something about heredity, and the biologist learned something about working with humans as experimental subjects.

Bateson invited him to come to Cambridge to see the sweet peas in person "which would give you a clearer idea of what the laws [of heredity] mean more than

[9]Bearn 1993, 59–61: Garrod to Bateson, 1902.
[10]Bearn 1993, 42.
[11]Garrod 1902.
[12]Garrod 1908.

anything else."[13] But working on human heredity could be very perplexing. Nettleship noted some of the intrinsic problems with human studies:

> The difficulties in getting anything like complete pedigrees of several generations in a human family when one is inquiring about a heritable disease are very considerable [...] The facts in regard to a set of siblings are sometimes tolerably complete and sometimes in regard to parent or a grandparent but the collaterals or earlier generations can only be unearthed in very rare instances.[14]

Selective breeding, of course, was not an option as it was in plant or animal studies.

> In human clinical work one has to take what one can get, and hitherto it has been rather seldom that one finds a full record of the unaffected or of sexes or of order of birth or consanguinity.[15]

Bateson told Nettleship, "I have been spending a good deal of time collecting medical pedigrees lately,"[16] but the work was not progressing. He had examined some family histories of deaf mutism and concluded, "They are not analyzable." He examined data on Huntington's chorea. Transmission through an affected parent seemed "almost universal," but too many affected children were born. The ratio of normal to affected was "hopelessly wild."[17] Disorders of muscles were too irregular to fit any particular pattern of heredity except pseudohypertrophic muscular paralysis with a male-dominant system. Among families with Leber's disease (blindness), too many affected family members were also evident.[16]

The dentist J. G. Turner (1870–1955) sent him a family pedigree in which dental enamel hypoplasia appeared to be directly transmitted from parent to child over five successive generations. There were 11 normal and 21 affected offspring. Turner thought this might represent a Mendelian dominant character, but Bateson again could not see how Mendelism could explain the overabundance of affected offspring.[18, 19]

Bateson was forced to ask whether, in fact, human disease was hereditary, at least in the Mendelian sense.

> I am more and more inclined to think that the transmission of some of these at least depends on processes quite distinct from what one ordinarily finds in the heredity of variations.
>
> With the rarest exceptions all the characters experimentally investigated [in plants and animals] can fairly easily be shown to follow Mendelian rules, but after making widest allowance for errors and misstatements of all sorts, it seems to me most unlikely that these several diseases can be fitted [to the Mendelian model].[20]

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[13]P 329: Bateson to Nettleship, 1905.

[14]B 2663: Nettleship to Bateson, 1904.

[15]B 2664: Nettleship to Bateson, 1905.

[16]P 329: Bateson to Nettleship, 1907.

[17]P 329: Bateson to Nettleship, 1906a.

[18]B 2800: Turner to Bateson.

[19]Turner 1906.

[20]P 329: Bateson to Nettleship, 1906b.

Was disease familial because a "pathogenic organism" was passed from parent to child and then caused maladies in successive generations?[21] Bateson confessed that, "The more cases I go through, the more I doubt whether they are truly hereditary" at all.[21]

Nettleship promptly responded to that suggestion. He might know little about heredity, but he had learned a good deal about infections in his clinical years. He told Bateson: "About germs as a cause of hereditary [disease], I suppose one must not say more than that according to our present knowledge, the idea is almost inconceivable.[22] Specific diseases recurred precisely in subsequent generations: "transmitted for generations with exactly the same characters and localizations." He did not "think it would be fair to the poor over-burdened micro-organism to run him in for such as that...You must kindly find some other explanation".[22]

Nettleship attempted to elucidate such human cases by collecting pedigrees of many eye diseases: ectopia lentis, coloboma irides, aniridia, ptosis, color blindness, and retinitis pigmentosa. He investigated a large family in France with stationary night blindness and worked with local priests and physicians to reconstruct the condition in prior generations. The family tree was presented by Nettleship at a meeting of the Ophthalmological Society in 1907 and incorporated 2121 individuals over 10 generations. Healthy parents never produced affected children. The character was always directly transmitted from one parent to children. In affected branches of the family, there were 255 offspring, and 53% inherited this ocular condition.[22] Bateson accompanied his friend to the meeting and was not hesitant in his praise: "No doubt the paper must prove [to be] a classic." In his opinion, the pedigree in this case was most consistent "with a simple Mendelian dominant" character.[23, 24]

Two years later, Nettleship was asked to present the Bowman Lecture at the Ophthalmological Society. His topic was "On some hereditary diseases of the eye," and he used the Mendelian theory to analyze pedigrees from several different ocular disorders. Cataract appeared to be a dominant, as was glaucoma. Retinitis pigmentosa, of which he collected more than 1000 pedigrees, appeared to be of two types. Certain families demonstrated a dominant pattern, while others were more consistent with a recessive type. He emphasized the important role of heredity in causing diverse eye diseases.[25]

Archibald Garrod thought Mendelian theory might also help elucidate some of the rare neurologic diseases which also recurred in successive generations of certain families. He urged Bateson to accept an invitation to speak before the Neurological Society in February 1906.[26] In some respects, Bateson appeared to be an unlikely lecturer before such a clinical body. He told the audience that he wondered

[21]B 329: Bateson to Nettleship, 1906c.

[22]B 2674: Nettleship to Bateson, 1906b.

[23]Nettleship 1907.

[24]B 2689: Nettleship to Bateson, 1907.

[25]Nettleship 1909.

[26]Bearn 1993, 73.

"whether anything I could say would have a sufficiently direct bearing on the subject in which you are interested [...]." He titled his paper "Mendelian heredity and its application to Man," realizing that such application was "rather for the future than for the present."

He noted that the "application of Mendelian rules to mankind has not made the progress that was to have been expected [...]." But he was able to show pedigrees of several families with dominant traits (brachydactyly and cataract), recessive traits (alkaptonuria and perhaps albinism), and sex-limited characters (color blindness and hemophilia). He expressed his gratitude to Garrod and Nettleship for assisting him with such human material.[27, 28] Nettleship was also present at the meeting. He wrote to Bateson the following week that the "nerve men" had received a "not un-needed fillip," a little push to study human heredity and disease in their own patients.[29]

The London physician Alfred Gossage (1864–1948) sent Bateson several pedigrees showing the inheritance of tylosis palmaris et plantaris, curly hair, and heterochoma irides. Through multiple generations, the characters were transmitted directly from one parent to about half the children. Gossage designated these as dominant characters.[30]

The inheritance of exostoses (extra knots of cartilage on bones) was another object of his study. Typical families showed direct heredity for at least three successive generations.[31] He eventually collected 67 such families. The character was passed from parent to child, affecting about half the offspring. Males and females were equally involved. When he presented this material to the *Westminster Medical Society*, he could confidently label exostosis as a dominant trait.[32]

The two workers continued to collaborate on other human disorders. A disease of muscle spasms, myotonia congenita, was inherited in the same fashion and was a "clear dominant."[33] Malformation of the skull and the clavicle (cleidocranial dysostosis) also showed direct heredity in 17 pedigrees. Gossage suggested dominant inheritance as the most likely mechanism for its heredity.[34]

Zoologist George Mudge (1870–1939) was on the faculty of the London Hospital Medical School and investigated segregation of human characters involved in crosses of Canadian Native Americans and white Europeans. Eye color, hair texture, facial hair, and nose patterns were found in all combinations among children of such families. But in subsequent generations, latent features not evident in the first generation reappeared. Such segregation of hereditary characters convinced

[27]Bateson 1906a.

[28]Bateson 1906b.

[29]B 2668: Nettleship to Bateson, 1906a.

[30]B 2765: Gossage to Bateson 1910a.

[31]B 2766: Gossage to Bateson, 1910b.

[32]B 2765: Gossage to Bateson 1910a.

[33]B 2766: Gossage to Bateson, 1910b.

[34]B 2767: Gossage to Bateson, 1910c.

him that Mendelian laws functioned in humans as well as plants and animals.[35] He sent Bateson a circular in January 1908 announcing a lecture series "On Inheritance" to be held at his medical college.[36] To further publicize studies in heredity, Mudge and Bateson collaborated to establish *The Mendel Journal* that first appeared in October 1908.[37] Mudge presented an article in the first issue which summarized recent evidence that several human characters segregated in accordance with Mendel's laws: albinism, brachydactyly, night blindness, and asthma.[38]

The Scottish physician Harry Drinkwater (1855–1925) was another pedigree collector. He contacted Bateson in May 1907 after finding a "somewhat remarkable family" in Staffordshire with an inherited deformity (brachydactyly) with shortened digits of the hands and feet. About half of the children were affected, males and females equally. The pedigree was eventually expanded to seven generations.[39] Drinkwater presented his findings to the *Royal Society of Edinburgh* where the paper was "very well received" and generated discussion on the connection between such clinical findings and the Mendelian theory of heredity.[40] Drinkwater later spoke before the Liverpool Medico-Chirurgical Society in 1908 and designated the character brachydactyly as a dominant trait.[41]

Further studies on asthma continued the collaboration. Drinkwater sent Bateson a three-generation pedigree to review. Affected children were born only from an affected parent. About half the offspring suffered from asthma.[42] The expected ratio of normal to affected children was "in accord with Mendel's theoretical 50%."[43]

The public forum "Science Lectures for the People" had been a regular fixture of intellectual life in Manchester since the 1870s. Many well-known scientists of the era presented their findings to a layperson audience as a means of sharing the importance of science with the modern English public. Drinkwater presented "A Lecture on Mendelism" there early in 1910. He described how the mechanisms for heredity applied to various plants and animals. As a physician he was particularly interested to show that these same principles governed human heredity as well. He utilized examples of brachydactyly, night blindness, and hemophilia to support his claim that Mendel's work was indeed relevant for people.[44]

The last doctor in this presentation is Redcliffe N. Salaman (1874–1955), a general practitioner in Barley about 10 miles south of Cambridge. Since the spring of 1906, he had been studying segregation of traits in potatoes in his garden after

[35]Mudge 1907.
[36]B 1099: Mudge to Bateson, 1908.
[37]Kim 1994.
[38]Mudge 1909.
[39]B 2783: Drinkwater to Bateson, 1907a.
[40]B 2784: Drinkwater to Bateson, 1907b.
[41]Drinkwater 1908.
[42]B 2758: Drinkwater to Bateson, 1908.
[43]Drinkwater 1909, 88.
[44]Drinkwater, 1910.

Bateson had suggested this as a means to learn about heredity. He reported his work in the first volume of the new *Journal of Genetics*.[45] Coming from a Jewish background, Salaman also noted certain hereditary conditions almost exclusively within Hebrew populations. The absence of alcoholism and the frequent appearance of the neurologic disorder amaurotic family idiocy (Tay–Sachs disease) appeared to be the result of inbreeding among the Jews for more than 2000 years. A "homozygous combination of factors" would result from such consanguinity. The recessive inheritance of the characters then followed "in accordance with the laws of Mendel."[46]

1 Discussion

William Bateson's opinion on the relevance of Mendelism to human disease underwent a complex metamorphosis in this first decade of the twentieth century. His initial hint that the theory might explain the incidence of rare diseases in consanguineous families was eroded by the complexity of pedigrees for other more common disorders. By 1906, he was ready to admit defeat in trying to analyze human conditions. It all seemed so simple and straightforward in his plant and animal studies.

But the careful pedigree collection by his physician colleagues eventually convinced Bateson that Mendel's laws did indeed apply to all living creatures. By the end of the decade, he was able to state with confidence that Mendelism was indeed relevant to medical practice. The "Bateson boys" had spoken before many medical society meetings to present pedigrees illustrating the action of Mendelian dominant, recessive, and sex-limited inheritance. Bateson was now prepared to explain these issues to a wider medical audience.

A conference on "Heredity and Disease" was sponsored by the *Royal Society of Medicine* over four days beginning in November 1908. There were 18 speakers representing diverse interests and opinions on the relevance of heredity to the understanding of human disorders. The ophthalmologist N. Bishop Harman (1869–1945) commented that "the medical profession is profoundly interested in the question of the applicability of the laws of heredity as propounded by Mendel."[47] One observer noted, "The place was filled."[48]

Bateson was a keynote speaker and repeated the claims made by his medical colleagues that it could now be convincingly demonstrated that Mendelism applied to humans. He acknowledged the assistance provided by his medical friends in collecting well-researched pedigrees illustrating the segregation of human diseases.

[45] Salaman 1910/11a.

[46] Salaman 1910/11b.

[47] Harman 1908/09.

[48] B 673: Mudge to Bateson, 1908.

The list of human characters, which followed Mendelian patterns, continued to grow. Chorea, ectopia lentis, distichiasis, ptosis, and brachydactyly acted as dominant traits. Albinism and alkaptonuria were common in consanguineous families as expected of recessive disorders. Hemophilia, muscular dystrophy, and color blindness followed the unusual pattern of affected males with unaffected female carriers that fit the pattern for sex-limited segregation.[49]

George Mudge then presented families with other traits, which appeared to follow the Mendelian model. Hair color, eye color, and asthma acted as dominants, while albinism segregated as a recessive character.[50] Alfred Gossage discussed his pedigrees for two skin disorders. Epidermolysis bullosa and tylosis palmeris et plantaris families had equal numbers of affected and normal individuals. This was exactly what was predicted under the Mendelian segregation of a dominant trait. He concluded that:

> In order to explain these facts [of heredity], one necessarily required a theory, and the only theory which offered an explanation was that of Mendel [...]. Importantly that theory did not merely depend on human observations, but had in fact been validated by breeding studies on plants and animals where experimental investigations in the laboratory could be conducted.[51]

During this decade, Bateson's interpretation of Mendelian theory as it pertained to human heredity spread widely throughout the medical community. His personal relationships with many physicians allowed him to collect human pedigree data and then to show the physicians how Mendel's theory explained the segregation of diverse human traits. Bateson, his assistant Reginald Punnett (1875–1967), and the physicians who understood the relevance of Mendel's work to the daily practice of medicine discussed these findings before numerous medical society meetings throughout England.

Bateson benefited immensely from his collaboration with British medical men. In 1906, he was about ready to forget the notion that Mendelian heredity applied to man. But by 1909 when he prepared his book *Mendel's Principles of Heredity,* intensive work with physicians had convinced him that, in fact, many human characters followed the patterns of inheritance predicted by the theory.[52]

The application of Mendel's laws to humans appealed to many physicians because it was a logical extension of the rule-of-thumb observations they had been using every day in attempting to understand the relationship between heredity and disease. British physicians had utilized the pedigree format to summarize inheritance of human conditions since the 1880s.[53] About the same time, the French psychologist Theodule-Armand Ribot published his observations on patterns of

[49]Bateson 1908/09.
[50]Mudge 1908/09.
[51]Gossage 1908/09.
[52]Bateson 1909.
[53]Leslie 1881.

disease and heredity in English translation. He recognized three distinct patterns of human inheritance:

1. Direct—successive parent-to-child transmission of the trait;
2. Indirect—traits which ran in the family, often occurring in several siblings of one generation, often recurring in collateral relatives such as aunts, uncles, and cousins;
3. Sex-limited—a characteristic indirect pattern of affected males, and unaffected females transmitting the character to their sons.[54]

Clinical examples of such characters were well recognized by 1900. The advent of Mendelism explained how segregation of hereditary elements in egg and sperm could produce such recognizable patterns for the inheritance of specific human characters. The direct pattern was Mendelian dominant. The indirect pattern was Mendelian recessive. The sex-limited pattern was characteristic of the Mendelian sex-linked form of heredity.

By 1910, Bateson and his medical colleagues had successfully communicated in understandable terms the potential and important role for Mendelian heredity in providing workable solutions to the day-to-day clinical problems of human heredity.

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Part III
Genetics and Medicine

Pedigrees and Prejudices: Pre-WWII Inherited Disease Classification at the US Eugenics Record Office

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Abstract Pedigree charts became readily used during the era of classical genetics as symbolized recordings of inherited human diseases. They were particularly prominent tools for disseminating the work of the Eugenics Record Office (ERO), in Cold Spring Harbor, Long Island, New York. ERO-trained fieldworkers organized data on pedigree charts that became national inventories of various diseases including tuberculosis, syphilis, and alcoholism.

This chapter explores ERO superintendent Harry H. Laughlin's use of pedigree charts to maneuver the flow of information of inherited disease to medical and public audiences. An advocate of the pedagogical power of visual displays since his days as a college professor, Laughlin's ERO work further propagated the use of pedigree charts to visualize the invisible. On one level, the charts served as scientific tools to display spatial arrangements of hereditary patterns of disease. On another level, they were rhetorically used to persuade society that eugenics operated within mainstream science of the era.

This chapter analyzes the relationship between inheritance and disease as represented in the pedigree charts that the ERO prepared and distributed during Laughlin's superintendence. The explicit and implicit uses of various formats of ERO pedigrees are examined, including their appearance at the International Eugenics Congresses of 1912, 1921, and 1932, at the Chicago World's Fair of 1933–1934, in many county and state fairs across the USA, in routine ERO mailings across the country and to Europe, in the correspondence between physicians seeking to update classifications of inherited human disease, and in popular biology textbooks and marriage manuals.

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Within the burgeoning field of eugenics history, pedigree charts have received minimal attention. This remains somewhat puzzling if one accepts the claim that "almost all studies of human heredity" in the early 1900s "tended to involve the collection of pedigrees." It was, after all, these studies in particular that provided the "facts of human inheritance necessary for the construction of eugenic breeding programs."[1] This lack of attention may, in part, be due to the longstanding marginalization of the study of images in favor of text, at least within the history of science and medicine.[2]

Pedigree charts were hardly a new concept of representing information during America's Progressive Era. Indeed, they had been used for centuries across the globe in attempt to trace human lineages back to the Biblical Adam. The term "pedigree," or etymologically *pied de grue* (a crane's foot), derives from the symbol used in medieval genealogical tables or trees that, looking like the multi-pronged avian's foot, denoted a succession of generations.[3]

The medical use of pedigree charts in the USA was pioneered in 1845 by Philadelphia physician Pliny Earle as he visually documented five generations of one family's history of color blindness.[4] Yet, this representation of heredity from a medical viewpoint was little copied throughout the nineteenth century. But as the medical science community clamored over the US government devoting considerably more resources to the proliferation of its agriculture and farm animals than it did its own human population, the pedigree chart reemerged in the study of humans during the "classical era" of genetics.[5] Slowly throughout this period, the pedigree

[1]Ludmerer 1972, 55. This chapter is culled primarily from a presentation delivered at "The Biological Future of Man" conference (Nuremberg, 2012), with bits added from work shared at "A Cultural History of Heredity IV: Heredity in the Century of the Gene" invited workshop (Exeter, 2006) and at the "International Symposium on the Topology of the Body" (Nagoya, 2008). The author extends his deep and enduring gratitude to conferees at each of these events as well as the staff of Special Collections, Pickler Memorial Library, Truman State University, Kirksville, Missouri, USA, and of the American Philosophical Society Library, Philadelphia, Pennsylvania, USA.

[2]Sander Gilman 1988 and Stafford 1991 have long noted this point. For an excellent overview of the importance of carefully chosen images, see Tufte 2001. One notable exception is Mark Jackson 1995, who has focused upon the visual representation of feeblemindedness in early twentieth-century eugenic literature. For a review of the "mapping" mentality of geneticists, see Gaudillière and Rheinberger 2004.

[3]For an interesting etymological ramble through pedigrees and the nomenclature of nature, see Potter and Sargent 1974. For a more thorough review of the use of pedigree charts in human medical practice, see Resta 1993, Rushton 1994, Nukago and Cambrosio 1997.

[4]Rushton 1994, 12–14.

[5]As an example of the apparent lack of attention on humans, Downing 1918: 149 argued that the "expert dairyman carefully inquires into the purity of strain and ancestral performance of the animal he mates with his choice cows. The farmer insists on a hog with certified ancestors. We have sense enough to apply such knowledge of heredity as we possess to our farm stock. It seems

chart became a standardized scientific tool to medical audiences, using simple, readily recognizable symbols to denote particular meaning regarding heredity and disease. In due course, this tool eased communication about the developing understanding of hereditary patterns of human disease, bridging classical genetics from the theoretical, to the experiential, to the clinical.

In the USA, the greatest popularization of the pedigree chart as a tool to visualize inherited human characteristics or traits emanated from the work of the Eugenics Record Office (ERO), in Cold Spring Harbor, Long Island, New York. ERO-trained fieldworkers organized data gathered from throughout the USA on charts—what they termed "Mendelian Blanks," a phrase that attests the bias of their outlook—to represent the incidence and prevalence of particular traits or characteristics that were thought to be hereditarily passed along familial lines.[6] The leading US publication of popular science, *Scientific American*, claimed that the ERO pedigree charts represented a true "inventory of the blood" of the nation.[7]

The concept of a Progressive Era in US history (approximately 1890–1920) invokes myriad views. One such view represents the time when the USA strengthened its position in relation to other leading nations worldwide. Doing so required a double-faced, Janus-type look into both its past and its future. As a nation just over a century old, the USA had expended considerable effort, first in fighting to maintain its independence and, more recently, to hold itself intact as a nation. Over that century, the USA had also accumulated an expanding genealogical record.

Within some circles, it was thought that the nation's strength and endurance was closely correlated with the physical constitution of its people. The New England physician, Edwin M. Fuller, argued that the relatively young USA still had a chance to fend off becoming laden with hereditary disease.

> The older a nation grows, the larger the percentage of hereditary diseases are manifest, and . . . after a century's growth, our nation appeals in silent language to our profession for remedies and intelligent barriers which may be stationed at the portals of society, that the ignorant and easily captivated masses may be warned of the approaching dangers to society and individuals.[8]

A major shift in thoughts about heredity and disease followed the rediscovery of Gregor Mendel's work during this era. While working at the newly opened

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little enough to ask that we should exercise as much good sense in producing children as we do in the production of hogs and corn." Such claims were still pouring forth a decade later. M.R. Ferris 1929, secretary to the Council of the Institute of American Genealogy, the National Adoption Information Clearinghouse, wrote to Laughlin with the sentiment, "Certainly you will agree that the systematic preservation of the lineages of human beings in the interest of better citizenship is infinitely more important than the registration of livestock pedigrees in the interest of better beef." Kimmelman 1983 analyzed the agricultural context within which human eugenics arose.

[6]The Mendelian leaning of the ERO has been widely noted. See, for example, Rushton 1994 and Turney and Balmer 2000.

[7]Collins 1913.

[8]Fuller 1887, 206.

University of Chicago, Harvard-trained zoologist, Charles B. Davenport, summarized Mendel's findings for an English-reading audience.[9] Within a few years, Mendel's principles of genetics were being applied not only to plants and animals but to humans as well. Davenport became director of the Carnegie-funded Station for Experimental Evolution in Cold Spring Harbor in 1904. Six years later, he established a division of this station, the Eugenic Record Office (ERO), that focused solely upon eugenics. For the next 30 years, America's most significant advances in promoting eugenics stemmed from this office.[10]

In 1910, Davenport hired Harry Laughlin to supervise work at the ERO, and together they organized that office around specific goals of operation, several of which specifically involved the production, storage, and analysis of pedigree charts.[11] Above all else, Laughlin repeatedly distinguished the need for ERO pedigree charts to delve beyond those typically used by genealogists. The genealogist, he argued, "strives to work out the family net-work, giving . . . names, dates, and connections." What was missing, however, was "a description of the natural, physical, mental, and temperamental qualities of each member listed." Once this information is provided, Laughlin concluded, we will have a "record of practical pedigree-value, one which can be used in tracing the descent and re-combination of natural qualities within the family-tree."[12] Laughlin summarized, the "usual outline of the genealogist . . . is merely the skeleton" upon which ERO efforts must "clothe it with the sinews and organs of natural traits" if pedigree charts are to "have any scientific value."[13] Yet he envisioned this mere charting of biological information as only a beginning. "Individual Analysis Cards," listing all of each pedigree members' constitutional traits, tendencies, and disorders, were also required to complete the "critical biological biography" for each family. For "when displayed in this manner," the "bare facts concerning the natural capacities and shortcomings of various members of a family . . . constitute an instructive guide for the family."[14]

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[9]Davenport 1901. For a biographical overview of Davenport and his contributions to hereditary thinking, see MacDowell 1946 and Kevles 1985.

[10]For extensive historical accounts of the ERO, see Allen 1986 and Watson 1991.

[11]To review of Laughlin's contributions to eugenics, see Bruinius 2006, Hassencahl 1970, Reilly 1991, and Wilson 2002, 2003, 2006, and 2013.

[12]Anonymous 1920, 77.

[13]"A Few Points to Observe in Writing up Notes." Elsewhere (Harry Laughlin Papers, 1939a, p.15), Laughlin acknowledges that the genealogists' biographical accounts were of some help to eugenics research as "records of human functioning which check[ed] constitutional traits diagnosed or collected from other sources." See also "Eugenics and Other Sciences," 1920 and Davenport et al. 1911.

[14]"Brief Instructions on How to Make a Eugenical Study of a Family," Harry Laughlin Papers, 1915, sections II and II. Banker 1923, p. 306, suggested the word "ecography" to account for the complete biological and historical component of family histories. The ERO was not alone in providing instructions of the construction of human pedigree charts. J.F. Munson 1910, a physician working at the Craig Colony for Epileptics in Sonyea, New York, published easy-to-follow guidelines in the *New York Medical Journal*.

When constructed with critical care, pedigree charts and the accompanying analysis cards—collectively referred to by the ERO as "scientific genealogies"—would be able to serve multiple purposes. On one hand, they provided essential information for every individual to "inquire into the natural endowment of its . . . members and by pedigree study to find out how the traits of each would be transmitted in given matings—to be calculating and forehanded in mate selection, so that the offspring will present fortunate combinations of desirable family traits." To this end, Laughlin argued, "every family should establish a permanent Family Pedigree Archive, for only through the information conveyed by such may the facts of fortune be worked out—or, to put it in the old way, may one see where the finger of destiny points." Indeed, it would serve a greater value still if "several branches of one's own family" could have their investigations "coordinated by a family association" whereby a "most excellent and useful scientific pedigree record" of the whole family would be produced, a task requiring "but little effort on the part of each branch." [15] Such a study, he noted, becomes "almost priceless" to a given family, particularly after the "oldest person consulted in preparing it has passed away." For, "as a rule, an individual is personally acquainted with but three generations of his or her kin and connections, and without personal knowledge and care [,] character analysis is very difficult."[16] Indeed, it "should be considered a filial duty as well as a duty to society to secure at the earliest opportunity from the oldest living members of one's family detailed facts concerning those who still live in the memory of their contemporaries."[17] It will be "a happy day for our national welfare," he championed, "when the keeping of . . . [a family pedigree] archive becomes a national family habit." Each family merely "needs but an organizer" to accomplish this goal. [18]

Ever the organizer himself, Laughlin envisioned his own pedigree archiving task on a much grander scale. Similar to what he urged each family to acquire, Laughlin sought for the ERO to become the national pedigree archive. By acquiring "all authentic family history studies," the ERO "seeks ultimately to have an index of the network of the family kin and of the natural heritable traits of all of our better American families." As this "ideal[ized goal] becomes realized, it will become less difficult," he concluded, for "representative families by using the [ERO's] files . . . to work out" their futures in "practical pedigree—i.e., trait prediction—fashion."[19]

To achieve this national aim, Laughlin coordinated the collecting and recording of family data through an extensive outreach program. From 1910 through 1924, he and Davenport trained teams of "fieldworkers" (primarily young college-educated women) in the principles of human genetics and provided them with skills neces-

[15]"The Permanent Family Pedigree Archive," Harry Laughlin Papers.

[16]"Brief Instructions on How to Make a Eugenical Family Study," Harry Laughlin Papers.

[17]Davenport and Laughlin 1915, 3.

[18]"The Permanent Family Pedigree Archive," Harry Laughlin Papers.

[19]"Eugenics," p. 6, Harry Laughlin Papers.

sary to gather extensive family histories.[20] Laughlin exposed fieldworkers to a series of lectures and lab activities on eugenics. The range of topics he addressed included chromosomal structure, anthropometrical measurement, elementary statistics, and discussions of the medical conditions deemed to be, at least in part, hereditary such as skin pigmentation, insanity, cataracts, and epilepsy. Additionally, he led these students through an experimental study of cross-fertilized and purebred corn in order to allow them to personally uncover the Mendelian laws regarding the segregation and recombination of hereditary traits. In subsequent discussions, students used visible evidence obtained from their corn experiments as analogies for the transfer of "defective" traits and "unfit" matings in the human population. Students were also provided with ERO-established guidelines instructing them how to make a eugenic family study. To gain experience in charting family pedigrees of actual "social defectives," students were sent on supervised educational visits to study the patient populations in nearby clinics at King's Ridge, Amityville, Letchworth Village, and Central Islip. They also visited the immigration control facilities on Ellis Island.[21]

In contrast to Laughlin's encouragement of America's best families to submit their own pedigrees to the ERO, he focused ERO fieldworkers' efforts to document the pedigrees of those he deemed as "socially defective" or "socially inadequate." Fear was already looming over the increasing numbers of "degenerates" in the USA before the Great War. State legislators deemed such individuals as the "greatest problem that confronts our nation," and they claimed the "degenerates" were present in "a greater multitude" than anyone could count.[22] Supportive of their concern, Laughlin and his fieldworkers provided the essential ingredient that legislators had been missing: specific quantification of the "social deviants" who, it was argued, by their "inferior blood" were viewed as a great and costly "menace to society."

Although the ERO acknowledged that information about family histories has "for many years" been obtained through the admissions material, medical examinations, and letters from relatives regarding "defectives" in "the better organized hospitals and institutions," such information was "far from satisfactory." The ERO claimed that "experience had shown that there is only one way to get a satisfactory family history of a stranger and that is to go, or to secure a trained assistant to go, to the various members of the family and with tact and patience and time secure the

[20]For a telling account, see Bix 1997. Laughlin 1929 claimed to have overseen the training of 258 fieldworkers between 1910 and 1924.

[21]Laughlin, "A Corn Breeding Experiment," Harry Laughlin Papers. Henry H. Goddard (1910), noted eugenicist and superintendent of the care of the institutionalized feebleminded in Vineland, New Jersey, also supplied specific instructions for fieldworkers in the preparation of pedigree charts.

[22]*Report of the Commission on the Segregation, Care and Treatment of Feeble-Minded and Epileptic Persons in the Commonwealth of Pennsylvania*, Legislation Pursuant to Joint Resolution, 14 June 1911.

necessary facts."[23] Using fieldworkers to "go to the homes" and to "interview persons that can and will give the desired information" would reputedly enhance the precision and accuracy of the data obtained. First, however, such workers would glean all they can about a patient from the institution's office files. Although they were encouraged to focus upon the specific trait being studied, fieldworkers were also urged to embrace further opportunities to "learn of other traits that may be significantly or incidentally associated with the primary trait."[24] "Just before starting out to visit the relatives and friends," the fieldworker was instructed to visit the patent "in his ward or cottage." Then, "armed with recent personal knowledge of the patient, which assures her cordial welcome," the fieldworker proceeds to visit the patient's home and "interviews the relatives, friends, and family physician." The fieldworker was encouraged to "see as many relatives as possible," since "facts omitted or overlooked by one [relative] were often recalled and told in full detail by another." Once the data was collected and recorded, a pedigree chart would be constructed.[25] Fieldworkers were sent out with the assurance that "the parents or other relatives of the patient" will be "pleased to think that the hospital or school takes such an interest in the patient as to send a visitor to the home."[26]

As many members of the "restricted" and "extended" families as possible were to be recorded on the pedigree chart.[27] Fieldworkers were urged to "lay great stress upon the reliability of the sources" of the information that they obtained, but to also check the "testimony of one informant against another." The traits and personalities of those individuals in the collateral lines (i.e., any line other than a direct ancestor) of the pedigree were to be strongly considered since a better understanding of their genotype would "throw light upon the germ plasm of the propositus." Fieldworkers were warned "Don't diagnose!"—and to use terms including 'insane,' 'feeble-minded,' 'criminal,' 'neurotic,' and 'normal' with great caution. Rather, they were instructed to provide sufficient details to "enable an expert to draw some conclusions from the data."[28] Standard symbols were to be used to represent afflicted individuals, specific lines of generational lineage, and specific traits and afflictions. The ERO produced a *Trait Book* to ensure that standard symbolic

[23]Davenport 1915, 18.

[24]Davenport et al. 1911, 7.

[25]Ibid., 1–2.

[26]Davenport 1915, 18.

[27]The "restricted" family consisted of the propositus, his siblings, and the consorts and children of these siblings; the father of the propositus and the father's siblings and consorts and their children; the father's father and the father's mother as well as the corresponding relations on the mother's side of the family. The "extended" family included, in addition to the restricted family, a history of the uncles and aunts by marriage, the consorts and children of the cousins, the siblings of the grandparents and their consorts and children, as well as their children's children and of the eight great grandparents. Davenport 1915, 6.

[28]"A Few Points to Observe in Writing Up Notes," Harry Laughlin Papers.

representations were known.[29] Some disease or "defective" conditions were so frequently studied that they acquired specific color representations on pedigree charts. For example, red was used to encode for epilepsy, green for insanity, violet for criminalistic tendencies, and black for feeblemindedness.[30] Finally, fieldworkers were alerted to provide the names and address of "defectives who need institutional care." As such, the data that they collected became particularly "useful information . . . when application is made for admission" to respective institutions.[31]

The ERO relied upon the pedigree chart as its most common tool of assimilating and promulgating information about the nation's reproductive stock. Such charts served practical measures by "determin[ing] . . . the eugenical fitness" of a contemplated marriage, "gauging the specific educability or the hereditary potentialities of a given individual," and determining the "intrinsic value of . . . [a] family, whenever such knowledge may aid . . . [that] family in directing . . . the education of its youth and in encouraging biologically fortunate matings of its marriageable members."[32] Originally, a tool for genealogists and biographers, this chart was modified by fieldworkers and others at the ERO so that it could just as easily be used to express biological aspects of all the individuals within a given family. By incorporating all of the known and gathered data about a particular family on one sheet of paper, these charts maintained a visual simplicity.

Overall, pedigree charts objectified, quantified, and visualized many previously invisible aspects of disease. They penetrated into the germ layer giving new insight into the genotypic level regardless of whether any aspect of the disease was phenotypically expressed.[33] In that way, they allowed for better discrimination of hereditary difference between individuals. But as ERO efforts demonstrated, they also provided a new way of imaging or representing disease.[34] As such, they became a conceptual tool for more fully appreciating patterns of inheritance for particular diseases. They also revealed a new structural knowledge that gave a better glimpse of the movement of disease via the germ plasm throughout a given family.

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[29]Among the disease traits or characteristics listed were alcoholic, blindness, Bright's disease, cancer, chorea, cripple, criminalistic, deafness, dementia, dropsy, eccentricity, encephalitis, epileptic, goiter, general paralysis of the insane, gonorrheal, hysteria, ill-defined organic disease, insane, kidney disease, locomotor ataxia, manic depressive insanity, migrainous, neuropathic condition, obesity, paralytic, paranoia, pneumonia, senile, sexually immoral, shiftlessness, softening of the brain, syphilitic, traumatic insanity, tubercular, vagrant, varicose veins, and vertigo.

[30]Davenport et al. 1911, 4.

[31]Ibid. 2.

[32]"Brief Instructions on How to Make a Eugenical Study of a Family" 1915, Harry Laughlin Papers.

[33]For a contemporary discussion of genotype, see Johannsen 1911. Sapp 1983 further contextualizes the genotype-phenotype distinction as iterated during this period. The word "idiotype" was used somewhat synonymously with "genotype" in literature of the period, particularly in that of the constitutionalists' writings on the body.

[34]For an overview of the social construction of genetic disease, see Yoxen 1984 in contrast to Child's 1999 history of ideas approach.

In and of themselves, these tools exhibited connections and offered some cautions as to what to look for in existing and future generations. On their own, however, they did not offer infallible explanations of particular patterns of inheritance. Many different humans had gathered information for pedigree charts, and thus, this process left considerable sources of error. Perhaps an even greater source of error arose from the potential of missing information in one or more generations. Quite often, fieldworkers and others relied solely upon the subjective views of one family member to account for various states of disease in all of that individual's known relatives. Even if that individual divulged all that he or she knew, much of this view may have stemmed from hearsay. Others, it was noted, may have been cajoled by fieldworkers into giving information that they thought the fieldworkers wanted to hear. Finally, no system was in place to verify either the information that was collected or its recording. Or, in other words, as critics claimed, they were "insufficiently critical to establish what actually is true."[35]

Within the world of medicine, pedigree charts became shorthand representations of the presence and potential patterns of disease. As with any shorthand system of symbolization, minimalist abstractions were rendered. In this case, humans were disembodied into some type of representational simulacrum in which they appeared as only bits or bytes of select information. This idea advanced reductionistic representations of humanity by offering a tool that diminished the concept of the human. The disembodiment of humans to mere boxes and circles encoded with information was consistent with reductionist thinking common of that era which began looking at the body more as distinct components rather than as a whole patient.

Within a short timeframe during the "classical era of genetics," pedigree charts gained an iconic status.[36] Though mere lines, circles, and squares, they held a power to persuade viewers to think about heredity within their own family. Curiously, these little mini-exhibitions of knowledge served both individual and societal needs. On one family's pedigree chart, each individual was highlighted as was his or her interconnectedness with everyone in an entire family, at least regarding a particular trait or disease. These charts seemed to introduce labels of either normality or deviance upon potentially all members of the family represented therein. But the ERO also used vast collections of pedigree charts as a form of collective data, expanding their apparent range of observation, in a manner that supported their overarching efforts of societal reform. Such efforts were aimed, in part, to convince American society that eugenics was working well within mainstream science of the era. Pedigree charts were, so Laughlin argued, an "obvious" choice to unambiguously document and visualize the "practical application" of eugenics schemes.[37]

[35]Ludmerer 1972, 59.

[36]For coverage of other icons related to heredity, see Nelkin and Lindee 1995 and Rheinberger and Gaudillière 2004.

[37]Laughlin 1912, 121.

As the ERO was actively involved in educating the public, Laughlin worked diligently to keep the message of eugenics paraded before the populace. As a public servant, he oversaw the design of a multitude of easy-to-understand pedigree handouts which, using simplistic diagrams and brief accompanying text, were used to relay particulars about the genetic principles underlying human eugenics for the lay public. He distributed these handouts freely to thousands of individuals who contacted the ERO. As part of his routine, Laughlin would ask people to complete two family pedigree charts which he included in his mailings. He urged the recipient to be as accurate and complete as possible in identifying all the hereditary traits in each family member according to the list he enclosed. If desired, the ERO was "glad to supply . . . small rubber stamps" of squares and circles, free of charge, to ease the completion of the charts. He urged people to "recast" the chart "two or three times" before drawing up a final copy, to incorporate all "new kinsmen . . . discovered" in the process.[38] After completing both forms identically, the recipients were to return one of them to the ERO for "secure filing" where it would remain "permanently available for reference by persons with legitimate concern" for such records.[39] The other, he suggested, should be kept for their own family records. His actions were aimed at providing families with a tool that expanded the genealogical tree recorded in family bibles, helping them to better visualize the genetic traits present in their family's recent past. This task also fulfilled Laughlin's self-serving interest of supplying data to the ERO beyond that generated by the fieldworkers.

Similar letters were sent to community clubs and organizations as well as to libraries. He closed his form letter to libraries acknowledging their help in "aiding pedigree study of the human family" by "securing valuable permanent records which otherwise would not be prepared, or if prepared, would be lost to the family and the state."[40]

By the late 1920s, nearly 400 US college courses were taught on eugenics.[41] Laughlin directed a series of letters to professors of biology, sociology, and psychology urging them to adopt his pedigree charting methods. Professors were asked to supervise student's completion of the ERO's standardized pedigree forms, and Laughlin left it up to the professors to collect the forms to return to the ERO or to "eliminate" any of the pedigree charts that were, according to the professor, "inaccurate or scantily prepared."[42] Professor U.G. Weatherly of the Indiana University claimed that this project "furnished the very best possible kind of laboratory material." There "could be no more effective method of getting young

[38]Davenport 1915, 9.

[39]"Family-Tree Folder," p. 1, Harry Laughlin Papers. For a discussion of the confusion over various attempts in analyzing these pedigrees, see Laughlin's "Report on Researches in Eugenics and Heredity" 1939.

[40]Laughlin, "Letter to Libraries," p. 2, Harry Laughlin Papers.

[41]Allen 1983, 116.

[42]Laughlin, "Memorandum of Suggestions to Instructors," Harry Laughlin Papers.

people in contact with the serious problems of family inheritance," he added. Students are "led not only to take a vital interest in the family history," but this pedigree analysis gave them "a sound and impelling interest in the future fate of their own groups and of the race."[43]

In what was undoubtedly his single greatest success in educating the masses, Laughlin organized a eugenics exhibit around the theme "Pedigree-study in Man" as part of the 1933 Chicago World's Fair. Consistent with the Fair's "Century of Progress" theme, Laughlin incorporated many recent eugenic advances within his exhibition.[44] He created a series of panels which, when viewed according to a specific order, presented the principles of human heredity as a puzzle which exhibit goers could solve based upon their own personal and family experience. Since "no one was stationed permanently at the eugenics exhibit," it was "necessary that the charts be self-explanatory and well adapted for conversation among mutually interested visitors."[45] To ensure that his exhibit caught the attention of every age and social class, he employed a variety of practical laboratory setups. Some stations were set up with the Midwestern American farmer in mind, invoking parallels between human stock and livestock breeding and crop production. The socially elite were catered to with a "test for instinctive appreciation of quality and elegance." In this test, ten animal fur samples of varying quality were placed on a table. Using score cards, fair goers were asked to "consider quality and elegance in relation to the appeal [that the furs made] to you personally" and then to rank the samples from best liked to least liked. People's own findings could then be applied to corresponding pedigree charts that outlined how certain favorable traits in a human population could best be propagated.[46]

Part of the effort to improve general eugenic knowledge was aimed at approaching marriage in a more discriminating manner. If young people, "before picking out their life partners, are taught to realize the fact that one marries not an individual but a family," then "better matings will be made."[47]

ERO efforts had long warned that unfit marriages would bring about distasteful and unproductive offspring. Laughlin's exhibit at the Chicago World's Fair incorporated pedigree charts showing how both desirable and undesirable traits could be passed along family lines. By placing two pedigrees side by side, he drew particular contrasts between the presidential Roosevelt family and the "degenerate" Ishmael family. Similar to the Jukes and the Kallikaks, the Ishmaels from Indiana were used as a representative family of over 1750 individuals in which eugenicists traced the

[43]"Anonymous, Family Pedigree Study as College Laboratory Work," 1927, 84.

[44]The "Century of Progress" theme was selected in attempt to "demonstrate to an international audience the nature and significance of scientific discoveries and the methods of achieving them." Chicago Historical Society: History Files—A Century of Progress, 1998.

[45]Laughlin 1935, 161.

[46]Laughlin 1932.

[47]Popenoe and Johnson 1922, 164.

linear passage of "defective germ plasm."[48] By studying the passage of ancestral lineage, viewers were urged to drop any lingering views that marriage was purely a human choice and adopt the more socially desirable belief, at least according to the eugenicists, that responsible Americans pursued marriage mindful of eugenics.[49]

The pedigree chart proved to be a valuable tool for the developing field of human genetics in several important ways. It offered a concise and clear way of demonstrating a perceived hereditary linkage regarding a particular disorder or disease. Laughlin's coordinated gathering and distribution of family pedigree information was designed, in part, for the eugenic attempt to maintain a healthy reproductive stock within the US population. As such, his use of these charts further substantiated the "hardening" that had occurred in beliefs about the nature of heredity during the late nineteenth century. In particular, the regular appearance of these tools strengthened "hard hereditarian" claims that inherited defects and disease were solely dependent upon a nonmalleable nature.[50] The heavy reliance upon these charts strongly suggests that the alteration of the reproductive stock of the American people during the Progressive Era became intensely focused upon nature rather than nurture. Further investigations remain to be undertaken in order to more fully appreciate the roles whereby pedigree charts secured such an iconic permanence in the field of human genetics.

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[49]For Laughlin's own account of the success of this exhibit, see Laughlin 1935. For a comparable assessment of the eugenics exhibition at the Second International Congress of Eugenics in 1921, see Laughlin 1923.

[50]Carlos López-Beltrán 1994 described that this malleable view existed in the "soft hereditarianism" beliefs of the early nineteenth century in contrast to the more objective qualifications of a nature-based, "hard hereditarianism" later in the century.

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Aldred Scott Warthin's Family 'G': The American Plot Against Cancer and Heredity (1895–1940)

Toine Pieters

Abstract According to many, the genetic technology used in cancer is a promising test case of twenty-first century 'genomic medicine'. However, it is important to realize that accounting for the genetic or hereditary factors in cancer medicine is not new. Since at least the eighteenth century, medical doctors and patients have tried to establish links between heredity and cancer. Following the excitement over the rediscovery of Gregor Mendel's theory of hereditary transmission (1900), there was renewed interest in the question of a linkage between heredity and cancer. Researchers began to pay attention to the statistical use of family studies as a means to calculate Mendelian ratios of disease inheritance. In 1913, the Michigan University pathologist Aldred Scott Warthin (1866–1931) published his first study of a pedigree with a so-called inherited susceptibility for cancer. Family G's susceptibility was associated with the risk of creating an 'inferior stock'. Given the number of studies on heredity and disease and the vogue for eugenics at the beginning of the twentieth century, one would have expected strong support for Warthin's study. Family G (one of the longest systematically studied cancer genealogies in the world and currently associated with Lynch syndrome) might have been accepted (if not for purely scientific reasons) as part of the eugenics gospel as an exemplary case of a degenerative stock. After all, Warthin was a rising star within the American medical establishment and had become part of John Kellogg's eugenic priesthood in Michigan. Ultimately, none of these likely

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scenarios materialized. I will show in this chapter how the cancer idiom of heredity that was associated with shame, fatalism and stigmatization came to be regarded as counterproductive in the fight against cancer and was suppressed at the time by the powerful American Society for the Control of Cancer.

Keywords Lynch syndrome • Family G • Eugenics • Colorectal cancer • Genetic condition • Family history

1 Introduction

Many consider cancer genetics to be the most promising test case of genomic medicine of the twenty-first century. Cancer, which is now accepted as a family of genetic traits and diseases, is inextricably bound to the discovery in the 1990s of specific genes collectively known as 'mismatch repair genes'. Although this understanding of cancer was innovative in terms of the science and technology involved in cancer medicine, it is important to realize that accounting for genetic and hereditary factors is nothing new in and of itself. It has long been known that cancer in the human species may run in families. Since at least the eighteenth century, medical doctors and patients have tried to establish links between heredity and cancer. As for other medical conditions, heredity's visibility, meaning and legitimacy have fluctuated over time. The same holds true for the role of a family's history in medical research and medical practice.

Collecting and understanding family histories has been part of medicine since the early nineteenth century. However, it was not until the 1850s that medical researchers developed an interest in family trees as a means to study and visualize the influence of heredity on cancer. The use of genealogical methods by medical researchers interested in the hereditary transmission of cancer is best exemplified by Paul Broca's (1824–1880) much-cited history of the so-called 'cancer family' of Madame Z[1]. After publication of Broca's pioneering study, international discussion about medical family studies and cancer continued as part of an ongoing debate on the question: 'Is cancer a hereditary disease?' At that time, there was no consensus concerning the nature and the magnitude of the hereditary factor in cancer. The American pathologist Aldred Scott Warthin (1866–1931) and his pedigree of Family 'G' were very much part of this debate from the days of Weismannism (1890s) to the age of brave new biology (1930s).

In her book *Moments of Truth in Genetic Medicine*, Lindee has provided an intriguing window on the amount of labour involved in the construction and maintenance of scientifically legitimate human pedigrees.[2] Pedigrees as a token of family identity blend folk, emotional, social and technical knowledge. From the nineteenth century to the present time, pedigrees as an integral part of medical family research have had multiple roles in framing illness, disease and social

[1]Lynch 1985, 12–13; Carlson 2001, 147.

[2]Lindee 2005.

abilities. The father of eugenics Francis Galton's (1822–1911) early use of pedigrees was exemplary in his study of the inheritance of genius and artistic ability.[3] To further our understanding of how the use of pedigrees as a tool in medical research has changed over time and within specific contexts, studies are needed that focus on the multidimensional historical trajectories of family studies. Thus, in this chapter on the genesis of a specific American cancer pedigree, I focus on Family G. This family was one of the longest systematically studied cancer genealogies in the world and is currently associated with the occurrence of hereditary non-polyposis colorectal cancer or Lynch syndrome. I will show how science, medicine and the public sphere have shaped and reshaped the identities of Family G and their pedigree as an object and tool of medical research from the 1890s to the 1930s in the American context.

2 The Birth of a Medical Pedigree: Family G

From 1893 to 1900, the young American pathologist Aldred Scott Warthin spent his time in pathology laboratories in Vienna, Austria, and Dresden and Freiburg, Germany. Warthin, who had a strong interest in the biological sciences, must have taken notice of the various scientific and popular discussions about the hereditary transmission of diseases or mental qualities during his study trips. It is likely that Warthin also took the opportunity in Austria and Germany to study the expanding literature on the biological and medical aspects of family research. If so, he must have noticed that the results from medical family research were as diverse as the methods of compilation since they were based on family histories, hospital records and replies to enquiries.[4] Most doctors at the time treated hereditary aspects as part of a nosographical description, whatever their views on the magnitude of the hereditary factor and the mechanism of transition. They usually spoke in terms of a potentiality and disposition to disease as part of a constitutional diathesis. In general, the term 'heredity' stood for a tendency for certain maladies to develop within a family.[5] Only the predisposition to develop the disease was inherited, not the disease characteristics. Expression depended on circumstances, for example, shock, misery or strain. The perspective of plasticity of expression was compatible with existing medical traditions and biological theories. Furthermore, the more often the disease characteristics occurred in pedigrees, the greater the chance that they would return in later generations.[6]

In 1895, upon his return to the University of Michigan, Warthin was appointed a demonstrator in pathology. Barely a year later, he assumed charge of the pathology

[3] Kevles 1985; Paul 1998.

[4] Gausemeier 2005.

[5] Snow 1893, 15; Butlin 1887; Butlin 1895; Jacobsen 1946, 13–17; Krush 1977.

[6] Snelders et al. 2007, 226.

laboratory where he worked at the university hospital in Ann Arbor.[7] Warthin loved to take a roundabout route home from work through Ann Arbor's German quarters. The familiar laborious German atmosphere and the chance of practising the German language made him feel comfortable. During one of these rounds, he ran into his family's young seamstress Pauline G, who looked unusually depressed. He questioned Pauline about her grief and learned that cancer was rampant in her family. Quite a number of her relatives seemed to have cancer, have died of cancer or were about to die of cancer. Pauline felt vulnerable and was afraid that she too would get cancer. Unfortunately, history would prove her fears to be correct. Like her mother, Pauline fell victim to a rapidly developing cancer of the uterus.

Although Pauline had initially only provided meagre details about her family history, her narrative corroborated Warthin's ideas about a family susceptibility or hereditary disposition to disease. Warthin thought that his own family was a cancer resistant and Pauline's was cancer susceptible. He had always been surprised about the nature of family histories and the so-called cancer statistics that were used in discussions about familial cancers. Rarely were clinical examinations supported by microscopic examinations, and only occasionally was an entire family history obtained extending over several generations. Moreover, most statistics provided little information beyond the fact of the multiple occurrence of cancer in certain family groups or generations.

Since Warthin was in charge of the pathological laboratory of a state hospital, this meant that he controlled a 'heavy traffic' of dead bodies from the general Michigan population. Warthin was aware of the fact that in terms of statistics, he was lucky. He had access to a significantly more representative collection of family histories and anatomic specimens than could be found in the more highly reputed charity hospitals of larger cities. Starting from the seamstress' story, Warthin and his co-workers painstakingly documented stacks of coded pedigree charts year in and year out, thus showing both the genealogy and pathology of countless relatives of cancer-susceptible families.

The point of departure of what is now known as 'Warthin's Family G' was the seamstress's German grandfather and grandmother. In the 1830s, the couple had crossed the Atlantic and settled in what was known as 'wild Washtenaw County' in what is now the Freedom Township near Ann Arbor. Like many others, Pioneer G and his wife purchased a land grant from the US government following the Indian Removal Act. They cleared woodlands, built a small log farmhouse, cultivated crops and bred children. In 1856, at the age of 60, Pioneer G died of what is believed to be cancer. He left his wife, who had no history of cancer, and ten children. If his granddaughter Pauline had not passed the information of a presumed familial cancer burden to Warthin, it is doubtful that Pioneer G and his offspring would

[7]This impressionistic account of Warthin's early research work on medical hereditarianism is based on

Warthin 1914; Stone 1927; Simpson 1931; Lynch 1985; Bentley historical library, University of Michigan; Aldred Scott Warthin papers, 1893–1931; Box 1: 'Dear friend' letters from Vienna (1893/1894); and Sir William Osler correspondence (1899–1919).

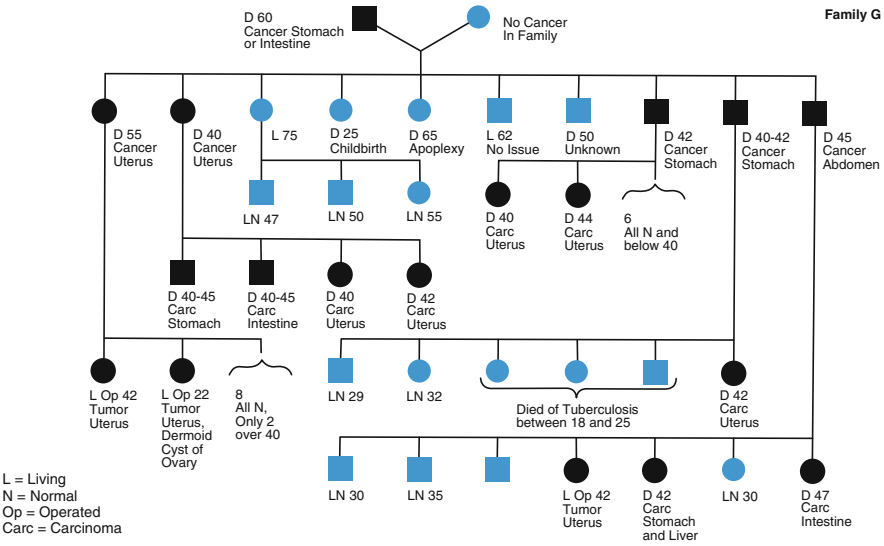

Fig. 1 Pedigree of Family 'G': By courtesy of Ca—A Cancer Journal for Clinicians (Copyright 1985)

have entered into the annals of medicine as an exemplary case of a multiple family occurrence of cancer in the Michigan area.[8]

In 1913, the Family G made its first appearance in medical literature as part of one of the most extensive early statistical studies of the influence of heredity on cancer. Out of 1600 cases of cancer, Warthin claimed that 15 % had a history of multiple family cancers. Family G was presented by Warthin as the first of four families with complete records of the descendants of a cancerous grandparent, and this stood out prominently because of the striking proclivity of cancer shown in two generations (see Fig. 1). Of the 48 descendants of the cancerous Pioneer G, apparently 17 had died or were operated on for cancer of the uterus or stomach. This family 'tendency', apparently present in the family line before the surname beginning with G, was introduced by marriage, and Warthin argued that it was so striking that Family G showed a so-called inherited susceptibility to cancer. In addition, Warthin pointed out a marked association between susceptibility for cancer and tuberculosis. The two susceptibilities seemed to run together and were believed to indicate a progressive degenerative inheritance and were associated with the development of an 'inferior stock'.[9,10]

Motivated by studies on heredity and disease, and the vogue for eugenics in the wake of the excitement over the rediscovery of Gregor Mendel's laws of

[8]Warthin 1914; Krush 1971; Remini 2001, 257.

[9]Warthin 1913.

[10]Lynch 1985.

inheritance in 1900, one would have expected strong scientific support for Warthin's study.[11] Only a few years before Warthin published his study, the internationally known cancer expert William Roger Williams quite plainly stated, 'those pathologists whose horizon does not extend beyond cells and microbes, have overlooked the chief factor in the cancer problem—that is to say, predisposition'.[12] And did not the highly reputed New York pathologist Isaac Levin (1874–1945) almost simultaneously announce that he was about to revise his view on the hereditability of the dreaded disease from 'no' to 'yes'?[13] The question of a possible familial susceptibility to cancer also excited lively interest from a new field of research—experimental animal breeding. The American researchers Ernest Tyzzer (1875–1965), Clarence C. Little (1888–1971) and the famous 'mouse lady' Maud Slye (1869–1954) pioneered efforts to trace Mendelian characteristics of cancer heritability in experimentally created lines of inbred mice.[14] If not for purely scientific reasons, Family G might have been selected as supporting the gospel of eugenics by serving as an exemplary case of a degenerative stock to be used during a eugenics exhibit.[15] After all, Warthin was a rising star within the American medical establishment and had become part of John Kellogg's (1852–1943) eugenic priesthood in Michigan.[16] However, none of these likely scenarios materialized. By 1914, the cancer research and treatment landscape was changing dramatically and so was the 'susceptibility' for cancer theory in scientific news on heredity and cancer.

3 Heredity, Eugenics and the Organized Fight Against Cancer: Family G Revisited

However promising as part of the emerging field of Mendelian genetics, medical support for the cancer hypothesis on hereditability waned. By the turn of the twentieth century, some surgeons argued that the popular belief in cancer as a hereditary disease could have negative health consequences of its own. For

[11]Rushton 1994, 59–84.

[12]Williams 1908, 374.

[13]Levin 1912.

[14]Mc Coy 1977.

[15]Eugenics can be seen as a biological theory of human improvement that was informed and vitalized by revolutionary developments in biology and medicine at the end of the nineteenth and early twentieth century. These scientific insights seemed to promise a new cure not only for a wide range of diseases but also for social problems. The social applications of the biological sciences have initiated debates about social differentiation, scientific responsibility, medical ethics, reproductive autonomy and human rights that resonate until the present day. Eugenics can equally be regarded as a social and cultural philosophy of individual and collective identity within the context of modernity; Kevles 1985.

[16]Lynch 1985; Robbins 1914 and 1915.

example, a presumed, but not proved, hereditary disposition to cancer could lead to depression and so cause cancer.[17] Twenty years later, with the rise of the organized fight against cancer, notions of hereditary cancer would become a major object of medical, social and political concern.[18]

With the support of the American Society for the Control of Cancer (ASCC), new cancer hospitals and research institutes and their specialists spread the message of *Do Not Delay*: cancer is curable, if and when detected early.[19] Within this context, we see attempts at a transformation of the responsible healthy citizen into a 'sentry patient' or 'homo medicus'—a patient ever watchful for the first signs of the dreaded disease.[20] The cancer idiom of heredity that was associated with shame, fatalism and stigmatization became regarded as counterproductive to the 'Do Not Delay' message.[21] The 'cancer prevention propagandists', as Warthin rather cynically called them, strongly believed that one of the major reasons for laymen to delay seeking medical attention was the creation of cancer-phobic states of mind by unfounded notions of hereditary and a predestination to certain doom.[22] Not surprisingly, in the propaganda literature of the ASCC, little, if any, attention was paid to a hereditary factor in the aetiology of cancer.[23] According to ASCC protagonist, the clinical pathologist James Ewing (1866–1943), in the interests of the American public, this hereditary doctrine ought to be combatted. Yes, people might pass on a liability for cancer, but cancer was not expressed until other factors were brought into play. A major building block for Ewing's anti-hereditary argument was statistical evidence from life insurance companies. Why bother with a theory of susceptibility to cancer when these companies had found no statistical evidence to pay serious attention to a history of 'cancer in the family'?[24]

The 'hereditary factor' might have been deleted completely from the 'Do Not Delay' campaign script, but did this mean that cancer and heredity were no longer up for medical debate? As Robert Proctor has shown in the interwar period, ethnic or geographic differences in cancer rates were commonly discussed in terms of racial or constitutional predispositions.[25] Given the unproblematic nature of these discussions, it is of interest to trace possible changes in the appreciation of Warthin's ongoing medical research on family cancers.

[17]Snow 1885.

[18]Patterson 1987, 38.

[19]Aronowitz 2001, 356.

[20]Pinell 2000 and 2002.

[21]Patterson 1987, 38; Aronowitz 2007, 144–162.

[22]Childe 1906, 144; Warthin 1926, 838.

[23]Bloodgood 1914; Special Committee for the control of cancer 1920, 10–11; Council on health and public instruction of the American Medical Association 1924; American Society for the Control of Cancer 1940.

[24]Ewing 1928, 109–114.

[25]Proctor 1995, 221.

In 1925, Warthin published a further study of the 'cancer' Family G.[26] In the introduction, he regrets that his first report met with little favour among the Alliance against Cancer. However, apparently the animal investigations of Maud Slye, Clarence Little and others had encouraged him to continue his research of cancerous grandfather G's offspring, which stood out as the best documented family with cancer and cancer fraternities identified in his previous study. Once again with the cooperation of the seamstress Pauline—who despite her regular visits to Warthin's department and awareness of the importance of early detection died of cancer prematurely—Warthin created a follow-up pedigree chart of the by then 144 descendants (three generations) of the original German settler and his wife. Out of the 146 individuals, 28 known cases of cancer had reportedly occurred, which was an incidence of 19.2 %. The accumulation of cancer cases was argued to be significantly in excess of the expected 10 % according to the law of probability for the whole population. According to Warthin, these findings suggested a recessive familial susceptibility to develop cancer and shown in females in the generative organs and in males in the gastrointestinal tract. He also noted (as in the case of the seamstress) a marked tendency to the sudden development and rapid course of the disease. However dramatic in terms of the presentation of clinical and statistical findings, once again, Warthin's writings on heredity, cancer and medical family research did not meet with much acclaim.

First, genealogy as a scientific method for studies on human heredity was increasingly put up for debate. The excessive popular use of pedigrees at eugenics exhibits and growing criticism against explaining human heredity in simple Mendelian terms undermined the authority of medical family research.[27] Moreover, animal and twin research had emerged as new standard methods of genetic research. Second, Warthin's public accusations of the ASCC's neglect of a hereditary factor for cancer did not help his cause.[28] And third, the 'Do Not Delay' supporters continued to keep doctors and lay people away from the perceived fatalistic associations between cancer and heredity in individuals and families.

Even in his position as editor of the Annals of Internal Medicine and president of the American Association for Cancer Research, Warthin was unable to distinguish himself from a voice crying in the wilderness. Although highly regarded as an internationally distinguished pathologist, Warthin's views on the influence of heredity on cancer in individuals and families remained controversial. Ultimately, however, Warthin was undeterred and was not influenced by his peers.

Shortly before Warthin's death in 1930, his eugenic manifest, *The Creed of a Biologist*, pleaded for the eugenic measure of marriage restrictions for those with a demonstrated heritable cancer susceptibility.[29] Warthin was especially concerned

[26]Warthin 1925.

[27]Kevles 1985.

[28]Warthin 1926; George A. Soper to Aldred Scott Warthin, letter dated 7 December 1926, Bentley Historical Archives, Warthin Papers Box 1.

[29]Warthin 1930.

about the reproduction of so-called *durchschlag* families like his Family G with a marked unhealthy family susceptibility for cancer and an associated predisposition to tuberculosis.[30] In Warthin's opinion, his new category of cancer families could only survive by breeding with individuals from families with no history of cancer and by avoiding all known extrinsic cancer-causing agents. 'He should not smoke; he should not engage in any industry in which...irritating products are used. He should not expose himself to irradiation'.[31] Given a proper and healthy regimen, the burden of cancer could be reduced even in cancer families like Family G. Although Warthin's views on the nature and magnitude of the hereditary factor differed from the mainstream, ironically he shared the optimistic and plastic nineteenth-century notion of coping with the natural history of cancer with his fierce opponents in the Alliance Against Cancer. Despite the development of new biological and medical theories in the first part of the twentieth century, doctors in the consulting room continued to regard health and disease as malleable states of being.

4 Conclusion

In my chapter, I have shown that the American cancer community was far less receptive to associations between heredity and cancer than might have been expected from the general popularity of debates on heredity, disease and behaviour in the nineteenth century. The translation and understanding of the hereditary risk factor in cancer medicine and the specific consequences for prophylaxis and treatment depended as much on the medical as on the socioeconomic and political contexts of doctoring cancer. My hypothesis is that the specific American resistance against an association between heredity and cancer in individuals and families has its origin in the rather radical translation of the 'Do not Delay' ideology by the ASCC. As part of the ASCC's economic struggle for existence, its leaders chose a straightforward and aggressive message: early detection and surgery were the only means to fight the dreaded disease. Anything that might hinder the circulation of this message was regarded as offensive, even if this implied resistance against the attractive world of brave new biology. ASCC's behaviour was in line with the curative focus that met the immediate needs of twentieth-century patients in American medicine.[32] ASCC was the leading force in the American war against cancer and was dominated by hospital doctors and entrepreneurs who shared a preference for private and technical forms of medical prophylaxis and treatment as part of a 'Do not Delay' ideology. This approach seems to be more significance in the rejection of eugenic measures than a general disapproval of eugenic measures in

[30]Warthin 1931.
[31]Warthin 1931, 696.
[32]Burnham 2015.

the face of a dreaded disease. In her book *Eugenic Nation*, Alexandra Stern has convincingly argued that the fear of disease could just as well have fuelled eugenic thinking and measures.[33] But, in the ideas and concepts across medicine and society, cancer has always been a possible, but not a necessary, outcome of a presumed hereditary or genetic predisposition, and this has created the flexibility that enabled interest groups (including Family G members) to explain and use hereditary and genetic 'at-risk' factors to their own advantage.

I also showed that in circulating between various realms, the pedigree of Family G began to take on a life of its own between 1895 and 1931 from the age of Weismannism to the age of a brave new biology. I argue that as part of this process, identity formation went both ways; as Family G changed, so did its handlers. In being 'revisited' in the medical literature in 1936 (four generations/305 descendants), 1971 (five generations/more than 650 descendants) and 2005 (seven generations/more than 929 descendants), the visibility, meaning and legitimacy of 'Family G' as a 'high-risk' cancer family continued to change.[34]

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Epilogue

Following the early scientific paper trail of Family G does not do justice to the pain, hardship, sorrow and stigma the family members had to endure throughout the twentieth century right into genomic age in coping with their genealogical disease burden and their role as objects of research. The long-term process of collecting family history data has involved intensive and emotional discussions with researchers and relatives about health, disease, death and other related aspects of personal biographies. The major question for the expanding Family G continues to be: How might the 'new' knowledge that is generated by participating in medical research benefit them?

For more than a century, scientific ideas circulated within the family about the aetiology of their disease burden from a recessive familial susceptibility (1930s), a cancer-susceptible genotype with a possible underlying viral oncogene mechanism (1970s), hereditary non-polyposis colorectal cancer (HNPCC) or Lynch syndrome associated with a possible genetic mechanism (1980s) to germline mutations in DNA mismatch repair (MMR) genes (1990s).[35] The understanding of those ideas

[33] Stern 2005.

[34] Hauser 1936; Lynch 1971; Douglas 2005.

[35] Lynch et al. 2004; Boland 2013; Necochea 2007.

within Family G circles was always associated with the hope for a cure, but, at the same time, the knowledge that their close cooperation with scientists had not yielded major therapeutic benefits or a dramatic change in the family's biography.

Following a frantic race, the headline news in 1994 that researchers had cloned the specific disease genes associated with Lynch syndrome and development of a genetic test was imminent was hailed as a victory within Family G. Predictive genetic medicine was believed to succeed where other medical approaches had failed, and the promise for an all-in-one cure for their genealogical misfortunes seemed more tangible than ever. President Bill Clinton exemplified this optimism when he announced the 'first draft' of the human genome in June 2000. Clinton claimed that for our children's children, cancer would only be known as a constellation of stars.[36]

However, in approximately 2001, the first results of the genetic tests were shared among Family G members, and their optimism quickly dwindled due to the development of disruptive family disputes over the issue of testing status. Those family members who had tested positive were confronted with complex preventive monitoring (e.g. colonoscopy) and surgical trajectories. They felt excluded by those family members who had tested negative and had no immediate medical obligation and the other way around. The professional writer Ami Mckay and Family G member, who lives in Canada, wrote and produced a most insightful radio documentary for CBC Radio 'Daughter of Family G' concerning the rather difficult decision to undergo genetic testing, what it meant to be tested and how she and other family members tried to cope with their test results. I would like to encourage all readers to learn more about this penetrating radio documentary. You will find a direct link to it here: http://www.mutantme.com/daughter-of-family-g/.[37]

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[36] http://www.thehumangenome.co.uk/THE_HUMAN_GENOME/Cancer.html.

[37] See for more information on Ami McKay's ongoing activities as public gatekeeper and Family G reporter: http://www.mutantme.com/daughter-of-family-g/.

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Genetic Discrimination in the Doctoring of Cancer and Alcoholism

Stephen Snelders, Charles D. Kaplan, Frans J. Meijman, and Toine Pieters

Abstract The genomics revolution of the early twenty-first century has stimulated the need for new appraisals of the risks of genetic discrimination in health care. Historical memories of genetic discrimination have raised serious concerns of the misuse of genetic information in the doctoring of patients. This has led to political action such as federal legislation in the United States to protect patients in both clinical practice and trials. Whether scientific knowledge of the inherited susceptibilities to disease need necessarily translate into new stigmatization and discrimination of specific populations at risk for disease has become an important topic in community genetics. Our study of the historical experiences of the application of genetic knowledge in the doctoring of cancer and alcoholism patients in the past

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century suggests that not scientific theories and evaluations by themselves lead to genetic discrimination. The crucial factors in determining whether genetic discrimination will occur are the social perceptions and evaluations of both disease and the specific at-risk population by not only the physicians themselves but the general population embedded in a specific sociohistorical context.

Keywords Genetics • Race • Ethnicity • Cancer • Alcoholism • Eugenics

1 Introduction

Medical innovations in genetics and genomics are impacting the way doctoring is proceeding in the office and the clinic. The concept of *doctoring* emphasizes a long-term perspective on the practice of medicine that focuses, despite the mystifying effects of technology and the dehumanizing consequences of large health-care delivery systems, on the sociohistorical processes and personal experiences of physicians in communicating with their patients and their families, characterized by "good days and bad days".[1] Medical practices have intersubjective characteristics that easily can be overlooked in our present era of a highly protocolized medicine. This concept extends to doctoring healthy people in preventive and diagnostic processes for diseases on the basis of their genetic susceptibility. Genetic information is increasingly of importance in contemporary doctoring expanding the horizons of personalized medicine through providing the tools for more accurate diagnoses and the introduction of new kinds of costly precision medicines.[2] However, this extension has raised concerns of new sources of discrimination that can endanger fundamental democratic ideals of equal treatment ("equipoise") and individuality and can have severe public health consequences. These new sources join the increasing awareness of the role of discrimination in the aetiology of disease and health disparities as well as the gene-environment interaction.[3]

The theories of biopolitics have critically addressed how abuses of power based on biologically formulated ideologies are increasingly used in conjunction with technology to control populations.[4] This process has caused a turn towards the integration of social science in the field of bioethics as bioethicists now are increasingly working in clinical teams to help physicians make morally defendable decisions in the doctoring of their patients.[5] These mutually reinforcing concerns of biopolitics and bioethics have also resulted in a widening of the scope of the concept of discrimination itself. Stigma, the basic social act of "marking" a person for social exclusion and discriminatory

[1] Charmaz 1991.

[2] Struse and Montoya 2001.

[3] van Os et al. 2010; Stuber et al. 2003.

[4] Foucault 2008; Patton 2007.

[5] Borry et al. 2005.

social practices, has been widely recognized as a critical social determinant of health disparities and is also applicable to the adverse consequences of genetics in doctoring.[6]

Today's new concerns of discrimination in medical guise have a dramatic historical legacy. The most prominent example from the past century is eugenics. This legacy continues to cast a shadow that can affect doctoring in many unforeseen ways. Of critical importance in understanding this legacy and its possible unforeseen consequences for scenarios of a genetics-informed doctoring for the future is the study of the historical uses of genetic information in medical knowledge and doctoring in the past century. To avoid the methodological pitfalls of overly general observations and conclusions, we have chosen in this paper for a highly differentiated analytical approach. Applying the comparative historical method, the doctoring of specific diseases of a very different nature are analysed for their particular differences in terms of the uses of genetic information in the social communication of doctors and their patients. In this study, we compare two very different relapsing diseases: cancer and alcoholism. These diseases not only differ radically in their standard medical classification, with cancer being primarily somatic and the province of internal medicine while alcoholism is clinically viewed as a neuropsychiatric disorder to be treated by the psychiatrist. These diseases and their patient populations have also been viewed and morally judged by the general public and by doctors themselves in very different ways. Generally somatic diseases are viewed as a "tragedy" where the patient cannot be blamed for their condition and the primary responsibility is imputed to the physician. In contrast, psychiatric disorders are often seen as "social inconveniences" in which the patient can be blamed for not adequately responding to "societal challenges".[7]

In the first part of this paper, we will discuss in-depth debates on genetic discrimination in doctoring by focusing on examples of the debate in leading medical journals around one particular important form: ethnic and racial discrimination. We will highlight important contributions to this discussion from the period 2003–2007. In the second part of the paper, we will present the conclusions from our historical research into the genetics of cancer and alcoholism, looking at debates in the twentieth century, detailing their relevance for the contemporary debates and linking past and present forms. We will conclude the article with a brief summary and implications for future developments.

2 Genetic Discrimination, Ethnicity and Race: Early Twenty-First Century Reports

In 2007, a survey of 1199 adults conducted in the United States found that the majority of the respondents had trust in the access of their doctors and of medical researchers to the respondent's genetic test results; however, they mistrusted the

[6]Hatzenbuehler et al. 2013.

[7]Damasio 1994, 40.

extension of access to the test results to health insurers and employers.[8] The report of these survey findings did not control for race and ethnicity of the respondent. We would expect lower degrees of trust in all categories of the medical system from ethnic and racial minority groups given the historical record of discrimination against these groups, despite American constitutional ideals. The results of the survey of 2007 suggested that the critical focus in the controversy on doctoring and genetic discrimination were not physicians and medical researchers per se, but the wider sociocultural context of special interest groups. Another critical focus was the context of public opinion. The public outcry over the race comments of the legendary scientist James Watson, who won the Nobel Prize for his part in discovering the structure of DNA, was exemplary in this respect. In the same year, 2007, he suggested that Africans were less intelligent than Westerners.[9]

The genomics revolution sparked a reappraisal in medicine of human biological differences and the application of racial and ethnic categories in assessing populations and patient groups.[10] Doctors were provided with an accelerating amount of genetic and epidemiological information concerning racial and ethnic differences to understand the aetiology of disease, categorize persons for genetic research and choose drug therapy for patients.[11] In promoting the new technology to produce this genetic information, biotechnology firms for a long time managed to avoid openly confronting the issue of race while at the same time delivering "racialized" products to the market.[12]

Many doctors seemed to feel that if race and ethnicity needed to be considered in making early diagnosis and treatment choices, then let it be so if this led to better care for the patient from these racial and ethnic groups. Critics, however, reminded the public that the reassessment of the category of race in light of the new genomics could have serious unintended consequences.[13] Using race and ethnic categories in doctoring raised concerns about reinforcing deep-seated prejudices of racial and ethnic groups with demonstrable genetic susceptibilities for disease. Minority groups, such as African Americans and Hispanics, experienced a double bind about receiving care founded on a race-based medical technology. They tended to be suspicious about race-informed genetic medicine yet felt they had no choice but to consume a race-based treatment for their own benefit.[14] These deficits in

[8]Hudson 2007.

[9]http://www.independent.co.uk/news/science/fury-at-dna-pioneers-theory-africans-are-less-intelligent-than-westerners-394898.html, accessed 17/05/2016.

[10]Cooper et al. 2003; Phimister 2003.

[11]Bhopal 2007.

[12]Duster 2007.

[13]Braun 2002; Kahn 2005; Ross and Fernandez-Esquer 2005; Cho 2006.

[14]Lynch and Dubriwny 2006.

communication of genetic information in doctoring to ethnic and racial minority group patients continue to affect the wider public health situation, providing yet another cause of health disparities in the general population.

We can refer to two American examples from around 2005 to explore the potential for race and ethnic-based doctoring of healthy individuals at risk for cancer and alcoholism. Racial factors were identified and used to advocate specific public health measures against smoking, in order to reduce the elevated risk of cancer presented by a susceptible black population.[15] Around alcohol abuse, a similar development could be observed. The Collaborative Studies on the Genetics of Alcoholism (COGA) multisite research programme of the National Institute on Alcohol Abuse and Alcoholism (NIAAA) documented significant differences among ethnic groups in both alcohol responsiveness and alcoholism susceptibility. As a means to reduce health disparities in alcoholism, NIAAA promoted studies of the links between alcohol dependence and genetic markers in specific racial and ethnic groups.[16] The question remained whether by classifying patients of minority racial and ethnic groups on the basis of genetic and epidemiological information, and not adjusting for confounding socio-economic factors such as income, discrimination and stigma, the risk was taken of unwittingly contributing to the development of unsound and discriminatory health policies that increase rather than decrease health disparities.

The historical legacy of racism, discrimination and inequalities in health care suggests that by the time evidence-based genetic medicine is firmly in place, doctors might have curbed their enthusiasm for applying racially and ethnically informed genetic knowledge because of a strong backlash from a public committed to norms and values of democracy and equality of opportunity. This is transpiring already as exemplified by the new strict guidelines of the US National Institutes of Health on research pertaining to informed consent in basic and clinical research that encompasses genomics.[17] New critical appraisals of translational research are challenging ossified concepts of linking bench and clinical science adding public and community links to the communication inherent in doctoring.[18] This is leading to more attention to community engagement and understanding the viewpoints and social context of patients beyond the immediate family. This includes the creative use of systematic focus group research to account for patient beliefs, culture and worldviews about disease diagnosis and treatment as a basic precondition to the creation of clinical guidelines affecting racial and ethnic groups.[19]

[15]Risch 2006.

[16]Russo et al. 2004.

[17]McGuire and Beskow 2010.

[18]van der Laan et al. 2015.

[19]Jones et al. 2006.

3 Genetics and Inequality in History: A Historical Perspective on the Context of Doctoring

Despite these encouraging new developments, the question remains whether the identification of genetic information with specifically defined ethnic and racial groups in itself predicts discrimination, stigmatization and possibly fatalism in the doctoring of specific diseases and patient or risk groups. A historical perspective sensitive to the sociocultural context in which physicians personally experience the current genomics revolution impacting their practice will lead to new insights, informing the future development of medical policy.[20] Here, our own historical research data tied to cancer and alcoholism provide a clinically significant contribution.[21] The specific use of genetic categories in doctoring highly depends upon the specific socio-historical context of medical practice. Without knowledge of this context, critics may be dangerously misleading both the physician and the patient, positing the hypothesis that differential genetic susceptibility to disease and therapy will necessarily lead to negative stereotypes and social consequences. Our research shows that the wider public perception of a particular disease or therapy interacting with the socio-demographic and behavioural characteristics of risk and patient groups associated with the disease has the determining influence.

The idea of an inherited susceptibility for cancer was already a research item in the first half of the twentieth century. For instance, the question of differential racial and ethnic susceptibilities was a topic of importance for the League of Nations, the predecessor of the United Nations. Breast cancer was regarded as a typical example of a cancer with a strong ethnic susceptibility, with different risk factors for different races. The scientific concept of hereditary predispositions that was then in vogue, including theories of racial susceptibility, did not however necessarily lead to racially or ethnically informed public health approaches in this historical period. Even in the racist Third Reich with its extreme eugenic policies, approaches varied with a remarkable focus on policies and programmes reminiscent of contemporary public health approaches that aimed at the prevention of the expression of cancer dispositions. For many German physicians, cancer was a constitutional and multifactorial disease, with diverse causes: genetics, diet, stress, radiation and/or industrial pollution. Part of the attraction of genetic approaches to these physicians was that they were not necessarily exclusive, but rather complementary to environmental approaches.[22]

Advocates of eugenic measures were far more prominent over the past two centuries in the case of alcoholism. Since the beginnings of the medical debate on "racial degeneration" in the 1850s, alcoholism was accorded a primary position in the theory of hereditary genetic defects. Alcohol "poisoned" the hereditary material of the abusers and of their descendants. This poisoning could be expressed in various mental and physical diseases including addiction itself. Alcoholism was

[20]Kelsey 1996; Vijverberg et al. 2010; van El et al. 2012.

[21]Snelders et al. 2006; 2007a; 2007b; 2008.

[22]Proctor 1999; Snelders et al. 2006 and 2007b.

therefore widely recognized as a medical condition. However, the alcoholics were not predestined to submit to their craving, but rather were only predisposed. Countervailing factors, including their own will, could act against addiction. Therefore, the individual alcoholic who gave in to his condition was not only ill but also morally weak and therefore of an inferior status. In the late nineteenth and early twentieth century, the eugenicists evoked a new biology that combined "medicalizing" with "moralizing". The doctoring of the alcoholic was to be informed by the new science of genetics in its clinical decision-making. This new biology would provide the diagnosis of "good" patients who were treatable and had the requisite "moral fibre" and "bad" patients who were hopeless cases.[23]

In the 1930s, new and more subtler genetics-informed differentiations among alcoholics were constructed between "made" and "born" drunkards and between treatable and untreatable categories of alcoholic patients. The treatable group received medication as well as forms of psychological, social and physical therapy, regardless of the therapeutic context. The untreatable group of "hopeless degenerates", however, ran a risk of being sterilized, or worse, as in National Socialist Germany, "referred" to concentration camps.[24]

Hence, despite scientific theories of alcoholism as a treatable and genetically influenced disease similar to theories of cancer, the doctoring of alcoholism took quite a different course than the doctoring of cancer. The social definition of the patient group was of the utmost importance. Persons suffering from alcoholism were often subjected to a process of social exclusion. The social ignorance and fear of the alcoholic resulted in a stigmatization of the alcoholic and consequent discrimination, leading to a corresponding exclusion from social support and needed medical services. In the United States, this exclusion process was accompanied by symbolic crusades that often involved pejorative stereotypes of minority racial and ethnic groups including Irish Americans, Italian Americans and African Americans.[25] In contrast, this process of social exclusion was never really possible with cancer patients. The public visibility of cancer and the unquestionable support of cancer patients continued into the twenty-first century. For example, a rally in Washington in September 2006 attracted over 10,000 participants. It is hard to imagine a similar show of public support for alcoholics despite the efforts to make their disease more visible as a public health problem of major concern to the American public.

4 Conclusions

To summarize, our research on the history of cancer and alcoholism concludes that the degree of discrimination and stigma related to doctoring patient groups depended primarily on the sociocultural context of the specific disease in question. As mentioned

[23] Snelders et al. 2007a.

[24] Proctor 1999; Snelders et al. 2008.

[25] Gusfield 1963; Tracy 2005.

in the introduction, this dependence persists through the present day when certain diseases such as cancer identified with the body by the general public are viewed as a "tragedy", while those such as alcoholism identified with the mind are viewed as moral disorders of "willpower" by public opinion despite existing medical knowledge that shows this divide to be spurious. Today, as in the past century, in doctoring patients, physicians seem still to be more constrained by the social and moral judgments of the general public and special interest groups of the specific diseases and the socio-demographic characteristics of their patient groups than by scientific concepts of inherited susceptibility.

This convergence of the present state-of-the-art contemporary doctoring with the historical data points to an important observation, relevant for our appraisal of future developments in the field of genetics and doctoring. The employment of genetic scientific categories does not necessarily have discriminatory effects (in health services, employment and insurance) although the risk groups might justifiably have such fears. Our review of the current developments in providing safeguards for informed consent in genomics and empowering communities in translational research shows signs of optimism in alleviating the sources of fear among patient populations with serious health disparities. However, today as with yesterday, it is the sociocultural context in which the public and physicians differentially perceive specific diseases that have a determining influence. We have learned from the history of cancer that there can be real positive effects of a genetics-informed historically sensitive doctoring on the reduction of health disparities. As described above through a more specific and sensitive recognition of the special medical needs of ethnic and racially defined communities made apparent by focus group research, genetic research can lead to better health care without discrimination and stigmatization. Making these special needs more visible to the public through social marketing, improved medical education and the communication of medical and genetic information to both the ethnic and racial subpopulations and the general population are the lessons that we have learned from our historical research. Participation of at risk groups in research, policy making and the development of services might also be a safeguard against the mistakes of the past. In addition, it is important for doctors to realize that genetics is not the sole key to unlocking the secrets of the causes of disease, but that it contributes in the development of constantly evolving conceptual tools for assessing needs and inequality and guiding health policy and practical action. As such, differences and similarities in the inherited susceptibility of specific diseases might even provide the basis for reinventing a more "holistic" and preventive approach for doctoring. Instead of reinforcing notions of biological determinism, doctors may emphasize the overwhelming importance of environmental factors associated with the expression of a genetic susceptibility to a disease. In doing so, doctors would reduce the risk of masking important differences that other individual characteristics might be able to reveal. This emphasis would include an understanding of the prospects and limitations of the wider sociocultural context and would hold significant promise for improving the responsiveness of at-risk individuals and their peers to doctoring in a new era of genomic medicine.

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The Genomization of Biology: Counterbalancing Radical Reductionism

Ricardo Noguera-Solano, Rosaura Ruiz-Gutierrez, and Juan Manuel Rodriguez-Caso

Abstract The term 'genome' was coined in 1920 by the German botanist Hans Winkler to describe the genetic material contained in the cell nucleus. Winkler's idea was a holistic one that emphasized the relationship between the material in the nucleus and the cytoplasm. With the passage of time, this original idea has been modified in parallel with scientific and technological progress that has led to holism being sidelined in favour of an increasingly radical reductionism. These advances have brought about significant changes in the understanding of the phenomena of heredity, from the heuristic power of the concept of the genome, resulting eventually in 'genomization', that is to say, seeking understanding of the phenomena of inheritance exclusively through the 'understanding' of genomic material in physical terms, taking a step beyond 'geneticization'. In this paper, we present the way in which genomization has followed a path that parallels the progress in genome studies, with the consolidation of the genomization of biology deriving from achievements such as the Human Genome Project and the consequent reassertion of reductionism as the dominant view. We will base our reconstruction on the original material of the authors who contributed to the knowledge of the genome, during the twentieth century in particular, combined with reflections on the impact of genomization on different fields of knowledge down the years. In this way, we hope to put forward a proposal that not only emphasizes the need to reconsider the way in which the historiography of biology has been carried out but also the impact

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that radical reductionism has had on the understanding and dissemination of contemporary biology.

Keywords Genome • Genomization • Reductionism • Geneticization • Organicism

1 Introduction

In 1920, the German botanist Hans Winkler (1877–1945) proposed the term 'genome' to describe the haploid number of chromosomes of a species, given that for each species, there appeared to be numerical consistency in the chromosomes, implying that this was among the 'material foundations of the species';[1] Winkler, of course, did not think that there was a direct relationship between the number of chromosomes and the characteristics of the species, since he was very well aware of the phenomenon of polyploidy; nor did he suggest a strong engagement with what was already known as 'phenotype'. His proposal was a construction based on the observation of chromosomal structures that appeared to show numerical regularity in a given species and which could be observed through a microscope. In an earlier paper, we showed how aspects of the history of the concept have developed during the twentieth century in parallel with advances in biology and support powerful novel heuristic biological research in the twenty-first century.[2]

The meaning of the term 'genome' changed from being understood only as a haploid set of chromosomes to a set of genes.[3] Since 1950, the term 'genome' has been related to DNA, but beyond this bare association lie developments in knowledge of the material of heredity and advances in molecular biology that have made other changes possible. After the 1950s, the concept of the 'genome' became generalized to mean a group of genes composed of DNA; subsequently, with greater technical precision, the concept was extended to the nucleotide bases.[4] In parallel with these changes, the concept was expanded, from considering only the haploid number of chromosomes to the diploid number of chromosomes.[5] This change was one of the most important because it led to the term 'genome' being thought of as including the totality of the material of heredity.[6] In a different way, the identification of extrachromosomal genetic material—such as that contained in mitochondria and chloroplasts—led to another remarkable change in the concept. For the first 30 years of its use, the term 'genome' was complemented by the term 'plasmon',[7] which represented the genetic material found outside the chromosome. Attempts were made to maintain the use of these terms to distinguish 'chromosomal

[1] Winkler 1920.
[2] Noguera-Solano et al. 2013.
[3] Winkler 1924a.
[4] Noguera-Solano et al. 2013, 218.
[5] Ibid.
[6] Ibid.
[7] Ibid.

inheritance' from 'extrachromosomal inheritance' up to the mid-1960s,[8] but with the discovery that the chromosomal material was of the same nature as the genetic material found outside the nucleus, the two concepts were merged, having the effect of extending the concept of 'genome' and rendering 'plasmon' obsolete.[9]

The concept of the genome originally arose within a line of research in which the interpretation of the nature and function of genetic material moved away from the gene-centric view or the 'nuclear monopoly' as the theory of the gene developed by Thomas Hunt Morgan and his school was known.[10] By the second half of the twentieth century, the genome concept, which had primarily been used in the field of botany, had become a heuristic research tool and was part of the new approaches to research in molecular biology and genetics. With these new uses, the genome had incorporated reductionist explanations, and it looked as though these new lines of research would be the culmination of the gene-centric vision, based on an understanding of the genome as the set of all genes containing encoded information that make the existence of an organism possible. This may be considered to be the first stage of genomization:[11] part of an illusion of being able to explain the whole organism as a function of its genome, as several authors have thought since the mid-1990s.[12]

With the development of the lines of research into genomes, including the human genome, the hard radical gene-centric vision was partially replaced by a 'genome-centric' vision, at least in the sense of understanding heredity and phenotypic expression in an integral way, such as gene interactions and epiphenomena, events that are much more complex and go beyond the simple expression of the information contained in a single gene or set of genes. At the end of the twentieth century, faced by the impossibility of understanding the nature of organisms in purely genetic terms, biological explanations that had shifted their focus towards the genome—in the sense of wanting to understand everything in terms of coding—a set of explanations was eventually constructed that gave rise to new questions,

[8]Ibid.

[9]Ibid.

[10]Harwood 1984.

[11]At this point, we would like to make a terminological clarification. As noted by Dutch physician and bioethicist Henk ten Have, explicit mention of *geneticization* began in the early 1990s with the work of Abby Lippman (1991, 1992, 1993), i.e., the extreme emphasis given to the use of genetic techniques, as well as the interpretation and description of health issues and disease based only on genetic explanations. In ten Have's words, 'this process implies a redefinition of individuals in terms of deoxyribonucleic acid (DNA) codes, a new language to describe and interpret human life and behavior in a genomic vocabulary of codes, blueprints, traits, dispositions, genetic mapping and a gene-technological approach to disease, health and the body' (ten Have, 2012). As we will see throughout the text, *geneticization* to *genomization* can even be thought of as synonyms, although the difference arises from the scope of the respective disciplines, genetics and genomics, and in that sense, the transition is from a more restricted to a broader vision, though always within the scope of reductionism.

[12]F.e. Lane 1997; Clarke 2003; Midanik 2004; Rock et al. 2007; Bell 2010.

new lines of research, and new sciences framed as the so-called omics sciences and, in particular, resulted in a space for the environmental factor being reopened as another element involved in the formation of the phenotype and nature of the organism, as well as new explanations integrated into lines of research in epigenetics. Much of the above, in our opinion, has reduced the scope of biological reductionism.

Linked to changes in the concept, which arose in parallel with changes and new developments in technology, a new framework was also being constructed. In this paper, we have called it *the genomization of biology*, understood as a process through which various explanations of biology have included knowledge of the nature, structure, physiology and organization of the 'genome' to explain the phenotypic nature of organisms in the structural, physiological and behavioural senses (when applicable).[13] Our goals in this paper are (1) to describe the role of the concept of genome for conceptual changes in molecular biology and human genetics, (2) to show overall conceptual changes based on the objects of study in biology that have shown some of the limitations of the reductionist and gene-centric interpretations, and (3) to show that the concept of the genome provides an alternative conceptual space to reductionist positions, from its early origin with Winkler's vision to the recent development of the '-omics' sciences.

2 'Nuclear Monopoly' Versus Cytoplasmic Inheritance

In the early twentieth century, Hans Winkler (1877–1945), then professor of Botany at the University of Hamburg, was determined to get to grips with the nature of heredity. Like many other German researchers, he had a keen interest in hereditary phenomena and the new research into genetics.[14] The most distinctive feature of this German tradition was an interest in the cytoplasmic material of heredity. Some of the ideas of this group of German authors in due course provided a counterbalance to what Winkler and others called the 'nuclear monopoly'.[15] This position, as we have already pointed out, assumed that heredity was controlled from the nucleus and was a controversy that caught the interest of several authors. One of them was noted plant geneticist and botanist Edward Murray East, who in 1934 noted the arguments of both sides in his reflection on the issue of the 'nucleus-plasma problem'.[16] In fact, East cites Winkler, mentioning examples of asexual reproduc-

[13]We use the term in a similar sense to Lane, 1997; Clarke 2003; Midanik 2004; Rock et al. 2007; Bell 2010, among others, though with certain differences, as we note below.

[14]Harwood 1993.

[15]On the topic of nuclear monopoly, see: Sapp 1987, 54–86; Harwood 1993, 315–350.

[16]East 1934—The nucleus-plasma problem. Amer. Nat. 63: 289–303; 402–439.

tion in brown algae, where, in East's opinion, in comparison with the nucleus, maternal protoplasm played no important role.[17]

In several experiments, working with phytocultures and members of the *Solanum* genus, Winkler began to take an interest in the relationship between parthenogenesis, chromosome number, and polyploidy, a common phenomenon in plants (and one which can result in new species). He had been interested, since 1908, in the phenomenon of parthenogenesis and its relationship with the reduction in the number of chromosomes.[18] In his examination of this relationship, Winkler proposed the term 'genome' for the first time in 1920 to indicate 'the haploid number of chromosomes, which, together with the associated protoplasm, constitute the material basis of the systematic unit'.[19] For Winkler, this 'genome' was located exclusively in the nucleus.[20]

As we can see in Winkler's proposal, there is a reference to 'associated protoplasm', which indicates that for Winkler, the nature of the specific type of the species is also related to cytoplasmic phenomena, in turn suggesting that, in the phenotypic structure of the body, not only is the information role of the nucleus included but there is also the possibility of the involvement of other cytoplasmic elements, in addition to possible interactions. This reflects the fact, as noted previously, that Morgan's theory[21] was received unsympathetically among researchers in Germany, who were far more interested in cytoplasmic inheritance and distanced themselves from Morgan's proposal, which was that the units of heredity were to be found in the nucleus, that they had a physical position within chromosomes, that changes in them were the cause of mutations, and that they followed Mendel's laws,[22] though these were neither sufficient to explain hereditary phenomena nor the developmental phenomena that brought about the nature of the organism.

In different papers written between 1908 and 1924, Winkler used terms that had already been coined, such as Hugo de Vries's 'pangene' and Wilhelm Johannsen's 'genotype' and accepted Thomas Morgan's chromosomal theory that genes are physically located in chromosomes. He did not, however, agree that the nucleus had a monopoly on inheritance. This was a point he contested in 'The role of the nucleus and cytoplasm in heredity' (1924),[23] a paper presented at the third meeting of the German Genetics Society in 1923. In this paper, he made a distinction between the

[17]East 1934, 300.

[18]Winkler 1908.

[19]In German: "...den haploiden Chromosomensatz, der im Verein mit dem zugehörigen Protoplasma die materielle Grundlage der systematischen Einheit darstellt, den Ausdruck". Winkler, 1920, 165. (Haploid chromosome: halving the chromosome number).

[20]Noguera-Solano et al. 2013, 213.

[21]Harwood 1984.

[22]Harwood 1984.

[23]Winkler, 1924a. F.e. see Pangene. In: Winkler, 1908,149; Genotype. In: Winkler 1924, 238; Morgan's theory. In: Winkler 1924a, 240–241.

'genome' (contained in the nucleus) and the 'plasmagene' (the material of heredity contained in the cytoplasm). This division had a direct impact on several authors.[24] Among these was the German botanist Friedrich Ritter von Wettstein (1895–1945), who placed still more importance on the hereditary material in the cytoplasm (or 'plasmon' as he called it), genetic factors that were sensitive to environmental conditions, that modulated changes during the development of the body and so could affect evolutionary processes.[25] Both Winkler and von Wettstein, but particularly the latter, were influenced by German botanist and geneticist Carl Correns's ideas of cytoplasmic inheritance.[26]

From the earliest reflections on the nature of the gene in 1933[27] to the most recent histories, surprisingly, little attention has been paid to the changing concept of 'genome'. In methodological and philosophical research into heredity, especially in recent decades, the concept has been looked at from within a postgenomic framework. This shows that there is a bias in the historiography that has disregarded the anti-reductionist, holistic vision in which the concept of the genome appeared.[28] We note here recent work by Maurizio Esposito, who has shown that much more may be noted on the importance of non-reductionist views of the first half of the twentieth century, such as organicism and holism, which emerged from German philosophy and science and spread later into other countries, such as Britain and the United States.[29]

3 From Botany to Other Biological Disciplines

The original concept of the 'genome' was limited to the structural composition of both sex and somatic cells, that is, the 'genome' was a structural assemblage. This perception was made possible by advances in microscopic observation associated with the development of karyotypes. Interest in the study of chromosome reduction was growing rapidly in studies of embryology and development in the early decades of the twentieth century and, later on, in botanical studies focused on hereditary transmission and the relationship between the nucleus and cytoplasm.[30]

Winkler stated that his primary concern was to establish the relationship between the number of chromosomes and the phenomenon of parthenogenesis. His observations led him to reflect on the minimum number of chromosomes essential for the origin of a new organism. In Winkler's view, 'genome' was the body or *structure*

[24]Harwood 1993.

[25]Von Wettstein 1924; von Wettstein 1926, 259.

[26]Noguera-Solano et al. 2013, 214.

[27]Demerec 1933.

[28]Noguera-Solano et al. 2013, 214.

[29]Esposito 2013.

[30]Noguera-Solano et al. 2013, 214.

with its respective protoplasm that jointly formed the basis of the systematic unit, that is to say, that in it lays the capacity for *originating* new organisms of the same species. It is appropriate then that the word 'genome' is a fusion of 'gene' and 'chromosome' (implying the set of all genes within the chromosome) and also alludes to the notions of genesis and soma (indicating the origin of a body).[31] Just to complicate matters, Winkler also seems to have used Johannsen's idea of the 'genotype' to refer to the sum of nuclear genes,[32] and, during the 1920s and 1930s, several authors used genome and genotype interchangeably to denote the group of genes located in the nucleus and plasmon to refer to the set of cytoplasmic genes.

This structural nature, consistent with microscopic observations, was, we believe, the first step towards a generalized genomization of biology, biology being understood as a science that studies life and whose objects of study are organisms and their relationships, whether of origin or interactions. Research and ideas about the genome were first incorporated into research in botany, then in genetics and molecular biology, and later in various fields such as systematics, zoology, palaeontology, and anthropology, among other biological disciplines.[33]

Even then, the term 'genome' made relatively few appearances in the scientific literature. Where it did, it was used mainly in the field of botany and referred to the 'number of chromosomes'.[34] In 1932, for instance, in the Proceedings of the Sixth Congress of Genetics, the word genome appears a couple of times, denoting the 'haploid number of chromosomes'.[35] Then, at some point in the 1930s, the term 'genome analyses' emerged to describe the practice of comparing the haploid number of chromosomes and the different states of polyploidy in plants such as wheat.[36]

In 1937, Theodosius Dobzhansky noted that the use of the terms 'genome' and 'genome analysis' was unfortunate, because it ignored the important variation occurring at the level of the gene. These terms, for instance, did little to acknowledge the recombination of chromosomal material during polyploidy that might have important consequences for a plant.[37] The nature of the genome, Dobzhansky believed, was not conserved in a homogeneous way during the process of parthenogenesis and that this was even clearer in the process of sexual reproduction.

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[31]The Oxford English Dictionary (OED) provides an etymology of the term coined by Winkler, *Genome*, consisting of an irregular form of gene + soma, the latter derived from chromosome, whereas Lederberg suggests an alternative etymology. 'As a botanist, Winkler must have been familiar with … –ome, … signifying the collectivity of the units in the stem'. Therefore, the genome should be understood as all genes collectively. See Lederberg 2001.

[32]Winkler 1924b.

[33]F.e. Emes 2003; Branco-Price 2005; Bonilla-Rosso 2008.

[34]See for instance *Cytologia I* (1930), 14; Müntzing 1930, 293.

[35]Jones 1932, 275; 369.

[36]See for instance Müntzing 1932; Müntzing 1935; Krishnaswamy 1939.

[37]Dobzhansky 1951, 216–217.

In making this point, Dobzhansky had started the transformation of the concept of the genome, from a set of haploid chromosomes to the idea of a complete group of diploid states of the cell. In 1952, the German botanist and geneticist Alfred Barthelmess (1910–1987) wrote one of the first histories on the topic of inheritance, in which he used the term 'genotype' in the same sense as Winkler's genome (the haploid number of chromosomes) but also in reference to all the genes of a nucleus 'a chromosome composition of all the genes of a cell', thereby extending the concept to diploid cells.[38] Similarly, the French word 'le génome', as used by Jean Rostand in the late 1950s, meant both the haploid number of chromosomes (as it did for Winkler) and also the complete set of genes in the nucleus.[39] However, it was not until the work of British geneticist John L. Jinks in *Extrachromosomal inheritance* (1964) that the term genome had come to mean 'the total chromosomal complement'. Jinks carried out a preliminary systematization of the terminology used up to the 1960s for the material of heredity and made two general divisions: the chromosomal and the extrachromosomal, referring to the former as the 'genome' (all the material of heredity in the chromosome as opposed to Winkler's haploid vision) and to the latter as the 'plasmon' (all the material of heredity in the extrachromosomal complement).[40]

Why did it take so long for this transformation of the concept of the genome to occur? Principally it is because the word itself remained in limited and specialized circulation. Although the publications of Johannsen, Morgan, and Hermann Joseph Muller among others had triggered a lively discussion of the relationship between genes and genetic material[41] and the gene as the basis of life,[42] most geneticists writing between 1920 and 1950 simply referred to the genetic material as the set of chromosomes.[43] From the point of view of the history of science, we may return here to what we mentioned above on how the stories of the ideas of inheritance focused on traditions that reinforced the reductionist view to the detriment of other traditions.[44]

In the decades that followed, the use of the term 'genome' became more widespread as can be seen in the writings of Gunter Stent (1924–2008), a molecular biologist who worked on the history of molecular biology,[45] and James Watson

[38]Barthelmess 1952, 293.

[39]Rostand 1957, 26.

[40]Jinks 1964, 4–5.

[41]Muller 1962, 175.

[42]Muller 1962, 188.

[43]C. H. Waddington, for example, uses terms such as genotype, nuclear material, collection of genes, and entire set of hereditary factors to discuss the material of heredity. See Waddington, 1939, 137; 322.

[44]See f.e. Esposito 2013, 95–102, 141–143.

[45]From Stent's view, 'the genome was the sum total of all genes of an individual'; see Stent, 1978, 15; 382.

(1928–),[46] promoter of the Human Genome Project, who believed that the 24 human chromosomes contained 3000 million base pairs or, as was believed at that time, 100,000 genes. Even Dobzhansky, who had expressed concerns about the term in the 1930s, came around to using it. In 1970, for instance, in a discussion of the problem of the sterility of hybrids, he wrote: 'A hybrid inherits its chromosomes from both parents, but its cytoplasm chiefly or entirely from its mother. Although the genetic information is transmitted mainly through the nuclei and their chromosomes, some of it is also carried in the cytoplasm. The genome and the plasmon of a hybrid can be distinguished. The sterility of some hybrids, especially among plants, is due to genome-plasmon incompatibilities'.[47] A little later in the same publication, he was more explicit about the way in which he was using the word 'genome': 'The analysis is made in terms of "genomes", that is, sets of 7 chromosomes each, differing in gene contents and gene arrangements, derived from different diploid ancestors'.[48]

With the widespread agreement that DNA was present both inside and outside the nucleus and with its double helical structure identified in 1953, the term 'genome' began to find use beyond the confines of the nuclear membrane. Although, in most cases, the genome still implied the set of chromosomal genes, in some contexts, at least, it came to mean the totality of genetic material in a cell—both nuclear and cytoplasmic—and as a result rendered the term 'plasmon' redundant.[49]

By 1955, there was considerable consensus as to the material nature of heredity. The German geneticist Richard Goldschmidt presented an overview of the nature of genetic material which held that any inquiry into the nature of the genetic material should begin with the following basic facts: (1) Chromosomes are the fundamental structures that, from bacteria to man, are in control of the characteristics of heredity. (2) All chromosomes are similar in structure and behaviour. In both morphology and at the genetic level, chromosomes are largely constant in size and number within a given species. (3) Chemically, chromosomes are always combinations of proteins (largely unknown) and deoxyribonucleic acid (DNA), which together form what cytologists called chromatin. Goldschmidt also suggested that genetic material in the chromosome consisted of a series of molecules of individual genes. However, despite his general approach on chromosomes and heredity, Goldschmidt believed that the germ plasm was the genetic material and did not consist of genes, but that these resulted from a structural reorganization. Although Goldschmidt considered that genes were not material substances, he clarified that DNA had been established as the main element of genetic material or at least was necessary

[46]James Watson defined the genome first as haploid set of chromosomes, with their associated genes. See Watson, 1970, 705.

[47]Dobzhansky 1970, 345.

[48]Dobzhansky 1970, 385–386.

[49]Noguera-Solano et al. 2013, 215.

for the functioning of genetic material, though he sustained that RNA could not be genetic material in the strict sense.[50]

In 1961, French molecular biologists *François* Jacob and Jacques Monod transformed the genome concept still further, describing it as akin to a genetic program: 'The discovery of regulator and operator genes, and of repressive regulation of the activity of structural genes, reveals that the genome contains not only a series of blueprints, but a co-ordinated program of protein synthesis and the means of controlling its execution'.[51]

Extending the idea of the genome from simply a repository of information to that of a much more complex unit opened the way for genomization to extend from the fields of botany and molecular biology into other biological disciplines, new research practices, and so-called big science [particularly the Human Genome Project (HGP)]. It was the determining factor in this process of the genomization of biology, a project that would critically influence the advances that biology made in the second half of the twentieth century, especially in the areas of human genetics and medicine, effecting what some term the genomization of human nature[52] and other areas such as agriculture and food production.[53]

4 Deepening Genomization

The development of recombinant DNA techniques—or genetic engineering—in the 1970s laid the foundations for a new area of scientific research on the genome, causing an explosion of research (formalized in programs and projects), scientific meetings, and publications. With the appearance of the first methods for sequencing genetic material and the publication of the first genome sequence (that of a bacteriophage), semiautomated sequencing technologies began to emerge in the early 1980s, with the first automated DNA sequencing machine, built by Lloyd Smith and colleagues, announced in *Nature* in 1986.[54] That year also witnessed emphatic discussion of the possibility of creating the Human Genome Project began, an event that suddenly propelled the concept of the 'genome' beyond the confines of the scientific community and into the realm of human health, medicine, and ultimately global society.

With respect to meetings, one of the first to focus on this new research was the *Symposium on the Genome and Chromatin: Organization, Evolution, and Function*, in Kaiserslautern, Germany, from 13 to 15 October 1978, where issues in plant genetics, chromatin, genomes, and chromosomes were discussed. Among its

[50]Noguera-Solano et al. 2013, 215–216.

[51]Jacob 1961, 254.

[52]Reardon 2005; López Beltrán, 2011; Wae 2014.

[53]Rock et al. 2007; Galesi 2014.

[54]Smith 1986, 674–679.

objectives were '(1) Orientation about current trends and results in our understanding of the organization, evolution, and function of the plant genome at the level of DNA (gene), the level of chromatin, and the level of the karyotype, and (2) Presentation of hypotheses and models which may be stimulating for further research'.[55] This meeting was particularly significant for bringing the new field of molecular biology to bear upon previous research on plants, where the concept of the genome emerged. A year later, from 11 to 21 July, 1979, the NATO Advanced Study Institute sponsored a lecture series on genome expression in plants held in Edinburgh, Scotland.

Another early conference related to these topics was a symposium at Steamboat Springs, Colorado, from 7 to 13 April, 1984, on genome rearrangement, where subjects such as recombination, gene expression, and regulation were discussed. The number of meetings on these issues has increased as a result of projects to map and sequence the genomes of different organisms including human beings with different approaches and objectives, such as the Human Genome Project, the Project of Genetic Diversity and the HapMap Project. Between 1986 and 1995, the fervour for research into 'genomes' was also evident in the publication of *Journals*. One of the first was *Genome = Génome* published by the National Research Council of Canada. From 1987 to date, other publications have appeared, for example, *Human Genome Review* (1990), *Mammalian Genome* (1991), *International Journal of Genome Research* (1991), *Advance in Genome Biology* (1992), *Genome Research* (1995), *Human Genome Project (newsletter)* (1995), and *Law and the Human Genome Science and Technology* (1995).[56]

Similarly, the publication of books on the genome has increased since the 1980s. These early books cover topics such as analysis and genome mapping, genome structure, function of the genome, the genome and cell differentiation and interaction, gene and phenotype, molecular medicine and genome evolution. One notable instance of this is Freeman J. Dyson's book,[57] which addresses issues of history and philosophy of science, and is one of the earliest reflections on the genome as a complex concept in modern science. We can see in these events and publications the widespread use and consolidation of the concept of 'genome' within the practice of modern biology. The study of the human genome has had a strong impact economically and has created new relationships between universities and industries linked to human medicine, agriculture, energy, food and veterinary science.[58]

We have, for example, the case of food, where genomization is understood to mean 'the redefinition of food consumption according to the needs for therapy, disease prevention, and enhanced wellness determined by the characteristics of an individual's genetic heritage',[59] a process that in the rush to 'individualize' food

[55]Nagl et al. 1979.

[56]Noguera-Solano et al. 2013, 216.

[57]Dyson 1999.

[58]Noguera-Solano et al. 2013, 216.

[59]Galesi 2014, 173.

exclusively according to genetic information inevitably falls into a worrying reductionism, at least from the perspective of ignoring all external factors. In the words of Davide Galesi, 'the genomization of everyday life is still a highly ambivalent phenomenon caught between the discovery of increasingly pervasive biological conditionings and the assertion of perhaps equally influential ideological constructs'.[60] What does the concept of 'genome' mean in this new era of biological research? There are various interpretations to be found in the scientific literature, most of which preserve its structural-functional nature.[61]

First, the genome can refer quite simply to the number of genes (as in 'the *Homo sapiens* genome contains between 60,000 and 70,000 genes distributed in 23 pairs of chromosomes'), an important metric for those involved intent on mapping the position of genes on chromosomes. A simplified characterization of this meaning is to conceive the genome as a specific number of chromosomes, this varying between species, but remaining constant within a species (all the genomes of a species are referred to as the 'pangenome').[62] It is worth mentioning the increasingly common use of the idea of genomization in relation to studies derived from the HGP, which seek to explain the particularities of human groups, based on '[fractionating] the genetic components [...] in various aliquots'.[63] This is being done to justify arguments and rhetoric aimed at maintaining the political status quo by establishing that the inferiority or superiority of a given group of human beings can basically be explained by its genome and thereby eliminates any possible influence of external factors.[64]

Second, the genome may refer to the number of base pairs in the nucleus or cytoplasm, a conception that has become increasingly common since the 1970s with the rapid interest in genomic sequencing and one often represented by the nucleotide bases that form the rungs of the double helix. Many scientific reports and journals define the genome in this way, ranging from the smallest genomes such as that of the bacteriophage *phi-X174* with just 5386 base pairs[65] to the human genome at around 3 billion base pairs.[66] In 1995, the first sequence of *Haemophilus influenzae* was completely sequenced (1137 bp).[67] More than 15 years later, over 180 genomes have been sequenced, including the genome of over 100 microorganisms.[68]

Third, the genome has also taken on a complex, more fluid meaning as a vast storehouse of chemical information. For evolutionary biologists, the still prevalent

[60]Galesi 2014, 184.

[61]Noguera-Solano et al. 2013, 216.

[62]Ibid.

[63]López Beltrán, 2011, 12.

[64]On the social implications of genomic studies, see also Reardon, 2005; Wade et al. 2014.

[65]Sanger 1977.

[66]Report of the Department of Energy, *Human Genome News*, 1990.

[67]Smith 1995.

[68]Metting 1997.

gene-centric explanation of biological diversity is gradually giving way to a more genome-centred vision. Genomes—or at least genomic data—can be stored on computers, opening up new avenues of research, such as synthetic biology. At the same time, advances in molecular biology have required the emergence of new scientific terminology, much of it based on the concept of the genome. 'Proteome', for instance, was coined in 1994 to describe the complete set of proteins that are expressed, and modified following expression, by the complete genome throughout the lifetime of a cell. This term is also used in a more specific sense to describe the group of proteins expressed by a cell at any particular given time. Similarly, the 'transcriptome' refers to the set of all RNA molecules, either at a particular time or throughout the lifetime of the organism, and the 'epigenome' acknowledges chemical changes in non-genetic components of DNA that are nevertheless heritable. Then there are 'transposons', 'integrons', 'introns', 'exons', 'retrons', 'invertrons', 'prophages', 'defective phages', 'plasmids', 'regulatory sequences', 'alternative splicing', 'gene interactions', and many other terms, all of which reveal the true complexity of genetic material and the need to integrate the important role of the environment—both internal and external—into explanations of the genome.[69] And with a better understanding of the variety of different forms that the genetic material can take—from bacteria with single strands of DNA to far more complex eukaryotic cells—so visual representations of the genome have had to change too.[70]

The impact of these processes of genomization has marked our own conceptions of heredity, such as the increasingly influential idea of heredity being horizontal through the processes of horizontal gene transfer, knowledge derived from the comparative analysis of genomes, or the increasingly accepted mechanisms of epigenetic inheritance, a line of research that has been heavily influenced by advances in the 'omics'.[71] Even given the widespread acceptance of biological theories that had once been marginal, such as the endosymbiotic theory proposed by Lynn Margulis,[72] many of these new 'omics' sciences have been decisive for new meanings of the genome.[73]

[69]Noguera-Solano et al. 2013, 217.

[70]Ibid.

[71]By this we are referring to disciplines that arose from the HGP, for example, proteomics, metabolomics, transcriptomics, lipidomics, as well as all the others that continue to emerge. A general definition of the suffix is 'Omics is a general term for a broad discipline of science and engineering for analyzing the interactions of biological information objects in various 'omes'. [. . .] The main focus is on: (1) mapping information objects such as genes, proteins, and ligands; (2) finding interaction relationships among the objects; (3) engineering the networks and objects to understand and manipulate the regulatory mechanisms; and (4) integrating various omes and omics subfields'. See about this site: Omics. (n.d.). Retrieved 2 August 2016, from http://www.nature.com/omics/about/index.html

[72]As a point of general interest, Margulis made the original proposal while married to Carl Sagan, which is why her surname is so given.

[73]Sagan 1967.

The idea of endosymbiosis, which Margulis first suggested in 1967, has also had an impact on our conception of the genome. Echoing Winkler's division between the nuclear genome and cytoplasmic plasmon, an endosymbiotic explanation of the eukaryotic cell suggested that it should be seen as a multi-genomed system with at least three different and specific kinds of DNA, nuclear DNA, mitochondrial DNA, and the (9 + 2) homologue DNA (according to Margulis, this kind of DNA has a common origin with flagella and cilia), as well as in the case of eukaryotic plants, chloroplast DNA. This vision of the cell strengthened evolutionary ideas about the continuity and diversity of life and from the 1960s onwards made it possible to apply the term genome to different structures: the viral genome (single- or double-stranded RNA or DNA), the mitochondrial genome (circular DNA like that of most prokaryotes), the plastid genome, and the nuclear genome. This view of the genome concept made it easier to imagine evolutionary possibilities beyond symbiogenesis, such as lateral gene transfer and the role of viruses in human evolution.[74]

As we can see, the positions in the old discussion on the prevalence of two different types of inheritance, between the prevalence of idea of 'nuclear monopoly' and the prevalence of the idea of cytoplasmic inheritance, represent at heart two aspects of a so far unknown phenomenon, the result of endosymbiotic processes that were understood in terms of symbiogenesis and the evolution of eukaryotic cells.

In spite of the significant differences in the arrangement of the hereditary material in these structures, the commonalities between the different genomes (they are all, at the very least, composed of nucleic acids) and the sequencing methods used to describe them mean that there is surprising agreement about the modern meaning of the term 'genome': in most contexts, it is understood to refer to the totality of the DNA (or RNA) or all of the material an organism has for heredity. This unification has been very clear in the language used in various biological disciplines, as well as in various other spheres such as the media, in academic and medical spaces, as well as business and commercial enterprises.[75]

5 Conclusion

We have referred to genomization as the process of incorporating knowledge from the 'omics' sciences into biological explanations in different disciplines (botany, zoology, genetics, molecular biology, systematics, evolutionary biology, evolutionary ecology) in order to understand the physiology, anatomy, behaviour, evolution and interactions of one species with another or its interaction with the environment. By genomization, we also understand the multi-faceted process of disagreement and moderation of the reductionist, gene-centred view of biology and the

[74]Noguera-Solano et al. 2013, 218.

[75]Ibid.

interpretation of organic phenomena resulting from events arising from intragenomic interactions and interactions between genetic information and the environment. We wish to emphasize the transition from geneticization to genomization, resulting from advances in genomic studies, which in short may be seen as a change of vision, at least as regards the scope allowed when considering biological phenomena, though not in its reductionist view.

As we have tried to show, the concept arose during discussions of research into heredity in parallel with Morgan's gene theory proposal, which was one of the key stages of genetic reductionism and which some authors would later refer to as being part of the gene-centric view. In the second half of the twentieth century, the greatest use of the idea of the genome—and its incorporation into the field of molecular biology—was mainly to be found, in our view, as a key event in the process of genomization, first, of particular areas of biology and then virtually the whole of biology, both in theoretical and practical disciplines, as well as other areas related to health, agriculture and the production of domestic animals, including conservation practices and ecology, and even in the increasingly common genomization of the application of justice, through the development of areas such as forensic genomics.

Based on the above, therefore, and following a similar line to Esposito, it is our belief that positions such as those suggested by Winkler should be understood by going beyond the dominant reductionist view in studies of heredity and its scope in various fields of knowledge. In its original sense, the genome provided a broad overview of the material of heredity and its relationship with the environment, not limited exclusively to understanding its physical aspect. Reductionism is a view of science that has reached its limit, and, although it remains methodologically useful, the complexity of the phenomena of life, for example, the understanding of genomes, requires us to move on to a vision such as organicism[76] that, though complex, can provide new ways of understanding genome, just as Winkler did in his day.

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[76]See f.e. Nicholson y Gawne, 2015.

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A Brief History of Uncertainty in Medical Genetics and Genomics

Reed E. Pyeritz

Abstract Uncertainty is a fact of life, and we all must cope with it in many different settings. More and more frequently, those of us in medical genetics and genomics must deal with uncertainty. My thesis is that, as our abilities to examine the human genome at ever more refined levels increase, so does the likelihood of encountering uncertainty as to the meaning of the information. The notion that uncertainty increases in some direct proportion to knowledge is far from confined to genetics and genomics. The leaders of the Age of Discovery (1450–1550) taught us that. To some degree, the notion of uncertainty in medical genetics has been formally recognized, even studied, over the decades. Examining the history of this concept can shed light on its status today, how we might confront and deal with uncertainty and what the future might hold. The era of precision medicine is here to stay. The concept holds great promise for directing specific therapy to patients who will most benefit from it and avoiding treatments in patients who are most likely to suffer adverse consequences or at best not benefit. But its application depends importantly on the proper interpretation of a person's genotype, as well as the clinical validity and utility of the genotype in a specific setting. Moreover, this evolution is taking place in a setting in which most health professionals are highly insecure in their knowledge of genetics, genomics, decision analysis and other relevant fields.

Keywords Uncertainty • Medical genetics

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1 Introduction

Soon after I was born, if my parents had inquired of my paediatrician how many chromosomes, he undoubtedly would have had to look it up, since genetics was certainly not part of usual practice in the late 1940s. However, after fairly straightforward research, my paediatrician would have confidently reported that, since I was not dysmorphic and was developing normally, I undoubtedly had 48 chromosomes, a 'fact' established in 1923. That was the prevailing state of knowledge and no authoritative source voiced any uncertainty. Then, in 1956, Joe Hin Tjio (1919–2001) and Albert Levan (1905–1998) corrected the record by demonstrating that normal human cells had 23 pairs of chromosomes. Science and medicine hardly blinked; one certainty was displaced by another, with no period of intervening uncertainty.

Over the succeeding six decades, the crude ability to count chromosomes by light microscopy evolved to the ability to sequence all 6.4 billion (or so) nucleotides of an individual's genome. How has this technological advancement affected our ability to interpret human genetics in terms of health and disease? My thesis is that, as our abilities to examine the human genome at ever more refined levels increases, so does the likelihood of encountering uncertainty as to the meaning of the information. To some degree, the notion of uncertainty has been formally recognized, even studied, over the decades. Examining the history of this concept can shed light on its status today, how we might confront and deal with uncertainty and what the future might hold.

2 Uncertainty as a Fact of Life and Science

Uncertainty is a fact of life, and we all must cope with it in many different settings. For example, financial analysts are always saying, 'markets hate uncertainty', to somehow justify their inability to achieve positive results for us. In contrast, one of the goals of science, no matter the discipline, is to reduce or, in the best circumstance, eliminate uncertainty. Joshua Lederberg (1925–2008), the Nobel Prize-winning geneticist stated toward the end of his life that '. . .the act of publication is an inscription under oath, a testimony'.[1] Yet we are increasingly aware of a disturbingly large fraction of published 'science' that is either not reproducible or outright fraudulent.[2] This clearly diminishes the willingness of our colleagues, our funders, our politicians and our general public to accept what we scientists state as the truth. For purposes of further discussion, however, I am not going to dwell on this source of uncertainty.

[1] Lederberg 1993.

[2] Altman 1983, Biagioli 2016.

3 Uncertainty in Medicine

Even as a part-time practising clinician, I recognize that uncertainty is pervasive in all aspects of medicine. For example, my oncologist colleagues are fond of stating that their patients have two possible outcomes: they live with uncertainty or they die. Of course, it is certain that we all will die, but this particular argument emphasizes that being treated for a chronic condition like cancer, even achieving a remission, is no guarantee that you will not die prematurely of that neoplasm. The entire discipline of decision analysis developed to explore the kinds of issues that clinicians, policymakers, economists and so forth should consider when forced to make choices when the alternatives were vague.[3]

Increasingly today, the focus of both translational and clinical research, as well as clinical practice, is on 'precision medicine'. This term has largely replaced 'personalized medicine', once it became clear to those who generate platitudes that all healthcare professionals have been schooled for centuries in personalizing the care they render. Although not frequently stated as such, one of the goals of precision medicine is the reduction of uncertainty. Specific care, be it counselling, surgery, medication, etc., will be delivered primarily to the people who need it and who will benefit the most. Achieving this goal has long been dependent on accurate phenotyping and clinical trials documenting an improvement in clinical history over natural history of a particular condition. Increasingly, however, both clinical trials and the practice of precision medicine depend on knowledge of the patient's genotype. This in turn depends on the analytic validity, clinical validity, clinical utility and ethical, legal, social and economic issues inherent in molecular testing, up to and including whole genome sequencing (WGS). Even the simplest indication for molecular testing is not immune from uncertainty, hence the need for accurate, understandable and sensitive counselling about the results. For example, testing the daughter of a woman with Marfan syndrome for her mother's mutation might seem completely straightforward, since a single nucleotide change might be at issue. Additionally, the testing of *FBN1* has been approved by regulatory bodies around the world. In this case, a negative result is definitive; the girl does not have Marfan syndrome. On the other hand, finding that she has inherited her mother's mutation will label her as having the condition, but offer little 'precision' as to what the future holds. It will certainly enable, even mandate, careful follow-up, prophylactic medication, exercise limitations and other measures to reduce the risk of a catastrophic event, but will not be entirely predictive. In all such cases, the issue of *variability* raises its spectre. As yet, we have little understanding of the various factors—the other allele, modifying genes, epigenetics and chance among them—that will influence the phenotype of the Marfan syndrome or any other Mendelian conditions. In a slightly more commonplace example, if rather than Marfan syndrome, the daughter was being tested for her mother's pathologic mutation in *BRCA1*, even finding the mutation would label her as being nothing more than a

[3] Weinstein 1986.

'carrier', because of the extreme of variability, *nonpenetrance* that pertains to many cancer-predisposing genes. So someone with a pathogenic mutation in *BRCA1* has a 90% and 55% lifetime chance of developing breast or ovarian cancer, respectively.

By testing panels of genes that are coalesced around a particular phenotype, such as the familial aortopathies, I weekly encounter these same issues, but in addition introduce one of the most important sources of uncertainty, the dreaded *variant of uncertain significance* (VUS). Typically, a VUS is a single nucleotide change that is not reported to cause the disease of interest (or any disease) in any database, but it does alter an amino acid, especially a conserved one across species. Struggling with the import of one or more VUS's in a molecular genetic test result is going to be with us for the foreseeable future. Eventually, various approaches, especially the bioinformatics analysis of 'big data', will reduce, and hopefully eliminate, the clinical uncertainty that attends VUS's. And we do get smarter about any particular VUS as time passes. Realistically, it is virtually impossible for any practitioner or clinic to keep accurate track of all the VUS's that patients accrue.[4] Further, it is costly in various ways to attempt to recontact individuals who had one or more VUS's as part of their lab report. As a result, the notion of a *duty to recontact* has occupied professional organizations in the past [5] and continues to be vexing today.[6]

As molecular testing expands beyond disease-focused panels, all the previous issues persist, but in addition the likelihood of *incidental findings* becomes a concern. These are presumed pathogenic (but also VUS) findings in genes unrelated to the condition for which testing was ordered, but revealed as a result of the testing. The *American College of Medical Genetics* famously identified 56 genes that, should a pathogenic mutation be found, as a part of reasonable medical care, it would need to be reported to the patient.[7] This has engendered considerable controversy, not the least of reasons being that the list is sure to expand. For a growing number of patients, such results are emerging not as the result of clinical testing but as part of research projects.[8]

Another current aspect of uncertainty is that whole exome sequencing (WES) ignores the 97–98% of the genome that does not encode 'traditional' proteins from sequences that have characteristics of 'traditional' genes The majority of the genome not sequenced by WES is somewhat *terra incognita*, but is increasingly recognized to encode microRNAs that have a role in controlling traditional genes as well as sequences that encode small peptides which may have unclear but potentially important roles in gene expression and function. So a report that states 'Your WES appeared normal' should engender less and less reassurance, and more and more uncertainty as to whether a genetic factor is involved in the disorder.

[4]Cheon 2014.

[5]Hirschhorn 1999.

[6]Pyeritz 2011, Hastings 2012.

[7]Green 2013.

[8]Wolf 2012, Wolf 2013, Pike 2014, Thorogood 2014.

4 Historical Underpinnings of Uncertainty

The presence of uncertainty in medicine and science, indeed, in life, has long been emphasized by some of society's greatest thinkers. Hippocrates (ca. 460–370 B.C.), in his *Aphorisms*, stated that 'Life is short, and Art long; the crisis fleeting; experience perilous, and decision difficult'.[9] Sir Thomas Browne (1605–1682), in 1643, recognized the frustrations inherent in uncertainty: 'It is better to sit down in a modest ignorance. . .than buy the uncertain knowledge of this life with sweat and vexation'.[10] Additional scholars, who we now consider among the founders of statistics, had their work stimulated by coping with uncertainty. More than 300 years ago, Jacob Bernoulli (ca. 1654–1705) in his *Ars Conjectandi* (1713) anticipated our growing infatuation with 'big data' when he noted that the variability in an estimate goes down as the sample size increases. Reverend Thomas Bayes (1701–1761) supported the notion of iterative databases when, over 250 years ago, he recommended that we sequentially learn from experience and steadily update our beliefs as more data become available. There is no evidence, despite the focus on morality in his principal profession, that he anticipated the duty to recontact as we became wiser. Closer to our own age, the US Supreme Court justice and legal scholar, Oliver Wendell Holmes, Jr. (1841–1935), emphasized that we need to become accustomed to uncertainty. In his *The Path of the Law* in 1897, he stated, 'Certainty generally is illusion, and repose is not the destiny of man'.[11] Shortly thereafter, in giving advice to medical students at Johns Hopkins, William Osler (1849–1919) cautioned, 'A distressing feature in the life which you are about to enter. . .is the uncertainty which pertains not alone to our science and art, but the very hopes and fears which make us men. In seeking absolute truth we aim at the unattainable, and must be content with finding broken portions'.[12] He also opined, 'Medicine is a science of uncertainty and an art of probability'.

Other than recognizing its existence, however, historically little attention was paid to the implications of uncertainty in medical practice. This began to change in the mid-twentieth century, especially with the pioneering work of Renée C. Fox (b. 1928) at the *University of Pennsylvania*. He empiric studies of the role of uncertainty in the practice of healthcare professionals and the expectations of society earned her considerable renown, as well as an appointment as the *Annenberg Professor of Social Sciences*. She documented that uncertainty was a persistent, yet changeable, attribute of medical science, research and practice.[13] Looking at it from the opposite perspective, she noted that the predictable rise in public expectation of the benefits of medical research was paralleled by a lowered tolerance for ambiguity in the implications of the progress.[14] She followed up on

[9] www.azquotes.com/author/22138-Hippocrates.

[10] Browne T. Religio Medici, 1643.

[11] Holmes OW Jr. The Path of the Law. Harvard Law Review, 1897.

[12] Osler 1922.

[13] Fox 1959.

[14] Ibid.

Osler's observations by recommending that medical students be specifically taught about uncertainty and how to deal with it. She also noted how inextricably linked the concepts of uncertainty and risk are, in many settings.[15] One of points of emphasis was to find scientifically adequate, culturally appropriate and socially effective means of appraising, communicating and, if possible, controlling risk—daunting challenges to say the least. This seminal work spawned considerable theoretical and empirical study of uncertainty. Hillman and Goldsmith, writing in the context of medical imaging, emphasized the importance of uncertainty to all medical disciplines. They asked, 'How does lowering uncertainty by a small fraction change the care patients receive or improve the quality of their health'?[16] They arrived at the pessimistic conclusion that searching for medical certainty is an impossible quest.

5 Empiric Studies of Uncertainty

Around the time that Renée Fox's studies were ending, empiric studies of uncertainty were being extended to medical genetics. The spousal team of Abby Lippman and Clarke Fraser conducted structured interviews with women who had undergone reproductive genetic counselling. They were able to parse the nature of uncertainties into three categories: (1) ambiguity about the total impact of a clinical condition, (2) dealing with the burden of decision-making and how others would view choices and (3) concern about their ability to fulfil their expected roles as parents.[17] This seminal work stimulated many further empiric studies in the then nascent field of genetic counselling. Genetic counsellors assist patients and parents deal with uncertainty by suggesting coping and adaptation strategies.[18] Understanding the prognosis of a condition, even if it is grim, can reduce uncertainty.[19] While severity of a condition and uncertainty are directly correlated, developing a sense of optimism and control can render parents of a child with a genetic condition less uncertain.[20] Uncertainty couched in the context of future research progress can lead to optimism, while expressed in terms of questionable, poorly understood outcomes can lead to disillusionment.[21] Our research group at Penn examined how health professionals conveyed the results of chromosomal microarray testing and how patients interpreted the results. Through interviews with families that received news

[15]Fox 1980.
[16]Hillman 2010.
[17]Lippman 1979.
[18]Lipinski 2006.
[19]Truitt 2012.
[20]Madeo 2012.
[21]Biesecker 2014.

of either a pathogenic result or a variant of uncertain significance in a child, we identified three domains of understanding:

First, did the parents understand the results?
Second, how did they interpret any uncertainty that was part of the results?
Third, how did they appreciate the personal meaning of the results for their child and themselves?[22]

We found that in the first domain, most families had incomplete comprehension of the results, regardless of whether the finding was clearly pathogenic or a VUS. Contributors to misunderstanding included receiving the results from a non-medical geneticist, receiving the results by telephone, having a protracted time to receive genetic counselling and having performed their own internet searches that proved misleading. Health professionals, who were not trained in medical genetics but who ordered chromosomal microarrays, also frequently had incomplete comprehension of the results, which included uncertainty of both pathogenic findings and VUS's.[23] Most of these non-geneticists did not view pretest genetic counselling necessary but felt ill-equipped to deal with uncertain results and wanted to refer the patients to medical geneticists subsequently.

Professor Barton Childs (1919–2010) of Johns Hopkins, one of the founders of the specialty of medical genetics, emphasized the distinction between how the scientist and the clinician view uncertainty. The biologist, for example, relishes uncertainty because eventually hypothesis-based investigation is stimulated. 'The physician, in contrast, must tolerate, even embrace, uncertainty and ambiguity'. One goal of clinical investigation is to 'reduce or eliminate the uncertainty and ambiguity of decisions that may have to be made in the absence of complete understanding'.[24] He went on to describe the evolution of the meaning of *gene*, from a statistical abstraction to a physical entity. The particular model of a gene one chooses will determine its interpretation, especially in a clinical setting. This need to define the particular nature of the gene in a particular setting or experiment has been emphasized in a recent best-selling book.[25] If we choose what Childs calls the *molecular gene*, then it is defined by its sequence. And as noted above, sequence data are becoming a commodity: relatively inexpensive to obtain and store, potentially determinable at birth or prenatally from mother's plasma and presumed to be invariant (other than through somatic mutation) throughout the lifespan. Over time, we have come to realize that this seemingly straightforward notion of the gene carries considerable baggage. As health professionals, patients and the general population consider 'sequence data', they are increasingly confronting concerns about ancestry, relationships, identity, privacy, clinical utility and ownership, to name but a few.[26]

[22]Reiff 2012.
[23]Reiff 2013.
[24]Childs 1999.
[25]Mukherjee 2016.
[26]Jackson 2011, Burke 2016.

Periodically, those of us involved in genetic and genomic medicine should step back and consider uncertainty in its broader contexts. Lawyers, judges and juries seem comfortable talking in terms of 'reasonable medical certainty' while recognizing its fallibility. At the other extreme of practical application, several noted scholars have produced books that tackle the notions of uncertainty in science and their implications for society.[27]

6 The Future

The era of precision medicine is here to stay. The concept holds great promise for directing specific therapy to patients who will most benefit from it and avoiding treatments in patients who are most likely to suffer adverse consequences or at best not benefit. Its evolution will be replete with fits and starts. Already there are examples of medical malpractice suits and settlements based on clinicians' failure to offer genomic testing in a variety of settings.[28] Malpractice claims are known to relate directly to the emergence of new technologies, and clinicians are often caught in a bind, in large measure based on uncertainty. They are at risk if they apply new technologies 'too early' and cannot interpret the results accurately or do not recontact their patients when better interpretations are possible.[29] If they delay (the 'late adopters') until the utility of the technology is more certain, they can be accused of withholding a beneficial service.

'Scientific results are always provisional, susceptible to being overturned by some further experiment or observation. Scientists rarely proclaim an absolute truth or absolute certainty. Uncertainty is inevitable at the frontiers of knowledge'.[30] So, for the foreseeable future, we all will have to continue to deal with uncertainty in genetics and genomics. David Botstein (b. 1942) suggested that the burden ultimately will be diminished, if not lifted, as we come to understand the function and dysfunction of all 6.4 billion base pairs (and the mitochondrial chromosome presumably).[31] Attempting this task is not trivial. For example, Jay Shendure chose a 6-base pair region of *BRCA1* to examine in detail. His group performed what they termed 'saturation editing' of those six bases and examined the in vitro impact on the protein's function. This required examining all 4048 possible combinations of sequence variation in that short region.[32] Imagine extending this sort of analysis to the 5000 nucleotides that comprise the coding sequence, before including the splice sites, controlling elements and so on. But even if this does come to

[27] Howlett 2011, Nowotny 2015, du Sautoy 2016.
[28] Marchant 2013.
[29] Pyeritz 2011.
[30] Achenbach 2015.
[31] Botstein 2012.
[32] Findlay 2014.

pass, we will have to deal with the interactions among the multiple variants we all carry and the epigenetic and other influences on expression.

A recent search of PubMed for references mentioning both 'uncertainty' and 'medicine' retrieved more than 68,000 citations. A search that linked 'uncertainty' and 'genetics' produced more than 30,000 references. There will undoubtedly be many more before those of us who apply medical genetics and genomics feel totally confident in our work. And long before that day arrives, there will have been numerous policies and standards of practice established based on uncertain results.

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Part IV
Countries

"Nature's Laboratories of Human Genetics": Alpine Isolates, Hereditary Diseases and Medical Genetic Fieldwork, 1920–1970

Pascal Germann

Abstract Genetic studies conducted in supposedly isolated populations played an important role in post-war era human genetics. Some of the most prominent figures in human genetics were involved, and they regarded the small, purportedly isolated communities they examined as ideal study units for investigating hereditary diseases and variations in genetic traits. Yet despite the significance of these "isolates", which were regarded as "laboratories of human genetics", the studies conducted have, in the historiography of human genetics, attracted only limited scholarly interest. In particular, scant attention has been paid to the connections between this post-war genetic fieldwork and earlier traditions that embraced medical studies of inbreeding and eugenic population research. In this case study, I will address this shortcoming by examining a long-term project of medical genetic research conducted in Swiss alpine isolates by Ernst Hanhart, a pioneer in medical genetics in Switzerland. Between 1920 and 1970, Hanhart and his co-workers systematically scrutinized isolated communities in the Swiss Alps, gathering vast amounts of genealogical, demographic, medical and blood-group data in order to investigate the suspected Mendelian inheritance of numerous pathologies. Having its roots in what Daniel Kevles called "mainline eugenics", Hanhart's project aimed at controlling the heredity of entire populations; it shared ideas and concepts with racial science and was much admired by Nazi eugenicists. After World War II, Hanhart's eugenic survey was recast as a medical genetic study of isolates. Typical of such post-war studies, the project now stood aloof from notions of traditional eugenics and racial science. However, I will argue in this chapter that World War II was not in fact a watershed moment cleanly dividing the older and new practices of hereditary research. By analysing the research practices in the field, I will show instead that there were some unexpected continuities. In order to mould alpine

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communities into “genetic isolates” and “nature’s laboratories”, Hanhart drew on social, ethnic and racial categories and narratives. As a consequence, the hereditary diseases scrutinized became linked with pre-existing categories of racial and social difference, and as my contribution will show, practices, concepts and ideas of eugenic population research and racial studies accordingly found their way into post-war medical genetics.

Keywords Medical Genetics • Eugenics • Isolate • Race • Hereditary Disease • Switzerland

In 1943, in the middle of World War II, the newly founded Swiss Society for Genetics held a conference in Schaffhausen. The participants had just created a national committee for human genetics, thereby laying the groundwork for the institutionalization of the discipline in the post-war era. Ernst Hanhart, a professor of medicine at the University of Zurich and one of the two keynote speakers, enthusiastically championed to the assembly the advantages of medical genetic research in Switzerland. “No country of the world”, he said, “is more suitable for the study of hereditary diseases than Switzerland”,[1] referring to the numerous remote villages in alpine valleys that were supposedly untouched by modern processes of migration. Working under the assumption that the inhabitants of these villages habitually married within their communities, Hanhart and other human geneticists regarded the villages as ideal sites for medical genetic field studies. As the reproductive isolation of these “isolates” made recessive diseases more easily detectable, Hanhart labelled them “nature’s laboratories of human genetics”.[2]

Between 1920 and 1970, Hanhart and his co-workers scrutinized these isolated communities, gathering vast amounts of genealogical, demographic, medical and blood-group data in order to examine the suspected Mendelian inheritance of a large variety of pathologies. Hanhart’s large-scale project was a success. His Mendelian analyses of over 60 pathologies were well received internationally, particularly in the post-war era, and some of these pathologies still bear his name. Hanhart syndrome, for example, is a rare birth defect that causes an undeveloped tongue and malformed limbs, and Hanhart–Rychner syndrome is an autosomal recessive metabolic disorder. Up until today, Hanhart is regarded as one of the pioneers of medical genetics in Switzerland.[3]

In many respects, Hanhart’s project was undoubtedly exceptional, particularly in its longevity; at the same time, myriads of studies performed in the time period between 1920 and 1970 were built upon similar research practices, methods and

[1] Hanhart 1943, 632.

[2] Hanhart 1954b, 925.

[3] See, for example, Beighton/Beighton 1997, 79. On the early development of human genetics in Switzerland, see Germann 2016, Ritter 2009, 175–196 and Geiser 2002.

ideas. Up until the 1960s, a great deal of the research on human heredity that contributed to the development of human genetics consisted of field studies that rested upon data gathering and statistical tools. The historiography of human genetics, however, has tended to neglect this important tradition in field research. Instead, historians of human genetics have established a narrative that focuses on innovations of laboratory research such as the development of biochemical methods for detecting genetic diseases and the discoveries of human cytogenetics in the late 1950s.[4] From such a limited perspective, human genetics seemed to evolve into a scientific research field only after World War II.[5] By neglecting the persistence of concepts, research styles and methods that were forged in the first half of the twentieth century and earlier, the dominant narrative also tends to clearly demarcate human genetics from eugenics. To be sure, this textbook narrative has been challenged. Recent historical accounts have broadened our view on a much wider range of scientific ideas and practices concerned with human heredity,[6] and since the 1980s, historical studies have shown that the science of human heredity and the movement of eugenics have an intertwined history.[7] However, most historians have stuck to clear lines of demarcation.[8] Many studies, for instance, have adopted Daniel Kevles's distinction between *mainline eugenics* and *reform eugenics*,[9] seeing the science of human genetics as evolving out of a reformed eugenics that had distanced itself from essential ideas and concepts of mainline eugenics, including the idea of forced sterilizations, the biologization of social problems, a strong hereditary determinism and the tied connections to racial science and ideology.[10] This interpretation has real merit but also some shortcomings. First, Daniel Kevles primarily addresses developments in the USA and Britain, and it is problematic to apply his interpretations to other countries. Second, his interpretation omits the persistence of those notions, methods, and practices that linked post-war human genetics to traditional eugenics. By focusing on a different national context and on a research field that has so far gained only limited attention, this chapter will highlight historical trajectories that challenge the dominant narrative.

In the following pages, I will shed light on the medical genetic fieldwork carried out in small, supposedly isolated communities, taking Hanhart's research in Swiss

[4]The most comprehensive history of human genetics is Harper 2008. This study is immensely insightful and well-researched but tends also to focus on the innovations of laboratory research.

[5]Susan Lindee, too, takes such a point of view in her excellent study of medical genetics in the "long 1960s" Lindee 2005.

[6]See in particular Thomaschke 2014, Gausemeier et al. 2013 and von Schwerin 2004.

[7]To cite just four classics: Kühl 2013, Paul 1995, Weingart et al. 1992 and Kevles 1985.

[8]An exception in this regard is Nathaniel Comfort's innovative study of the history of medical genetics in the USA. He argues against the notion that the study of human heredity evolved from a focus on eugenics to a focus on medical therapy. In his history of human genetics, eugenic *and* medical aims are shown to have persisted jointly throughout the twentieth century. Comfort 2012.

[9]Kevles 1985.

[10]See, for example, Kühl 2013 and Roll-Hansen 2010.

alpine valleys as an example.[11] On the one hand, Hanhart's program of genetic population research and data collection had its roots in what is called "mainline eugenics". Eugenic population studies in small communities often combined social, medical and genetic issues with questions of racial mixture and purity, and the collected data were deployed to construct huge pedigrees which buttressed gloomy scenarios of degeneration and served as visual arguments for eugenic measures.[12] On the other hand, as the historian Susan Lindee has emphasized, research in isolated populations was a booming field of human genetics in the 1950s and 1960s.[13] Some of the most prominent figures in post-war human genetics were involved, and these geneticists often explicitly rejected old eugenic and racial notions.

But was there really a watershed moment which divided the older and newer practices of genetic population research? To what extent were these traditions interconnected? In my paper, I will focus on the question of how categories of difference were used in medical genetic studies of isolated populations. In particular, I will highlight the role of ethnic and racial categories within this research process. No doubt these categories were pivotal for "mainline eugenics". But what role did they play in medical genetic fieldwork in the post-war era?

To begin, I will give a short outline of Hanhart's large-scale project, its eugenic framework and its international reception. Afterwards, I will highlight two pivotal aspects of medical genetic fieldwork: the construction of "isolated populations" and the construction of "hereditary diseases". In these two parts, I will focus on the categories geneticists drew on in order to shape and define the objects of their study. Finally, I will argue that the dividing line between eugenic population research and post-war genetic fieldwork was not as clear-cut as it would seem. The case study reveals some striking continuities in the use of racial, ethnic and social categories.

1 Ernst Hanhart's Long-Term Project and the Changing Framework of Eugenics

When Ernst Hanhart started his project in the 1920s, his aims were ambitious and at the same time amorphous. The project's goal was to scrutinize every single population in Switzerland that was regarded as "isolated" and to register all hereditary pathologies that could be found.[14] No doubt, Hanhart's study had its

[11]My analysis of Hanhart's project rests upon an examination of the comprehensive archival material contained in Archive MHIZ, Nachlass Ernst Hanhart, PN 56: 1–101.

[12]See, for example, the comprehensive survey that Herman Lundborg conducted of an "isolated" population in Sweden: Lundborg 1913. Hanhart often made reference to this eugenic population study.

[13]Lindee 2005, 58–89. So-called isolates were also important research sites for genetic studies of human variation. See Lipphardt 2014, Lipphardt 2013 and Lipphardt 2010.

[14]Hanhart 1924, 3.

roots in mainline eugenics. Tellingly, the project was funded by the affluent Julius Klaus Foundation for Heredity Research, Social Anthropology and Racial Hygiene. Typical of mainline eugenics, the Zurich-based foundation combined eugenic aims with racial notions of white supremacy. Falling under the foundation's aegis, stated its bylaws, were all efforts based on scientific grounds whose "final goal is directed towards the preparation and implementation of practical reforms to improve the white race".[15] Due to its generous funding of hereditary research, the eugenic foundation made crucial contributions to the development of human genetics in Switzerland.[16] For Hanhart too, the foundation's support was vital. Thanks to its periodic grants, Hanhart always had sufficient means to successfully carry out and extend his research in alpine isolates. In programmatic texts, he emphasized a eugenic agenda and asserted that his studies made eugenic population control possible.[17]

In the 1920s, Hanhart's project received little attention within the scientific community outside Switzerland. After 1933, however, the Swiss geneticist gained a high degree of scientific prestige in Germany. The Nazi regime's unprecedented support of heredity research and eugenics opened new opportunities and spaces of resonance, which Hanhart was willing to take advantage of.[18] He published widely in the newly founded journals of human heredity and racial hygiene; he gave lectures at conferences and, in cooperation with German colleagues, co-edited the first comprehensive compendium of human genetics.[19] Hanhart was able to align his research program with politically motivated research interests in Germany in two ways. Firstly, in his papers he often addressed research problems that arose as a result of eugenic legislation in Germany, such as the problem of precisely defining the medical conditions that met the terms of the sterilisation law. Hence, in German journals Hanhart particularly emphasized the "practical relevance" of his research.[20] Secondly, by reconstructing the ancestry of entire populations and the geographical distributions of hereditary diseases, Hanhart's surveys displayed affinities with racial research. Accordingly, in his publications Hanhart frequently raised questions about how geography, race and the frequency of hereditary diseases were interrelated.[21] Because of such alignments with research fields relevant to eugenics and racial politics, Hanhart's studies were widely and positively received in Nazi Germany. Indeed, authorities of German racial hygiene held up

[15]Stiftungsreglement 1922, 5–6.

[16]Germann 2016, 37–64.

[17]See, for example, Hanhart 1924.

[18]On human genetics in Nazi Germany, see in particular Weiss 2010, Cottebrune 2008, Schmuhl 2008 and von Schwerin 2004.

[19]Just et al. 1940. For example, Hanhart published several articles in "Der Erbarzt", a journal founded in 1934 by Otmar von Verschuer, a leading eugenicist in Nazi Germany. See UAZ, AB.1.0378, Dozierendendossier Ernst Hanhart (1891–1973), Verzeichnis der wissenschaftlichen Arbeiten.

[20]Hanhart 1938, 612.

[21]See, for example, Hanhart 1941.

his genetic field studies in the Swiss Alps as models for eugenic population surveys in Germany.[22] In 1941, the Reichs Ministry of Education funded an expedition by Günther Just, a leading German geneticist, to visit the field sites of Hanhart's comprehensive population studies.[23]

Up until World War II, research in Switzerland on human heredity was strongly dominated by researchers at the University of Zurich; after 1945, however, Geneva developed into the new centre of human genetics. The leading figures in Geneva were the ophthalmologist Adolphe Franceschetti and the psychiatrist David Klein, two medical scientists who would go on to become the most renowned human geneticists in post-war Switzerland. In 1955, Franceschetti and Klein jointly founded the first institute for human genetics in Switzerland, an institute which Klein headed in Geneva from 1955 to 1978.[24] But despite the increased importance of Geneva, Hanhart and his project attracted a great deal of attention after the war. Both Klein and Franceschetti emphasized the crucial, pioneering role of Hanhart's research. "We wish to stress the extreme importance of systematical investigations of isolated populations",[25] Klein stated in 1964, referring to Hanhart's studies. And Franceschetti simply asserted, "It was Hanhart's scientific merit that Swiss genetics enjoyed such great international prestige today".[26] Klein and Franceschetti finally convinced Hanhart to bring his data—against the will of Zurich-based geneticists—to Geneva, where they used it for further research.[27]

Internationally, Hanhart's project also attracted considerable attention in the post-war era despite the close relations he had cultivated with eugenicists in Nazi Germany. In 1947, for example, Hanhart was invited to London for the renowned Galton Lecture, the first such lecture held in 8 years.[28] This invitation was both an honour for the Swiss geneticist—the Galton Laboratory in London was, internationally, the most prominent research centre in human genetics in the post-war era—and a challenge as well. It was Hanhart's first journey to England and probably never before had he presented his project to an audience that was so

[22]At the Twelfth Conference of the International Federation of Eugenic Organizations in Scheveningen (Netherlands), for example, Hanhart outlined his research project in a paper that was enthusiastically received by the German delegates. In an internal report, probably written for the Reichs Ministry of the Interior, it was highly recommended that surveys similar to Hanhart's be conducted in Germany. MPIP-HA, GDA 37, Bericht: Internationale Eugenische Versammlung in Holland.

[23]UAG, PA 229, Just, Günther, Personal-Akte, Reichsminister für Wissenschaft, Erziehung und Volksbildung to Universitätskurator Greifswald, 23 May 1941.

[24]On the establishment of medical genetics in Geneva, see Geiser 2002.

[25]Klein, Ammann 1964, 129.

[26]CH-BAR#E9510.10#1987/32#558*, Az. 3.1-502, Prof. Dr. Ernst Hanhart, Ascona: Monographie über Mongoloidismus und Arbeiten über die Erblichkeit von Krankheiten und Missbildungen, 1961–1962, Franceschetti to Bundesrat H.-P. Tschudi, 12 July 1962.

[27]Archive MHIZ, Nachlass Hanhart, PN 56. 1:70, Institut de Génétique Médicale (Genève) to Hanhart, 1 December 1962.

[28]Deaf-Mutism in Switzerland 1947.

critical of traditional eugenics.[29] Just as in his prior talks which had aroused the admiration of German eugenicists, Hanhart presented pedigrees and first evaluations from his ongoing research project on alpine populations, and he repeated his credo that "Switzerland was an excellent theatre for genetic study because of the many villages which had been virtually isolated for centuries".[30] In one significant aspect, however, Hanhart's labelling of his study had changed. Omitting any references to a eugenic agenda, he now framed his project as a medical genetic study of isolated populations.

This example shows how Hanhart managed to cope with the politically changed climate after World War II and establish ties to the new leading centres of human genetics, which were no longer in Germany but in the USA, Scandinavia and, especially, Great Britain. The leading geneticists in these centres often distanced themselves from traditional eugenics and racial science, and in this new milieu, having altered his project's embedding and labelling, Hanhart continued his research project undeterred. By transforming his project from a population survey of racial hygiene into a program of medical genetic studies of isolates, Hanhart had successfully placed his research within a booming field of post-war human genetics.

To be sure, Hanhart stuck to his eugenic convictions in the post-war era. He continued to act as a eugenic marriage counsellor and as a eugenic expert in assessing sterilisation measures.[31] Until his death in 1973, the Swiss geneticist always regarded eugenics as an essential and incontestable application field of human genetics. Hanhart's rhetoric, however, shifted towards a more moderate stance that could be labelled "liberal eugenics"[32]. Instead of backing coercive measures enforced by the state, he now appealed to individuals to take personal responsibility by considering eugenic principles when they married. Like many human geneticists in the post-war era, he was convinced that eugenics should be put into practice not by force but through free choice.[33]

After World War II, Hanhart also became increasingly critical to racial science. As a consequence, the antiracist anthropologist Ashley Montague referred to him in 1962 as one of the human geneticists who fundamentally opposed the concept of race.[34] As we will see, Hanhart's view on this point was more ambiguous than Montague maintained, though indeed there were programmatic statements that justified such a claim. For instance, Hanhart had voiced doubts about assertions of "racial predispositions" for diseases; he had referred to the efforts of racial

[29]On the Galton Laboratory in London, see Harper 2008, 235–240. Especially critical of eugenics was Lionel Penrose, who occupied the Galton Chair at the Galton Laboratory. See Ramsden 2013, 47 and Mazumdar 1992, 251–253.

[30]Deaf-Mutism in Switzerland 1947, 703.

[31]Archive MHIZ, Nachlass Ernst Hanhart, PN 56.1:72 and PN 56.1:73.

[32]Habermas 2001.

[33]Hanhart 1951, 3; Hanhart 1954a, 173.

[34]Montagu 1962, 922.

anthropologists to divide the European population into racial types as "dilettantish";[35] and in 1953 he stated simply that "there are no true races".[36]

Thus, it would seem that at least after 1945 Hanhart had abandoned the old racial categories and conceptions of "mainline eugenics", and up until this point, the history of Hanhart's project seems to fit nicely into the dominant historical narratives of human genetics, confirming the view that World War II marked a watershed moment separating racial hygiene from human genetics or mainline eugenics from reform eugenics. Historians of eugenics and human genetics have stated that in the 1940s, a shift took place, which they called the "medicalization" of eugenic research.[37] Accordingly, it was argued that the medical genetic view of specific disorders and diseases left little room for the broader social and racial concepts that were fundamental to mainline eugenics. However, a closer look at Hanhart's research practices reveals that the old narratives and categories proved to be astonishingly persistent in post-war medical genetics. Rather than undergoing a "medicalization", the genetic research carried out in isolates was instead shaped by an interconnection of medical, cultural, social, geographic and racial categories. This mechanism can be seen at work on a very basic level in the researchers' choice of a population to study. Which conceptual tools, discourses, and social contexts enabled geneticists to define certain human groups as isolated populations and to demarcate them from other human groups?

2 Constructing an Isolate: Inbreeding Discourses, Biohistorical Narratives and the Legacy of Racial Anthropology

In a paper on Leslie Dunn's population genetics study of the Jewish Community in Rome, the historian Veronika Lipphardt highlights a basic prerequisite for carrying out genetic fieldwork: identifying a suitable population.[38] In medical genetic studies of isolates, three factors were particularly relevant to this task. First, researchers had to find a community that was regarded as "isolated" or "endogamous"; this entailed identifying evidence that the group's members generally intermarried. Second, the geneticists were seeking social groups, villages or regions in which certain abnormalities or diseases were frequent; to do so they were dependent on information, narratives and hearsay. Finally, on a more practical level, researchers needed to work in communities where cooperation was probable;

[35]Hanhart 1954a, 171.

[36]Hanhart 1953, 545.

[37]For the Swiss context, see, for example, Ritter 2009.

[38]Lipphardt 2010.

as Susan Lindee reminds us, people, in contrast to laboratory animals, can be "informative" and "insightful" but also "unreliable" and "resistant".[39] Indeed, Hanhart's handwritten research records reveal that, not uncommonly, subjects refused medical examinations or blood tests.[40]

As all three of these requirements make clear, before geneticists could start their fieldwork, they were reliant on a complex, multifaceted body of knowledge in order to find and demarcate a suitable field site. One important source of information was medical studies of "inbreeding" and "incest", a research area that boomed from the mid-nineteenth to the beginning of the twentieth century.[41] Frequently, the social groups that geneticists in the 1950s defined as "isolated populations" were identical to those that had earlier been identified as "inbreeding populations". The scientific preoccupation with inbreeding was itself a response to a remarkable socio-demographic change. As historians of kinship have shown, a significant increase in close-kin marriages occurred in Europe between 1750 and 1850.[42] Because of fears that consanguineous marriages would result in serious health consequences and possibly even the degeneration of entire families, a large number of medical studies scrutinized the supposedly negative effects of inbreeding.[43] The evolving scientific and medical discourse on inbreeding was shaped by moral judgements as well as social and racial biases. Medical reports led to a perception of inbreeding and incest as health-damaging vices typical of rural communities and depraved working-class districts, and in the increasingly anti-Semitic atmosphere of the fin de siècle, inbreeding and incest were also obsessively ascribed to Jews.[44] In Switzerland, the medical discourse on inbreeding focused particularly on rural villages. Since the end of the eighteenth century, scholars, doctors and scientists travelling in alpine regions had reported on widespread abnormalities, birth defects, and diseases. These pathologies were attributed to, among other causes, the allegedly frequent practice of inbreeding.[45] Hanhart used these old reports as heuristic tools: they could shape new research questions and serve as signposts for identifying convenient field sites.

Thus, the nineteenth-century discourse on inbreeding not only reflected a middle- and upper-class concern about health but also evolved into a medium that both negotiated and produced otherness. Although the practice of marrying within a close kinship group was particularly prevalent among wealthy urban residents, inbreeding became increasingly identified with the lower classes and ethnic minorities, who collectively served as a foil upon which the upper classes could project

[39]Lindee 2005, 3.

[40]Archive MHIZ, Nachlass Ernst Hanhart, PN 56: 1:3, Bevölkerungsregister.

[41]Kuper 2002; Ottenheimer 1996.

[42]Sabean et al. 2007 and Sabean 1998. On Switzerland: Mathieu 2007.

[43]Kuper 2002 and Ottenheimer 1996.

[44]On class-specific perceptions, see Kerchner 2002. On racial interpretations, see Lipphardt 2008 and Gilman 1998.

[45]Germann 2007.

their desire for social distinction. Accompanying this social framing of inbreeding, an anachronistic perception prevailed. By neglecting the fact that marrying within the family was in many regards a modern phenomenon, inbreeding became regarded as a relic from ancient times still persisting in places far removed from the civilized world. Because of this spatio-temporal framing, small isolated populations came increasingly to attract the attention of anthropologists, and their publications provided yet another source of knowledge for genetic studies of isolates, as we will see in the following example.

One anthropologist who was convinced that isolated populations in the Swiss Alps could provide a useful window into the past was Rudolf Martin, a German scholar whose anthropometric methods would go on to wield enormous influence on the international development of physical anthropology.[46] In 1897, 2 years before Switzerland's first chair in anthropology was created for him at the University of Zurich, Martin programmatically demanded a systematic investigation of Switzerland's remote alpine villages, where "for a long time inbreeding became a necessity".[47] Envisaged as a contribution to racial anthropology, Martin's project aimed at detecting primal racial types which, he hoped, could be identified in these rural populations because they had seemingly remained stable for centuries, hardly affected by racial intermingling. So decades before Hanhart emphasized the ideal conditions in Switzerland for genetic studies of isolates, Martin made a similar claim, for identical reasons. Hardly any country, he wrote, was as suitable for racial anthropological research as Switzerland because of its manifold "virtually isolated areas".[48] Subsequently, a large number of racial anthropological surveys were conducted in villages and regions whose populations were considered to be isolated, immobile and homogenous.[49] Hanhart's studies of isolates drew on information from these racial surveys, but there is an even more direct link between them and his work. Tellingly, it was Otto Schlaginhaufen, a racial anthropologist and the successor to Martin's chair at the Anthropological Institute in Zurich, who initially gave Hanhart the idea for his ambitious project.[50] The institute's research focus had given Schlaginhaufen a detailed knowledge of the demography, anthropology and health of alpine populations, and he convinced Hanhart in 1921 to conduct large-scale genetic surveys in areas that were the same as, or similar to, the ones where anthropologists had earlier carried out their racial studies.

As this glimpse into the inbreeding discourses of the nineteenth and early twentieth centuries shows, the geneticists who scrutinized isolates after World War II did not tread upon a terra incognita. Rather, a myriad of studies, reports,

[46]On Rudolf Martin, see Germann 2015a and Morris-Reich 2013.

[47]Martin 1897, 32.

[48]Martin 1897, 34.

[49]See, for example, Wettstein 1902, Wettstein 1910, Bosshart 1938, Schlaginhaufen 1939, Hägler 1941, Büchi 1942 and Hess 1950. On the history of racial anthropology in Switzerland, see in particular Germann 2016, Germann 2015b, Schär 2015, 297–324, Keller 2006 and Keller 1995.

[50]Hanhart 1961, 54.

notes, data and rumours provided detailed demographic, medical and ethnographic information on these village communities, classifying and demarcating them from each other. Long before geneticists caught sight of any actual villagers, they encountered *representations* of those villagers, and in choosing a community as a study unit and moulding it into a research object of human genetics, the geneticists of the post-war era drew upon those representations. I will elaborate on this point in the following example.

Several villages Hanhart studied were known to be inhabited by the Walsers, a German-speaking minority in the East of Switzerland. These Walser communities attracted the interest of Hanhart and other geneticists, who were convinced that these Walser populations featured the ideal conditions for medical as well as population genetic research. In the 1950s, in particular, expeditions of geneticists scrutinized many Walser villages extensively, taking blood and determining the genetic and morphological traits of nearly all the inhabitants.[51] In these large-scale projects, scientists from a variety of disciplines and from several Swiss universities worked together, collaborating as well with some of the leading human geneticists and blood-group researchers from the USA and Britain. The blood sampled in Walser communities was tested not only in laboratories in Bern, Zurich, Basel and Geneva, but as well in the internationally renowned Blood Group Reference Laboratory in London.[52] Hence, as field sites for genetic studies, these small Walser villages loomed larger than any other communities in Switzerland. But how did they come to be chosen as study units by Hanhart and other geneticists?

One important factor was the sheer quantity of information and scientific knowledge available. Geneticists were not the first scientists to haunt these Walser settlements. Before them had come linguists examining the Walser dialect, folklorists studying Walser customs, historians investigating past Walser migrations and, last but not least, the aforementioned racial anthropologists, who measured bodies and examined the supposed racial peculiarities of this minority.[53] Consequently, the geneticists were able to refer to a recognized body of scientific work and, in particular, draw upon an established "biohistorical narrative".[54] According to this narrative, which was tinged with Darwinism, in the Middle Ages the Alemannic tribe of the Walsers migrated to the eastern Alps, where they adapted superbly to the harsh living conditions high up in the mountains and, increasingly, supplanted the indigenous population. Thanks, the story goes, to their clear demarcation from the Romansh-speaking population and their pronounced tendency to intermarry, the Walsers preserved their original culture, language and race.[55]

[51]See in particular Moor-Jankowski and Huser 1959, 1957; Ikin et al. 1957; Huser et al. 1956 and Moor-Jankowski, Huser 1954.

[52]Ikin et al. 1957.

[53]See the bibliography of "Walser studies", including all the mentioned disciplines Carlen 1973.

[54]On the term "biohistorical narrative", see Lipphardt and Niewöhner 2007.

[55]See, for example, Gysi 1951, Ruepp 1935, Wettstein 1910 and Wettstein 1902.

Although Hanhart and other geneticists relied upon this historical and racial narrative to give weight to the idea that Walser settlements were endogamous communities clearly delineated from their Romansh-speaking neighbours, the boundaries in practice were anything but clear-cut. The villages that were scrutinized proved to be less homogenous than assumed. In their investigations, for example, Hanhart and his co-workers came upon families who they identified as "vagrant people" ("wanderndes Volk") or "gipsies" ("Zigeuner").[56] These pejorative terms referred to a minority—nowadays known as the Yenish—against whom state authorities in Switzerland, in collaboration with private associations, pursued a policy of discrimination and forced assimilation. Between 1926 and 1976, children of Yenish families were systematically taken from their parents and placed with foster parents or institutions.[57] The geneticists treated Yenish families as an "exogamous" group and demarcated them from the village population, although the Yenish had been living in the village for several generations. In drawing boundaries between the Yenish and Walser villagers, genetic fieldworkers drew mainly on genealogical criteria, labelling a family as "vagrant" if their forbearers had been naturalized after the Swiss nation state's founding in 1848.[58]

By choosing to define "vagrant people" through their genealogy and ancestry, and not through the social or cultural group they might belong to, Hanhart tapped into notions of heredity and race, notions which fed on eugenic and psychiatric discourses and which greatly contributed to the stigmatization of the Yenish in the first half of the twentieth century. Medical studies concluded, on limited empirical grounds, that there was excessive inbreeding among Yenish populations, and psychiatrists pathologized their travelling lifestyle as a hereditary disease.[59] Hanhart adopted such interpretations. In an article published in 1951, he used the Yenish minority to illustrate the proposition that measures of assimilation and education were futile in the face of the "power of heredity". Just as it would be "grotesque" to aim at "washing negroes white", you could not, Hanhart argued, eradicate the "instinctive vagrancy" of the Yenish people through re-education.[60] Following the logic of this notion, Hanhart and his co-workers classified families as "vagrant" even when they had—by choice or through coercion—settled down long before. This example illustrates how the themes of traditional eugenics and racial biology found their way into the classifying practices of genetic studies of isolates.

In order to mould and define "endogamous communities", genetic fieldworkers resorted to pre-existing social, ethnic and racial demarcations. In so doing, they strengthened these demarcations while at the same time giving them a biological

[56] Archive MHIZ, Nachlass Ernst Hanhart, PN 56: 1:3, Bevölkerungsregister

[57] Galle 2016 and Meier 2008.

[58] Archive MHIZ, Nachlass Ernst Hanhart, PN 56: 1:3, Bevölkerungsregister. This genealogical evidence referred to the young nation state's policy towards stateless travellers of either deporting or forcibly naturalizing them. See Meier and Wolfensberger 1998, 511–517.

[59] Dazzi 2008, 73–76.

[60] Hanhart 1951, 3.

underpinning. Indeed, the notion that Walser populations were isolated ethnic or racial groups was seemingly confirmed by the investigations of blood-group geneticists, whose findings showed that Walser populations featured a significantly higher percentage of blood-group O than surrounding populations. In no other European population, the geneticists claimed, had such a high instance of blood-group 0 ever been found.[61] These results attracted the attention of, among others, members of Walser associations, and the high percentage of blood-group 0 became part of their discussions of group identity.[62] Thus, biological traits detected by geneticists found their way into discourses of Walser identity which, in turn, influenced the research conditions for geneticists, who often emphasized that Walser communities were cooperative, showing interest in the research. There were hardly any refusals, the fieldworkers noted, to allow blood to be taken or medical examinations to be performed.[63] This was yet another reason why Walser settlements were regarded as convenient study units.

So far, I have argued that "isolated communities" were not pre-existing entities. Despite the claims of geneticists, they did not exist in isolation for centuries due to reproductive barriers. Rather, they emerged from an interaction between recent socio-demographic developments and discursive inscriptions. As the example of the Walsers suggests, isolates were shaped by biohistorical narratives, anthropological classifications, social distinctions and identity discourses. Furthermore, for medical geneticists, finding and moulding an isolate was just the first step. In the next section, I will elaborate on how genetic fieldworkers detected pathologies and transformed them into hereditary diseases.

3 Detecting, Recording and Controlling Hereditary Diseases

In the main, Hanhart's research practices in alpine isolates consisted of gathering activities. In order to collect information on diseases, anomalies and family histories, he and his co-workers drew on a variety of sources. They visited local doctors, teachers, pastors and councils, from whom they received valuable information. Additionally, they gathered medical data in asylums and medical practices. And as genealogical data was especially important, they visited archives and studied church records, including comprehensive family registers.

Ultimately this social, historical, medical and genealogical knowledge was synthesized into large pedigrees. By situating diseases within complex patterns of kinship and depicting family narratives and the vertical transmission of genes in the same diagram, these pedigrees effectively translated social relationships into

[61] See studies in fn. 51.

[62] See, for example, Nachbaur 2013, 109.

[63] Liechti 1953, 153–154; Knoll and Arendt-Knoll 1950, 54.

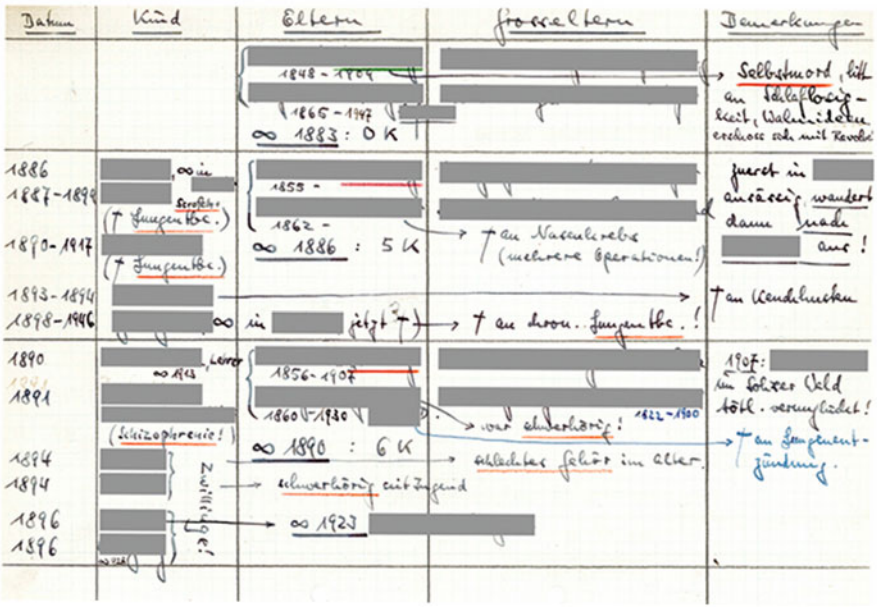

Fig. 1 Population register made in a Walser village in 1948 (MHIZ Archive, Hanhart papers, PN 56 1: 3: documents Paul Gysi)

biological ones, and vice versa, which explains why they played such a crucial role in the production of medical genetic knowledge. Historians have, with good reason, characterized such pedigrees as the "basic investigative"[64] or "bedrock"[65] tool of human genetics. But before pedigrees could be drawn, the information had first to be recorded in simple charts (Fig. 1) which were less organized and arranged. For our purposes, it is useful to note that because of their inherent "messiness", these first inscriptions can sometimes give valuable insight into the research process itself. Pedigrees tend to camouflage both the social practice of gathering—the fieldworker's agency—and the contingencies of research;[66] these simple charts, on the other hand, shed light on the process, which ultimately brought hereditary pathologies into being as epistemic objects.

The written record illustrated in Fig. 1 was made in a Walser village in 1948.[67] Initially, Hanhart had hoped to uncover cases of Pelger-Huët anomaly in this village, but not a single instance was found. However, the co-worker who performed the survey did not return empty-handed. As outlined in Hanhart's

[64]Nukaga 2002, 40.

[65]Lindee 2005, 62.

[66]Mary Bouquet has made a similar point when analysing the use of pedigrees in ethnographic studies. See Bouquet 1996, 60.

[67]Archive MHIZ, Nachlass Ernst Hanhart, PN 56: 1:3, Bevölkerungsregister.

research program for studying isolates, he systematically conducted medical examinations and compiled all available medical, demographic and genealogical data into a comprehensive population register, from which Fig. 1 depicts an extract. On the left side, genealogical relations are recorded. On the right, under the heading "remarks", we can see information on the cause of death, as well as on medical and social traits. At the top, on the right, the word "suicide" is underlined in red. Drawing on these data almost 20 years after the examination of the isolate, Hanhart published a study of the involvement of genetic factors in suicidality.[68] This example shows how data gathering in isolates could entail very different kinds of research. Basically, every recorded trait was a potential object of heredity study. Thus, these charts were the basic tools for constructing hereditary pathologies.

The charts' effectiveness was driven by two basic principles of data arrangement. The first organizing principle was genealogical—as we can see on Fig. 1, data is divided into three groups, representing three generations—and this vertical orientation is fundamental to assessing the frequency of pathologies in families; indeed, it is the underpinning of the very concept of a hereditary disease. This genealogical view of disease does not draw attention to individuals but rather focuses on what parents transmit to their offspring, essentially embedding hereditary disease in a narrative of provenance.[69] From this perspective, hereditary diseases are basically "family diseases"; moreover, these families can be located within broader kinship groups that are delineated by deploying social, geographic, ethnic or racial categories. Hanhart often attributed vague categories of ancestry to kinship groups by drawing on national (e.g. "Swiss families"[70]), racial (e.g. "Negroid", "Jewish", "Germanic" or "Romanic families"[71]) or social (e.g. "farmer families"[72]) frames.

These categories became even more important due to the second organizing principle, which essentially follows an epidemiological approach in arranging the gathered data. This may be surprising because medical Mendelism incorporates a concept of hereditary disease that rests on statistical patterns of vertical disease transmission and excludes horizontal considerations such as geographic and social distribution. However, as Jean-Paul Gaudillière and Ilona Loewy have convincingly argued, in practice notions of vertical and horizontal disease transmission often remained associated in twentieth century medicine.[73] Medical genetic studies of isolates are an example of this persisting association. Genetic fieldworkers were interested not only in genealogical patterns of human pathologies but also in their geographic distribution. When, for example, Hanhart summed up the results of his studies in the 1950s, he pointed out that the most striking finding of his research was

[68]Hanhart 1968/1969.

[69]Wailoo 2003, 249.

[70]Hanhart 1954a, 176.

[71]Hanhart 1954a, 173, 175, 178.

[72]Hanhart 1972, 228.

[73]Gaudillière and Löwy 2001.

the uneven geographic distribution of hereditary anomalies and diseases.[74] Some of the pathologies studied could only be found in a few small villages and nowhere else in Switzerland. In order to investigate such uneven distributions of hereditary diseases and traits, Hanhart announced a new field of research called "human genetic geography".[75] As a contribution to this field, he intended to prepare a "national topography of pathological mutations" that showed the geographic distribution of all known hereditary pathologies in Switzerland.[76] For reasons of health, he was never able to complete this project, but the endeavour was carried forward by younger researchers, among whom was David Klein. At an international symposium in Den Haag titled "Methodology of Isolates", Klein and his colleague Ferdinand Amman presented maps which drew on the data of Hanhart and other researchers to plot the geographic distribution of neurogenetic disorders in Switzerland. One of their findings concerned the distribution of cases of phenylketonuria (PKU), a congenital disorder of the metabolism. The two Geneva-based geneticists emphasized that in Swiss isolates several instances of PKU had been uncovered in families of "Gipsy origin".[77] The example shows how an epidemiological view of hereditary diseases could perpetuate racial notions.

In pursuing an epidemiological approach, Klein, Hanhart and other geneticists had to resort to a variety of pre-existing categories in order to detect and quantify uneven distributions of hereditary conditions. In dealing with regional differences between alpine villages, they frequently used geographical terms to capture the small-scale variations. As I have shown, however, local kinship groups and geographically defined isolates were also frequently placed within trans-local and transnational communities, and in order for the researchers to accomplish this, they had to deploy national, ethnic and racial categories.

In conclusion, I will end this section with the observation that these categories were not only present in the research process; they could be found as well in the practice of genetic counselling. As the most experienced researcher in alpine isolates, Hanhart was recognized as a medical expert in kin marriages and was frequently consulted by couples wishing to marry. According to a report published in 1972, Hanhart advised against 13 out of 30 planned first-cousin marriages.[78] How did he arrive at his conclusions?

As part of every consultation, Hanhart placed great emphasis on genealogical reconstructions, creating an extended pedigree that recorded the diseases and malformations of even distant relatives; but he drew as well on epidemiological considerations. In his report in 1972 Hanhart pointed out that "the risk of homozygosity for recessive disorders" also depended on "locality". This epidemiological

[74]Hanhart 1954a, 1955.

[75]Hanhart 1954a.

[76]Archive AIZ, Korrespondenz der Julius-Klaus Stiftung: Hanhart to Schlaginhaufen, 15 January 1963.

[77]Klein Ammann 1964, 128–129.

[78]Hanhart 1972.

perspective allowed him to factor in information on a couple's social, ethnic and racial identity. In one counselling session, for example, he advised against a marriage between first cousins even though there were only a few cases of diabetes and one suicide in their family. But Hanhart cited the family's Jewish and Eastern-European origins in his explanation and concluded that "close consanguine marriages were generally risky within this ethnic group" because they could feature "a high frequency of several recessive diseases".[79]

One of the most prevalent and tenacious allegations of scientific racism in the early twentieth century was that Jewish families from Eastern Europe were loaded with hereditary and infectious diseases.[80] In his assessment, Hanhart drew on such discourses of medical anti-Semitism; moreover, these racial notions were further perpetuated by Hanhart's conception of hereditary disease as being shaped by an interconnection of genealogical and epidemiological perspectives. Although he rejected the notion of "racial diseases", Hanhart deployed racial and ethnic categories in order to locate hereditary diseases within social groups and geographically defined populations. Hanhart's genetic counselling was based on the principles of voluntariness and self-responsibility; as such, his counselling practice seems to exemplify the "individualized" or "liberalized" eugenics of post-war human genetics. As this example shows, however, the discourses and categories of traditional eugenics could still persist within the medium of individual counselling.

4 Conclusion

Ernst Hanhart regarded the numerous remote villages of the Swiss Alps as "nature's laboratories". However, these communities were neither "natural" nor "laboratories". Rather, the geneticists encountered complex societies in which each community and family was interconnected with other social groups in manifold ways. No clear-cut boundaries separated the populations; in order to mould social communities into "nature's laboratories", genetic fieldworkers had to draw on pre-existing categories of national, ethnic and racial difference. These categories enabled the geneticists to reduce social complexity, define populations and naturalize cultural boundaries.

As a scientist who devoted himself to studying the medical and genetic differences between small alpine populations, Hanhart knew that broad racial classifications could never capture the complex diversity revealed in any close examination of the isolates, which is why he concluded that there were no "true races". Nevertheless, he deployed the concept of race as a tool to link hereditary diseases and "disease carriers" with ancestry groups and geographic areas. Thus, the use of race enabled genetic fieldworkers to combine a genealogical view focusing on

[79]Hanhart 1972, 241.

[80]See, for example, Weindling 1999 and Gilman 1998.

questions of ancestry with an epidemiological view aiming at the detection of geographically uneven distributions of diseases and traits. Rough racial categories made it possible to associate local entities such as families, villages or social groups with broader categories of difference that were solidified by established narratives, anthropological knowledge, identity discourses and state policies towards minorities.

In order to structure our understanding of the complex and entangled history of eugenics and human genetics in the twentieth century, historians have posited a number of conceptual distinctions, drawing a dividing line between mainline and reform eugenics, between eugenic hereditary research and medical genetics and between population and individual approaches to disease prevention. In the case study elaborated on in this paper, these boundaries are shown to be blurred. A program of eugenic population surveys—which fed on radical notions of racial hygiene and was admired by Nazi eugenicists—became smoothly recast as a genetic study of isolates, a booming field in post-war human genetics. To be sure, this transformation entailed changes in the eugenic framework. A close look at the collecting practices and the underlying categories involved in the research, however, reveals some striking continuities, particularly regarding the use of social and racial categories. Medical genetic fieldwork was shaped by an entanglement of medical, biological, social, historical and anthropological knowledge and practices. These manifold interconnections, this case study suggests, moulded the genetic fieldworkers' understanding of hereditary disease and influenced the practice of disease prevention. In the post-war era, old categories of difference were deployed and reified in order to demarcate the populations studied, arrange medical data and detect epidemiological patterns. In the process, eugenic notions of otherness, nineteenth-century discourses on inbreeding and racial anthropological classifications found their way into post-war medical genetics.

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Some Thoughts on Genetics and Politics. The Historical Misrepresentation of Scandinavian Eugenics and Sterilization

Nils Roll-Hansen

Abstract Human genetics has been—and still is—very much at the crossroads of science and politics. The confusion that comes from conflating science and politics is a chronic problem which is bound to need constant attention also in the future. The distinction between basic and applied research is both cogent and useful. It marks not a precise border, but rather a "trading zone" or overlap between science and politics, the social area where they meet and interact closely. In spite of all the criticism of this distinction during recent decades, I will use it to understand the history of sterilization and eugenics in a way that makes the autonomy of science defensible and important for a democratic political system. I will focus on the development of Scandinavian sterilization laws and practices and show genetic science was a main source of criticism and curtailment of eugenic policies.

Keywords Sterilization • Eugenics • Genetic science • Scandinavia • Twentieth century

Human genetics has been—and still is—very much at the crossroads of science and politics. It seems safe to predict that this situation will persist. Eugenics—policies aimed to improve the heredity of man—have a somber history. With the present rapid development of genetic technology and knowledge of human heredity, the problem of how to apply and how to regulate the new power that this knowledge provides is more pressing than ever. It is truly said that the only thing we can know for sure about history is that it never repeats itself. And nevertheless, it is a main source of experience for understanding the problems that face us.

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The historiography of eugenics has dwelt so much on the inhuman excesses of eugenic policies, especially in Nazi Germany, that it has obscured how similar present challenges and dilemmas are to those of the mid-twentieth century. As an insightful and politically engaged historian of eugenics put it a decade ago: "It is time to be more sophisticated in our accounts of eugenics, not just the sake of fidelity to the historical record but for a more adequate public policy."[1] In this study, I will focus on Scandinavia as a contrast to Germany, how eugenics developed under liberal democratic regimes as opposed to a totalitarian state. For the last half century, eugenics has been a paradigm case for sinister interaction of science with politics and Nazi Germany has been the paradigm example of eugenic ideology and its sociopolitical effects. This trend has fueled a biased interpretation of eugenics in Scandinavia missing the fundamentally different nature of political government, and its relation to science, in Nazi Germany and the Scandinavian countries at this time. It misses the difference between regimes where science becomes an instrument, ideological and technological, for the aims of the regime, and regimes where science also can criticize the political goals of the regime. The autonomy of science was a core principle in liberal defense against authoritarian and totalitarian politics, symbolized by Nazi Germany and Soviet Russia in particular.

In the late nineteenth and early twentieth century, there was widespread fear of degeneration due to modern conditions of living, industrialization, public health, and social care, etc. Eugenics arose as a movement to protect or improve biological inheritance. The term covers a wide range of different policies depending on what social ideals it included. Social historians and historians of science have often talked about eugenics as a "science." This can make sense in terms of an applied science, an application of human genetics organized to serve some more or less definite eugenic policy.[2] A problem with the historiographical use of such a broad concept of "science" is that it overlooks how important the difference between applied science and basic ("pure") science was for the historical actors, not least the scientists involved.

Sterilization has often been considered the most important expression of eugenics in the twentieth century, with the 1933 Nazi German sterilization law as the frightening example. The tendency to identify sterilization with eugenics has contributed to a biased understanding of Scandinavian eugenics. However, sterilization can be carried out for quite different reasons. Roughly, they can be characterized as biological and social. Biological motivation can be concern for unwanted detrimental heredity in immediate progeny or concern for such heredity in the population. The latter is eugenics in the strict sense, while the former belongs to medical genetics, as usually understood.[3] Important examples of social motivation

[1] Paul 2007, 15.

[2] My concept of applied science points to the political framing of the research and knowledge in a way similar to Ilkka Niiniluoto's concept of design science.—Niiniluoto 1993, 2014.

[3] Of course, individual interventions of medical genetics are likely in the long run to have substantial effects on the gene pool. In the broader discussion of eugenics as human control of the evolution of the species, this is an important issue.

are women's liberation and family planning. I will argue that such social motivations together with concern for immediate progeny were dominant in the lawmaking and practice of sterilization in Scandinavia.

1 The New Science of Heredity: Classical Genetics

While eugenics came of age in the late nineteenth century, the modern science of genetics was only established around 1910. The dominant paradigm of classical genetics was the chromosome theory presented by Thomas Hunt Morgan (1866–1945) and his students in *The Mechanism of Mendelian Heredity* (1915). By then, the fundamental distinction between genotype and phenotype including the stability and discontinuous change in genotype and its components the genes had been established. Hereditary change was not continuous, as orthodox Darwinians like the biometricians believed. It was stepwise through mutation and recombination of genes.[4]

This time lag meant that early eugenics was based on views of biological heredity that soon became outdated. Darwin's idea of small "gemmules" as material carriers of heredity was not much different from the speculations of Antiquity, more than 2000 years earlier. Only with the development of cytology and systematic application of scientific methods to plant and animal breeding toward the end of the nineteenth century was the ground prepared for a modern science of biological inheritance. A substantial part of the pioneering work was done in Scandinavia, especially in the Øresund region including Copenhagen in Denmark and Lund in Sweden.

The early eugenic programs and policies were thus largely based on vague folk ideas about biological heredity. This was the case for instance with the first sterilization laws introduced and practiced in the USA, from 1907 on. Only gradually did the eugenics movement absorb the new knowledge and insights of classical genetics. By the 1930s, representative eugenic organizations recognized that credible eugenic policies had to be abreast of the science of genetics. The terms "mainline eugenics" and "reform eugenics" have been used to designate these two phases of eugenics.[5]

The study of human genetics was seriously tainted and held back by its close association with the race and eugenic policies of Nazi Germany. In his 1961 presidential address to the American Society of Human Heredity, Leslie Dunn (1893–1974) emphasized that the study of human genetics had long lagged behind the general development of genetics. Primitive ideas of unit characters and simple Mendelian inheritance of psychic characteristics had long continued to influence psychiatry and eugenic thinking about feeblemindedness and mental disease. I will

[4]We now know that this was a somewhat simplistic model, but it provided the hard core of a very progressive research program in genetics through the middle of the twentieth century.

[5]Kevles 1985.

return to this in my account of sterilization and eugenics in Scandinavia. Another indication of this backwardness is the fact that only as late as 1956 was the number of human chromosomes precisely and correctly counted, at Lund University.[6] That same year the first International Congress of Human genetics was organized by Tage Kemp (1896–1964) in Copenhagen.

Dunn was concerned about the recurrent tendencies to conflate science and politics, i.e., not to be sufficiently clear about the difference between scientific knowledge and the political programs that applied it. Eugenics had "appeal to most people as embodying a noble conception,"[7] but the political enthusiasm stimulated wishful thinking and neglect of sober scientific method. The development in Germany after 1933 was a grave example. Even serious scientists had underrated the dangers of the eugenics movement until it was too late. The problem lay not only with those who supported the misuse of genetics, but also with those who failed "to point out, as teachers, the distinctions between true and false science," said Dunn.[8]

Dunn referred to William Bateson (1861–1926) as one of the founders of genetics who worried about conflation of science and politics. In 1919, Bateson insisted on the importance of the distinction between basic and applied science:

> The eugenicist and the geneticist will, I am convinced, work most effectively without organic connection, and though we have much in common, we should not be brigaded together. Geneticists are not concerned with the betterment of the human race but with problems of pure physiology, and I am a little afraid that the distinctness of our aims may be obscured.[9]

2 Eugenics in Scandinavia

Eugenic ideas were popular in Scandinavia during the first half of the twentieth century, but declined sharply from around 1950. Concern for the quality of biological inheritance to new generations was influential in many areas of social policy. This was part of a widespread concern for degeneration of populations in Europe from the mid-nineteenth century on. Marriage laws, institutions and laws for taking care of insane and mentally retarded, immigration laws and regulations, etc., were influenced by eugenic ideas. But by far the most attention has been given to the development of sterilization laws and practices.

The ideology of eugenics came to Scandinavia around the turn of the nineteenth century, primarily from Germany. It was inspired by German scientists and scholars like Alfred Ploetz (1860–1940) and Wilhelm Schallmeyer (1857–1919). The German term "racehygiene" ("Rassenhygiene") was at first most commonly used in

[6] Harper 2006.

[7] Dunn 1962, 3.

[8] Dunn 1962, 8.

[9] Quoted from Dunn 1962, 4.

Scandinavia. As racism became more unpopular in the years after World War I, the English term "eugenics," originally introduced by Francis Galton (1822–1911), was gradually preferred.

A Swedish Society for race hygiene was founded in 1909 as a branch of the German-based International society for race hygiene.[10] The Danish movement was headed by an Anthropological Committee where most members were medical doctors.[11] In Norway, the pharmacist Jon Alfred Mjøen started propagating his "Norwegian program for race hygiene" in 1908. He also became a central figure in the international eugenics movement.[12] Eugenics made an impact in Finland from the 1910s on. *Samfundet Folkhälsan i Svenska Finland* (The society for public health in Swedish Finland) became a driving force. Finland was an ethnically divided country with a culturally and politically dominant Swedish-speaking minority. It was not by accident that eugenics found its early supporters within this minority.[13]

This early period in Scandinavia was dominated by mainline eugenics. The supporters had their scientific basis as much in the humanities as in natural sciences. Some of the most active critics belonged to the pioneers of the new discipline of genetics. In Norway, this led to a polarized debate with harsh denunciation of Jon Alfred Mjøen (1860–1939). He was ostracized from the club of professional geneticist, and his own "Consultative Norwegian committee for race hygiene" mostly consisted of humanists, lawyers, and politicians. The initial attack was led by the young medical scientist Otto Lous Mohr (1886–1967) who later became an internationally influential human geneticist. At first, Mjøen had a considerable influence on public opinion and politics, but by the mid-1930s when the sterilization law was formulated and approved the geneticists had the decisive word.[14]

Scientific institutions for the study of biological heredity were founded in the same period. The practical motivations for developing a science of heredity had two sources. One was the demand for better varieties of plants and animals in a quickly modernizing agriculture, adapting to national and international specialization and trade. The other was concerns for eugenic or dysgenic consequences of social and population policy. Great scientific advances had been made with discoveries in cytology, microbiology, physiology and evolution through the nineteenth century. Genetics was emerging as a fundamental discipline of biology that promised to unify all these new branches of scientific knowledge. In the founding period around the turn of the nineteenth century, new theoretical impulses came from plant breeding in particular. Plants were well suited to clarify the existence of the genotype and its interaction with the environment in the formation of an organism. And such clarification was of practical use.

[10]Broberg and Tydén 1995, 68.

[11]Hansen 1995; Koch 1996.

[12]Roll-Hansen 1995, 155–161.

[13]Hietala 1995.

[14]Roll-Hansen 1980.

A pioneering institution of worldwide importance was the plant breeding institution at Svalöf in South Sweden, founded in 1886 and developed in close cooperation with genetics research at the nearby Lund University. A genetics institute was established at the University of Oslo in 1916 focusing on questions of human heredity. A State Institute for Race Biology was established at Uppsala in Sweden in 1921 under the leadership of Herman Lundborg (1868–1943), an active participant in international eugenics organizations. The Rockefeller Foundation gave extensive support to the development of research on human genetics in Denmark. An "Institute for human heredity and eugenics" was established at the University of Copenhagen in 1938.[15] Throughout this period, the Scandinavian countries were at the forefront in genetic research.

Neither "eugenics" nor "race hygiene" is necessarily racist in the sense of holding some races to be superior to others. But the movement was from the start highly discriminatory of non-European races. Before World War I, views that we today consider outrageously racist were generally accepted without wincing an eyebrow. For instance, the mild-mannered and benevolent founder of Norwegian psychiatry, Ragnar Vogt (1870–1943), in his book *Racehygiene* (1914) wrote: "It is not seeming for a blond blue-eyed, intelligent Nordic to degrade his hereditary material by marrying a negro."[16] This should be understood more as blindness to general contemporary prejudice than as malevolent discrimination of blacks. Sensitivity to racist rhetoric changed rapidly through World War I and its aftermath.

3 The Introduction of Sterilization Laws in Scandinavia

In the USA, sterilization laws were introduced in the early decades of the twentieth century from the 1910s to the 1920s, when mainline eugenics still dominated. The Scandinavian laws were introduced mainly in the mid-1930s when reform eugenics was taking over. In Denmark, the first law regulating sterilization of feebleminded was introduced in 1929, and two new laws with broader application in 1934 and 1935. In Norway, a sterilization law was introduced in 1934, and in Finland in 1935. Sweden introduced a sterilization law in 1935 and revised it in 1941 allowing a more coercive sterilization practice.

These laws emphasized that for ordinary citizens of legal age sterilization is a matter of own free choice. The problem of compulsory sterilization arose with underage and otherwise legally incompetent persons. Parents or other guardians were then responsible. These were paternalistic societies and individual freedom did not have as wide scope as today. A clear sign is that even normal legally competent adults had to apply for permission to have a sterilization, and originally there were strict restrictions on such permission. The Norwegian law was seen as

[15]Koch 1996.

[16]Quoted from Roll-Hansen 1980, 278.

particularly progressive by explicitly stating the possibility of having a sterilization permitted for "respectable reasons" (aktverdig grunn).

Genetic research had by the 1930s considerably narrowed the options for proposal on eugenic policy. The understanding of human heredity that gradually developed showed that there was little if any immediate threat of hereditary degeneration. Furthermore, the hereditary mechanisms responsible for physical diseases and mental retardation were so differentiated and complex that a negative eugenics was unlikely to be successful. Already in the 1910s, it was well understood by geneticists that for a hereditary disease depending on one recessive gene those individuals showing the disease represented only a small fraction of the total amount of such genes in the population. The effect of a negative eugenic policy discouraging or preventing these people from having children would be correspondingly small.

The Scandinavian sterilization laws of the 1930s were introduced with a high degree of political consensus. These were morally sensitive questions, and in a liberal democratic system a careful conservative approach was necessary. The laws had to be moderate compromises that accommodated traditional moral restrictive attitudes. Eugenic enthusiasts like Mjøen and Lundborg saw these laws as a first step in the direction of truly eugenic policies. It is significant that in other West European countries proposals for similar laws were abandoned. In England and the Netherlands, for instance, the opposition was too strong. The Catholic Church argued on moral grounds. Representatives of labor and socialism argued that such laws would discriminate suppressed social groups. The Scandinavian countries were Lutheran and relatively secular. They also had a more homogeneous population and small class differences, as well as a weak Catholic Church.

A truly eugenic justification, aiming to improve the population's gene pool, was no doubt an important factor in the introduction of the laws and significant for their practice during the first couple of decades. This was a heritage from nineteenth-century social-biological ideology ("social Darwinism"). But already in the debates leading up to the laws social justifications took on central importance. This is reflected in the formulation of the laws. They emphasized the welfare of women and children as main reasons for permitting sterilization. On the biological side, the likelihood that hereditary disease or disability could be inherited was also included as a main justification. But this was more in tune with the individually oriented medical genetics of the period after the Second World War than eugenics in a traditional sense.

It is notable that the moderate formulation of the Scandinavian sterilization laws was able to accommodate the radical changes in sterilization practice during the decades following the Second World War. These laws mostly stayed in force till the 1970s. By then sterilization had become a widespread means of contraception not only in Scandinavia.[17] A law demanding that ordinary people legally of age had official permission to be sterilized was obviously outdated.

[17]It has been estimated that in 1988 near half of US women of age 35–44 had either let themselves be sterilized or had a partner who had done so. See Tydén 2002.

A strong and consistent critic of eugenic sterilization was Otto Lous Mohr, medical doctor and professor of anatomy at the University of Oslo. He was also an internationally prominent geneticist, an authority on human genetics, and played an important role in the international politics of genetics in the 1930s and 1940s.

In his capacity as dean of the medical faculty of the University of Oslo, Mohr wrote an assessment of the government proposal for a sterilization law. Considering the present knowledge of human genetics, it was unlikely that the sterilizations approved by the law would have any positive eugenic effects, claimed Mohr. But he supported the law because a legalization of sterilization on social indications was important for family planning. Mohr was married to a medical doctor who was very active in this field. And his mother-in-law, Katti Anker Møller (1868–1045), started the birth control movement in Norway. There was a deluge of protest, even from some of her collaborators in the feminist movement, after her public lecture in 1914 on "The liberation of motherhood," arguing that abortion should be legalized. Mohr himself was a supporter of the Labour party but not a member. Mohr's combination of advanced genetic expertise with clear political motivation was the basis for his large influence on the eugenics debate in Norway.[18]

A Swedish counterpart to Mohr was Gunnar Dahlberg (1893–1956). When Lundborg retired as head of the State Institute for Race Biology, there was a long-drawn conflict over his successor. With support from his friends among the governing social democrats, in particular Gunnar Myrdal (1896–1987), Dahlberg finally was appointed in 1936.[19] This marked the transition from mainline to reform eugenics in Sweden. Dahlberg was a leading international expert in human genetics with good contacts with leading British human geneticists. The British medical statistician and zoologist Lancelot Hogben (1895–1975) gave a lecture in Oslo on the evening of April 8, 1940, and fled to Sweden when the Germans invaded the following day. In Sweden, he spent time with his friend Dahlberg before he was able to get back to England and started on an English translation of Dahlberg's 1940 book, *Arv och Ras* (heredity and race). The translation was published as *Race, Reason and Rubbish* in England in 1942. After the Second World War, Dahlberg was active in UNESCO's campaigns against racism.

Dahlberg and Mohr, like many of their liberal and left-wing British and American geneticist colleagues, sharply criticized mainline eugenics and were against the introduction of eugenic policies in the present situation. The scientific knowledge was too weak and the actual proposals in their judgment were likely to have social effects contrary to the general programs of improving the living conditions of women and other suppressed groups. But they saw human genetics as a science that was likely to lead to future results of great importance for human health including improved quality of the population's gene pool.

[18]More information about Mohr in Roll-Hansen 1980.

[19]Broberg and Tydén 1995, 91–95.

4 Eugenic Sterilization in Scandinavia?

Did Scandinavian countries introduce specific laws and other means to implement government-driven eugenic policies, like Germany after the Nazi takeover of 1933?

Marriage restrictions were an old tradition, and new marriage laws in the early twentieth century were influenced by the new knowledge of genetics. Immigration policies, e.g., with respect to Jews fleeing Germany after 1933, were clearly influenced by racist mainline eugenic ideas. But was there systematic government policy introducing practicing eugenic and racist laws like the German sterilization law of 1933 and the Nuremberg race laws of 1935? The sterilization laws are the obvious point of comparison since there were no laws parallel to the Nuremberg laws directed against full citizens' rights for Jews, including ban on marriage or sexual relationship with persons of "German or related blood."

The cultural closeness of Scandinavia to Germany, with a common religious basis in Lutheran Christianity, a similar type of centralized government run by civil servants with a high degree of trust in the population, and a similar development of social institutions and ideology up to the 1930s, makes for interesting comparison. This was underlined in the first overview of Scandinavian sterilization, *Eugenics and the Welfare State*, published in 1995, two years before the sensationalist mass media stories of August and September 1997. The final chapter of the book pointed to the political break of 1933 as a decisive moment of change. It was this political break "rather than a fundamentally different German social and cultural development, a German 'Sonderweg', that led to the realization of Nazi eugenic politics." Thus, "a comparison of eugenics in Scandinavia and Germany appears particularly well suited for a sophisticated analysis of the interaction between science, ideology and politics," concluded the book.[20]

The contribution on eugenics in Norway suggested that a misleading instrumental view of science deriving from a German philosophical tradition and popularized through the Frankfurt School of Sociology in the 1960s and 1970s had distorted the historiography of eugenics. This view, which became dominant in the sociology of scientific knowledge from the 1970s on, saw science as an instrument of politics and not as an autonomous source of insight and knowledge that politics disregarded at its own risk. It disregarded the difference between applied science (technology) and basic science with respect to governing values and potential for criticism of current politics. It was argued that such tendencies are found for instance in the overview of German eugenics, *Rasse, Blut und Gene* (1988) by Peter Weingart et al.[21]

In a 1999 paper, Weingart took up the challenge of comparing Scandinavian and German eugenics in order to illuminate the general processes of interaction between science and politics. He took Sweden as his case, and his analysis built on the philosophical foundation that Roll-Hansen had criticized as inadequate for a

[20]Broberg and Roll-Hansen 1995, 267–270.

[21]Roll-Hansen 1995, 151–155.

dependable historiography. Weingart concluded in harmony with the 1997 mass media rumors that there was "virtual identity of eugenic and rase hygiene discourse in Sweden and Germany as well as a striking similarity in the sterilization practice."[22] This may be true up to 1933, but not after the Nazi regime took over in Germany.

Weingart's use of statistics about sterilizations is revealing. It has been estimated that out of ca. 350,000 sterilizations under the Nazi law of 1933 ca. 300,000 were carried out before the start of the war in August 1939.[23] Weingart compares the 350,000 German Nazi sterilizations in the period from 1934 to 1945 with the 63,000 sterilizations carried out under the Swedish law in the period from 1935 to 1975 and calculates that proportionally this amounts to 0.4% of the total population in Germany and 0.8% in Sweden. He does not mention that in the period before the start of the Second World War the number of Swedish sterilizations was ca. 2800 compared to 300,000 in Germany which gives proportions of respectively 0.035% and 0.35%. The ten times higher rate in Germany demonstrates the brutal and rushed character of the Nazi sterilization program, a clear indication that the eugenic rhetoric was in fact different.

Weingart's analysis also disregards principal differences between Swedish and Nazi laws and practices. The Nazi law institutes compulsory sterilization and even allowed for physical force to carry out decisions by the specially established "Erbgesundheitsgerichte" (courts of hereditary health), while the Swedish law assumed a principle of voluntariness. Furthermore, the German Nazi law was biological. It accepted only biological hereditary reasons for sterilization and not social justifications. In Scandinavia, social justification was important from the beginning and soon became dominant. The mentally retarded was a target group in Sweden as in Germany. In the period up to the late 1940s, mentally retarded sterilized on so-called "eugenic indication"[24] was the largest group in the Swedish statistics. But as shown above their inability to bring up and support children was a main consideration in Sweden as well as in other Scandinavian countries. And from ca. 1950, the sterilization of mentally retarded decreased rapidly.

Weingart went on to argue that the similarity in eugenic sterilization practice is due to ideological similarity between German national socialists and Swedish social democrats. It supported an "autocratic philosophy" that subordinated "the rights of the individual" to "the interests of society." This, according to Weingart, might explain "how thin the dividing line between Social Democratic and National Socialist sentiments actually was with respect to the role of the state and eugenic practice."[25] It is curious how a prominent and trendsetting scholar in science studies

[22]Weingart 1999, 173.

[23]Bock 1986.

[24]This expression simply referred to biological heredity. It did not distinguish considerations for immediate offspring from eugenics in the strict sense, i.e., improvement of the gene pool of the population.

[25]Weingart 1999, 167.

plays down the central political contradiction of the mid-twentieth century. Social democrats were in front defending liberal democracy first against Nazi and then against communist dictatorship.

This ideological comparison of Scandinavian social democracy to German National socialism has later been picked up and developed by other scholars. The Israeli political scientist Alberto Spektorokowski has argued that a "progressivist and humanist type of socialist welfare loses moral ground when it adopts productivity views of society."[26] He compares the expression of this tendency in Soviet Russia, Nazi Germany and social democratic Sweden. In Sweden and Germany, the imposition of productivity as a fundamental aim led to a eugenic social policy. Soviet Russia was saved from eugenics by dialectical materialist ideology that outlawed the biological view of man implied by eugenics—at considerable cost for the science of genetics. Swedish social democracy was saved from the drift toward authoritarian political solutions by democratic control.[27] Spektorokowski does not, however, compare socialist to capitalist influence on social policy. One might argue that the latter was no less important than the former in promoting eugenics in the Swedish welfare state.

The criticism of the Swedish welfare state as a creation of social democratic politics tainted by eugenics, which Zaremba launched in 1997, was a main theme in an extensive 2003 review of Scandinavian scholarly literature on the history of eugenics by Thomas Etzemüller. He concluded that the political differences had little significance for the nature of eugenics in Germany and Scandinavia. Dictatorship only meant more radical eugenic measures. "German eugenics was the model. It was not discredited even by national socialism," was Etzemüller's verdict.[28]

The role of "modern welfare policies" as a driving force for eugenic measures in Scandinavia is also emphasized by Véronique Mottier in her comparison of government policy in Switzerland, Sweden and Germany. She points to social democratic promotion of collective goods at the expense of individual freedom as the fatal ideological difference to England and the USA—countries that avoided similar sterilization regimes. "Far from constituting an 'accident' in the history of social-democracy, the eugenic social experiments fit comfortably with core elements of social democratic ideology," claimed Mottier.[29]

Common to these accounts of how the ideology of the Scandinavian welfare state created pernicious eugenic policies is a lack of detailed knowledge about the historical events. The sweeping interpretation is based on a selective and limited set of facts.

Kris i befolkningsfrågan (Crisis in the population question) by Gunnar and Alva Myrdal (1902–1986), first published in 1934, is recognized as a representative and

[26]Spektorokowski 2004, 85.

[27]Spektorokowski and Mizrachi 2004, 352.

[28]Etzemüller 2003, 508.

[29]Mottier 2010, 142.

highly influential expression of the ideas that formed Swedish social democracy in the 1930s and 1940s. An English version by Alva Myrdal bears the more enlightening title *Nation and Family. The Swedish Experiment in Democratic Family and Population Policy*.[30] The original Swedish as well as the English version includes eugenic considerations in their discussion of sterilization. This discussion covers only a small fraction of the book. Its concern was family policy in a broad sense, covering issues from illegitimate births, contraception and sexual instruction to housing, home economics, education and recreation for the family. In the English version, sterilization was part of a chapter on "Planning the Size of the family."[31] The uncertainty of genetic knowledge and respect for voluntariness is stressed, as well as the importance of allowing purely social indications. The quotations and paraphrases that are used to document the eugenic character of the Swedish welfare state are taken out of context. They give a distorted and biased impression of the social policy program of Alva and Gunnar Myrdal. Their main message was neither eugenics nor economic efficiency but an egalitarian social policy to improve the living conditions of the lower classes.

Since 1997, a series of detailed studies have greatly improved our knowledge of how the sterilization laws were implemented. The Norwegian sterilization law, its introduction and changing practice, has been thoroughly studied by Per Haave. He has documented how eugenic arguments gained increased influence in the late 1930s and 1940s, in particular with respect to mental retardation. But even then it appears that social justification was a more important motive than eugenics in the strict sense of population genetics: The mentally retarded lacked the ability to bring up and support children.[32]

A similar extensive and conscientious study of the Swedish case by Mattias Tydén indicates that strictly eugenic justifications had more impact in Sweden than in Norway. However, it was not a government policy imposed from above, but rather a result of local grass-roots attitudes among the doctors and local health and social institutions and services that handled the individual applications for sterilization.[33]

For Denmark, Lene Koch has made a detailed and penetrating analysis of justification of sterilizations according to the 1934 law about care of mentally retarded. The text of the laws does not mention biological heredity and eugenic considerations. But in the individual applications arguments from unwanted hereditary effects are common. Koch and her assistants classified 5579 registered sterilizations under this law in the period 1934–1968 into three categories according to the indication for sterilization: "partially eugenic," "eugenic," and "non-

[30]Myrdal 1945. This English version is substantially expanded from the first Swedish 1934 version. The preface is dated Stockholm, August 31, 1940. Apparently the book was written then but not published till after the Second World War.

[31]Myrdal 1945, 212–217.

[32]Haave 2000, 212–215.

[33]Tydén 2002, 588–590.

eugenic"—where "eugenic" means justified by biological inheritance. The total number of sterilizations peaked at around 300 per year in 1937 to 1951 and then fell continuously to below 20 in 1968. Only a few percent of the cases were purely "eugenic." The relative frequency of the two other categories changed substantially through the period. The non-eugenic made up ca. 60% in the early years 1934–1943, fell to around 50% in the period 1945–1961, and then rose to ca. 70% in the period 1963–1967.[34] This course of development confirms the importance of social (non-eugenic) justification in the 1930s, the increasing weight of eugenic (hereditary) justification in the 1940s, and its decline from around 1960.

These detailed analyses of sterilization policy and practice in Norway Sweden and Denmark support the view that Scandinavian sterilization was motivated more by social considerations, improving the conditions of children in poor families, emancipation of women, etc., than by eugenic arguments to improve the quality of the gene pool. So far these Scandinavian studies have received little attention in the international scholarly debate. Weingart's 1999 comparison of Germany and Sweden has been widely accepted with little attention to the criticism.[35]

5 Persistence of Eugenic Sterilization

As already mentioned, the knowledge of human genetics was still weakly developed in the 1940s and 1950s lagging behind that of plants and animals. The lack of knowledge gave considerable scope for eugenics in spite of the criticism from liberal left-leaning geneticists like Mohr and Dahlberg. There was much uncertainty about interaction of recessive and dominant genes as well as the importance of environmental factors. This was crucial for instance with respect to the explanation of mental retardation, which was a main concern of eugenic policy.

Nils von Hofsten (1881–1967), professor of zoology at Uppsala University, was main advisor on sterilization for the Swedish government from the 1930s till the 1950s. He had published an influential textbook on genetics in 1919. In 1944, he published a paper on "(t)he hereditary effects of sterilization" which discussed the possible eugenic effects of Swedish sterilization practice. Von Hofsten argued that the new law of 1941 had the effect that was hoped for. The number of sterilizations of feebleminded had increased, and this was likely to have a significant effect on their frequency in the future.

In his calculations, von Hofsten assumed that 75% of feeblemindedness is hereditary and can be accounted for by classical Mendelian inheritance. This high degree of genetic determination was a common assumption among psychiatrists at

[34]Koch 2000, see especially Tables 3, 14, and 17 toward the end of the book (no pagination).

[35]Besides the literature already cited, there is a vigorous counter-story by Siri Haavie (2003) who emphasizes the humanitarian and social motives of sterilization. A more subdued criticism is found in Tydén 2010.

the time. Hofsten recognized that hereditary feeblemindedness is "not a biological unity," but assumed that it depended on an interaction of several recessive and dominant factors. He argued that such a complex mechanism could well be equivalent to a model depending on a single recessive gene. With random mating and 100% negative selection, this would give a reduction from 1 to 0.7% feebleminded in two generations. For Sweden, this would mean 19,000 fewer feebleminded. Even if this was an idealized calculation, it indicated a eugenic effect that was by no means insignificant, claimed von Hofsten in 1944.

As late as 1955, Karl Evang (1902–1981), the powerful head of the Norwegian health service from 1939 to 1972—with a five-year pause during the German occupation 1940–1945, argued that the sterilization of feebleminded ought to be intensified for eugenic reasons. This does not imply that he saw eugenics as the main reason for sterilization of the feebleminded, but it does show that eugenic arguments were still acceptable and influential. For genetic expertise, Evang relied on von Hofsten.[36]

Tage Kemp, head of the Institute for human genetics and eugenics at the University of Copenhagen, argued for a similar conclusion in his 1957 Galton lecture in London. He estimated that in Denmark selection against feeblemindedness could reach 50% for the period 1951–1970 if current practice was continued. But he cautioned that the effect of such negative selection "will depend on the way mental defectiveness is inherited." Kemp still assumed high heritability which he called "empirical genetic prognosis": with two mentally defect parents, 60–90% of the children would inherit the condition, with one such parent 30%.[37]

Hereditarian views continued to flourish among Scandinavian psychiatrists from the 1930s into the period after World War II. In this respect, they were similar to German and different from Anglo-American psychiatrists at the time. An official British report on mental retardation in 1933 (Brock 1934) presented German figures similar to those of von Hofsten and Kemp, but its own conclusions gave biological heredity less weight.[38] British research on human genetics starting in the 1930s contributed greatly to more precise understanding of the multiple genetic and not least environmental causes of mental retardation. The impact of Lionel Penrose's revealing studies of the causes of mental retardation was first felt in his home country Britain and in the USA. His results were synthesized in a classic 1949 monograph, *The Biology of Mental Defect*. Penrose was a pioneer demonstrating through clinical research both the diversity of the genetic causes and how important the environment was in molding different results within each genetic category.[39]

But it took time before the new insight into the underlying biological mechanisms, genetic as well as environmental, was digested and empirically assessed and integrated in current psychiatry. The director of the national Norwegian Psychiatric

[36]Roll-Hansen 2007, 71–73.

[37]Kemp 1957.

[38]Brock 1934.

[39]For Penrose's contributions, see Kevles 1985.

Hospital, Ørnulv Ødegård (1901–1983), was typical of psychiatrists' conservatism. In the late 1950s, he served as genetic expert for a commission to revise the Norwegian Marriage Act of 1918. An appendix to the commission's report, written by Ødegård, said that disappointing result of therapy in the early part of the century had led to exaggerated belief in heredity, but at present the tendency was "to underestimate what can be achieved by eugenic measures." His expert genetic authority was von Hofsten. The Norwegian marriage act that was finally passed in 1969 simply retained the 1918 clause that the "insane" and the "feebleminded" were not allowed to marry. There was no mention of eugenics.[40]

By this time, the earlier acceptance of strong inheritance in mental disease had been effectively undermined. The authoritative twin studies of Franz Kallmann (1897–1956), a German psychiatrist who emigrated to the USA in the 1930s due to his Jewish ancestry, published in the 1940s and 1950s, had apparently shown a high degree of inheritance of schizophrenia. The concordance numbers[41] were 86% for monozygotic and only 15% for dizygotic twins.[42] Though the results of other investigators indicated a smaller difference, Kallmann was widely accepted till the 1960s when Scandinavian researchers with improved methods of sampling confirmed that Kallmann was wrong. A Finnish study surprisingly showed no concordance for all of 16 pairs of monozygotic twins. And a large Norwegian study soon confirmed much lower concordance than Kallmann for both monozygotic and dizygotic twins, namely 28–38% concordance for monozygotic and 5–14% for dizygotic, which remain accepted figures until today.[43]

Already in the 1970s, the Norwegian government started a new revision of the marriage act using other genetic experts. Following their advice, the revision that was finally introduced in 1991 had no prohibition of marriage for mentally retarded or ill.[44]

6 Human Genetics Between Science and Politics

The historiography of eugenics has been important in forming present views on the relationship between science and politics. Because of the close and complex entanglement of science and politics, the history of eugenics is a particularly challenging topic. Historical analysis has been much occupied with threats implicit in general features of Modernity. Zygmunt Bauman's *Modernity and the Holocaust* (1989) is a characteristic expression of the worries that developed in the last

[40]Giæver 2003, 9–13.

[41]The concordance number is the percentage of twin pairs who share a certain property, e.g., schizophrenia.

[42]Kallmann 1946, 1953.

[43]Tienari 1963; Kringlen 1964.

[44]Giæver 2005.

decades of the twentieth century. A more recent study addresses the utopian scientism characteristic of much eugenic thinking up to 1940.[45]

Pessimistic views of modern science and its enlightenment ideal were well developed before the Second World War, not least in Germany. The neo-Marxist Frankfurt School of Social Research was a characteristic example. Their writings became an important source of theorizing about science and technology in the second half of the twentieth century. The School emigrated to the USA in the 1930s due to Nazi persecution and its ideology inspired radical student movements of the late 1960s in both the USA and Europe.

The word "eugenics" today invokes a powerful myth about the fatal union between an amoral science and an evil political ideology. But it is important to remember that this was not always so. Before the Second World War, few objected to the idea that an improvement of human biological heredity would be a good thing, assuming this could be realized with acceptable means. There were sharp criticisms of some means proposed, but in general the word had a positive ring. The present strong negative connotations are a product of historical reflection after the Second World War. Especially since the 1970s, "eugenics" has become associated with the racist ideology and population policies of the Nazi regime in Germany, leading up to the Holocaust and other atrocious deeds of genocide.

The first inclusive foreign-language overview of the history of eugenics and sterilization in Scandinavia was *Eugenics and the Welfare State*.[46] In the autumn of 1997, this sort of information gave rise to a sensational story in mass media around the globe about a eugenic sterilization program in the Scandinavian welfare states comparable in extent and brutality to that of Hitler's Germany. It started with an article by the historian of ideas and cultural journalist Maciej Zaremba in the Swedish national daily newspaper *Dagens Nyheter* on August 20, 1997.[47] This spawned a wave of sensational stories about large-scale eugenic sterilization carried out from the late 1930s till the 1970s. For instance, the *Washington Post* told about "a 40-year Nazi style campaign of forced sterilization." A *Reuter* telegram proclaimed that "Social democratic Swedish governments sterilized 60,000 women to rid society of 'inferior' racial types and to encourage Aryan feature." *The Guardian* declared that "The laws ... could have come out of a Nazi text book."[48]

The editors of *Eugenics and the Welfare State* commented in the preface to the 2005 second edition on the politics of this event. On the one hand, it was part of running debates over the heritage and future value of the Nordic welfare state. And on the other, it was involved with the controversies over the enlightenment ideal of

[45]Turda 2010.

[46]Broberg and Roll-Hansen 1995.

[47]Zaremba later developed his ideas into a book, *De rena och de andra. Om tvangsteriliseringar, rashygien och arvsynd* (The pure and the others. On compulsory sterilization, race hygiene and original sin), privately published, printed in Finland 1999.

[48]References in Broberg and Tydén 1999.

science and scholarship. It raised fundamental issues about the nature of science and the relationship between natural and human sciences.

> This media event in the summer of 1997 demonstrated both how historical interpretation can be a powerful weapon in the political struggle and how a combination of political fashion and media attention can distort historical accounts. The event raised, in a sharp and interesting way, questions concerning truth, honesty, and appropriate behavior for scientists and politicians as well as journalists. This time the focus was not on the social responsibility and moral integrity of natural science (in this case genetics) but on that of humanistic sciences. Perhaps the event can be taken as a reminder of the close interdependence of the natural and humanistic sciences: that they will fall or stand together and that the widening ideological gulf between them is a serious threat to a productive social role for the Western scientific tradition—taking science in the broad continental enlightenment sense including the natural as well as the humanistic sciences.[49]

The sensationalist misperceptions of the 1997 mass media reports made a lasting impact on both the public mind and the scholarly literature. As already pointed out, attempts to correct mistakes and outline a more correct story had little effect. Two recent examples are witnesses of continuing misunderstanding:

A recent book on sterilization policy in North America includes twenty pages of comparison with Europe. These pages are almost all on Nazi Germany which thus appears as typical of sterilization and eugenics policies in Europe. Revealing of a superficial understanding of social policy conflicts is a passing mention of Jon Alfred Mjøen and his Vindern Biological Laboratory as if his activities were representative of events in Norway.[50] Another example is a general analysis of the relation of science to public policy where "race science" is a main example besides Keynesian economics and climate science. Again it is primarily about Nazi Germany and how biology inspired the Holocaust. Nazi population policies built on perfectly legitimate science the readers are assured. It is a mistake to call it "pseudoscience," though it was mistaken and misleading. The scientific criticism is not taken into account. And the 1997 "discovery that Sweden had a long-standing, state-sponsored eugenics program" is accepted without question.[51]

Both these accounts exemplify that the instrumental view of science has dominated science studies since the 1970s. On this view, science affects society through its practical technological applications. Science and technology is seen as a whole. The distinction becomes unimportant. And the technological emphasis makes it clear that political control is needed. "Technoscience" is the common term for this unified compound of science and technology.

This instrumental view of science is not new to the late twentieth century, but it gained influence through the influence of the Frankfurt School on the cultural revolution driven by the student movements in the 1960s and 1970s.[52] A traditional alternative view, which could be called the classical liberal view of science, is that science does not only provide instruments and techniques for solving practical

[49] Broberg and Roll-Hansen 2005, ix.

[50] Hansen and King 2013, 154.

[51] Grundmann and Stehr 2012, 76–77, 114.

[52] An influential example of this pessimistic and instrumental view of science is Horkheimer 1947.

social and economic problems. Besides its technical practical usefulness, science is a basis for understanding the problems. In this sense, it comes before politics and is a precondition rather than an instrument for politics. In this perspective, science is also a platform for fundamental criticism and revision of fundamental social and political goals. To effectively serve this second social function, science needs autonomy from politics, an autonomy which is often denied by dictatorial governments. To uphold and defend such autonomy is difficult without a clear understanding of the difference between science and technology, or pure and applied science, as it has traditionally been called.[53]

In recent years, it has become clear that the combination of new reproduction technologies and free individual choice raises questions quite similar to those of the old eugenics debates.[54] The potential for genetic manipulation of children's hereditary potential, "designer babies," is rapidly increasing. As this technological potential is transformed into practice, it will unavoidably affect the gene pool and thus be eugenic in the strict sense, in effect if not in intention. This "backdoor" or "laissez-faire" eugenics is no less in need of regulation than the primitive old technique of sterilization.[55]

The science of human genetics has to face this predicament. As a science, its nature is to produce new knowledge and thus unavoidably inspire new technologies and practical procedures. Since the creation of modern genetic science a century ago, the possibility of humanity steering its own evolution as a biological species has appeared as both a utopian dream and a nightmare. From early on, it was pointed out that this was fundamentally a political question, depending on what kind of society was the aim. Socialist ideals of solidarity, equality, and freedom were influential among geneticists in the mid-twentieth century. This social responsibility of genetic science was the nerve of a document that was signed by leading geneticists on the eve of the Second World War. They were gathered at the Seventh International Congress of Genetics in Edinburgh when the war broke out in September 1939. The gist of the document was to uphold a positive view of future application of human genetics squeezed between Nazi misuse and Soviet suppression of scientific freedom.[56]

This study has argued that a proper understanding of the history of eugenics, and in particular the role of the science of genetics, is important for the future handling of pressing political issues about the application of scientific knowledge to social practice. The history of eugenics is also a primary example for any general theory about the interaction of science and politics, their "co-production" as it is often

[53]Details of such an argument are found in Roll-Hansen 2009, 2010.

[54]Buchanan et al. 2000.

[55]Bashford and Philippa 2010a, b.

[56]The document has been called "The Geneticists' Manifesto" and was authored mainly by Herman J. Muller. A perceptive account of the circumstances and Muller's thinking is found in Paul 1998.

called in present science studies. I agree fully with Diane Paul that in general present narratives of eugenics are too one-sided: "As a guide to future action, they are therefore profoundly deficient."[57]

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[57] Paul 2007, 15.

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Changing the Point of View: The History of Human Genetics as an Applied Science in the Federal Republic of Germany, 1945–1975

Heike I. Petermann

Abstract In the history of human genetics in Germany, there has been less focus on the aspect of it as an applied science than as clinical genetics.

The approach in this chapter is on the meaning of the term, the influence of human genetics on legislation, the specialisation in the Federal Republic of Germany and the first tasks in medical genetics.

This has several aspects, such as the initial situation, with the consequences of war and the UNESCO Statement on Race, the establishment of human genetics at universities forced by political will and the international relationships such as representation at international congresses. The term "Human Genetics" was used for the first time in the 1940s; until then e.g. "Erblichkeitslehre" characterised this field. The matter in the handbooks of human genetics was the same as in English ones in the first and in further publications.

The *Wissenschaftsrat* (German Science Council) stated in 1960 that a chair for Genetics is necessary at every medical faculty. This was the stimulus for the professionalisation of human genetics, and institutes for human genetics were established. But it can be stated that there was a personal continuity relating to the time before 1945. The scientific questions remained the same.

The field of activities was focusing in the beginning on paternity tests and genetic counselling. The initial problems were lack of appropriate premises, but also the unsolved question of reimbursement for these jobs.

The increasing knowledge in human genetics had no influence on legislation in the first decades after 1945. It was important for the first time in changing the abortion law at the beginning of the 1970s.

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"Changing the point of view" shows that development of human genetics occurred under the prevailing law. Today the actors are seen to be more critical.

Keywords Human genetics • History • Federal Republic of Germany • University

The history of human genetics in both parts of Germany is associated with and has the burden of the Third Reich. During the period from 1933 to 1945, human heredity was in the focus of population policy and institutes for racial hygiene, racial biology or human heredity were founded at universities but also as research institutes like the *Kaiser-Wilhelm-Institute for Anthropology, Human Heredity and Eugenics* in Berlin-Dahlem. After 1945, those no longer existed, but members of their staff continued their research in the field of human genetics. This influenced the appraisal of human genetics after 1945.[1]

Will this image change, if one looks at the process of the establishment of human genetics?

To change the perspective means to look first at the historical context: the foundation of the Federal Republic of Germany (FRG) by using archive files; second how human geneticists judged this time; and third whether applications were introducing this.

> Human Genetics is both a fundamental and an applied science [...] Because of its continued theoretical and practical interest, human genetics offers fascination and human fulfilment unparalleled by work in fields that are either primarily theoretical or entirely practical in subject matter.

So stated Friedrich Vogel (1925–2006) and Arno Motulsky (b. 1923) in 1986.[2] This statement can also be regarded as a description of the development of human genetics in the FRG and other Western countries.

In 1985, Richard von Weizsäcker (1920–2015), at that time president of the *Federal Republic of Germany*, stated that from day to day it became more and more clear to us that May 8, 1945, was the day of liberation of the inhuman system of the National Socialist dictatorship.[3] This was his remark about the development in the past 40 years.

1 The Situation in 1945 and Beyond

Germany (*Deutsches Reich*) was destroyed and laid in ruins and the population had little or no hope. With the Declaration of Berlin (*Berliner Deklaration*), the four victorious states, the United Kingdom of Great Britain and Ireland (UK), the provisional government of France, the United States of America (USA) and the

[1] Weiss 2010; See e.g. Weingart et al. 1988 and Kröner 1998.

[2] Vogel et al. 1986, X.

[3] Weizsäcker 1985, 2.

Union of Soviet Socialist Republics (USSR) took over the power. They determined four occupation zones in the country and a special status of Berlin. The executive was the Allied Control Council (*Alliierter Kontrollrat*) that reigned since July 1945 by orders, directives, proclamations and laws. Those were published in English, French and Russian with a German translation.

In the Nuremberg Trials (*Nürnberger Prozesse*) first the major war criminals (*Hauptkriegsverbrecher*) were accused, if they had not committed suicide like Adolf Hitler (1889–1945), Heinrich Himmler (1900–1945) and Joseph Goebbels (1897–1945), and were kept in captivity. Further trials were the USA versus e.g. physicians or judges. Important till today is the *Nuremberg Code* as a result of the "Doctor's Trial" (*Nürnberger Ärzteprozess*) with principles for research and human experimentation.

The last decision of all four occupation states was on February 20, 1948. After that, in the western parts the *Allied High Commission for Occupied Germany* (HICOG) continued in power till 1955. In the Soviet zone, the development was determined by the USSR and resulted in the foundation of the *German Democratic Republic* (GDR). All regulations which determined life in the western occupation zones and then in the FRG came to an end by the reunion of the both German states of October 3, 1990, and by the *Two-plus-Four-contract* from March 15, 1991.

The Potsdam Agreement (*Potsdamer Abkommen)* from August 2, 1945, was the basis for the purge of the political and economic as well as spiritual and cultural life of national-socialist elements, called denazification (*Entnazifizierung)*. About the success of this method, there exist different opinions and the practice of denazification was different in each occupied zone: the American authorities operated restrictively, while the British and French organs acted less strictly.[4] The results of denazification influenced the establishment of human genetics in the FRG.

At the universities most of the professors returned, because most of them were regarded unavailable during the war and therefore survived. Other colleagues might have been killed in the war or have emigrated because of the political situation. After the war, all people had the right to return to their job in the civil service, if he or she had passed the denazification according to Article 131 of the constitution of the FRG (Grundgesetz, GG).[5] This was not indisputable, because it included continuity from the German Reich (*Deutsches Reich*) to the Federal Republic of Germany (*Bundesrepublik Deutschland*). Above all, it meant that all civil servants of the National socialist government could return to their jobs.[6]

Immediately after the war, the western allies were forced to rebuild all structures that were important for the population, like the health system. Therefore, they sent

[4]First the USSR exited denazification in February 1948, followed by the USA in August 1949, UK and France in February 1950.

[5]Einigungsvertrag 1990, 891. According to the Einigungsvertrag (Artikel 6) § 131 became not valid in the federal states of Brandenburg, Mecklenburg-Vorpommern, Sachsen, Sachsen-Anhalt, Thüringen and also East-Berlin.

[6]No right to return had people judges as principal accused saying "Hauptbeschuldigter" or "belastet".

medical staff such as surgeons, anaesthetists and internists to inform their German colleagues about the state of the art in medicine.[7] In these groups, there was no human geneticist and so this field had to be reestablished by those who were still available.

2 The UNESCO Statement on Race

Starting in 1942, there were already meetings on how to rebuild culture and education after World War II. In 1945, the *United Nations* were founded and immediately afterwards the *United Nations Educational, Scientific and Cultural Organization* (UNESCO). In the preamble, it was clearly expressed that political and economic agreements cannot alone secure peace:

> That a peace based exclusively upon the political and economic arrangements of governments would not be a peace which could secure the unanimous, lasting and sincere support of the peoples of the world, and that the peace must therefore be founded, if it is not to fail, upon the intellectual and moral solidarity of mankind.[8]

This was continued with the demand as far as possible for the education for everybody and the free exchange of ideas and knowledge. And this is valid till today:

> UNESCO's mission is to contribute to the building of peace, the eradication of poverty, sustainable development and intercultural dialogue through education, the sciences, culture, communication and information.[9]

On December 10, 1948, the meeting of the UN plenary assembly passed the *Universal Declaration of Human Rights*.[10] This is based, like many comparable explanations of the time, on the experiences of the Second World War. In article 1, it was stated:

> All human beings are born free and equal in dignity and rights. They are endowed with reason and conscience and should act towards one another in a spirit of brotherhood.[11]

This was followed by the statement in article 2 that the fact that the rights and freedoms of this Declaration without distinction are valid for everyone, regardless of race, colour, gender, language, religion, political or other setting, national or social origin, possession, birth or other status. The essential characteristics of this declaration are part of the basic law (*Grundgesetz*) of the FRG in the fundamental rights.[12] In article 1 (2) is written:

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[7] Petermann 2007.

[8] UNESCO 2004, 7.

[9] www.unesco.org: introducing UNESCO: what we are. [06.10.2016]

[10] United Nations General Assembly (Dec. 10, 1948): General Assembly Resolution 217 A (III).

[11] Ibid.

[12] Those comprise the articles 1–19.

> The German people therefore acknowledge inviolable and inalienable human rights as the basis of every community, of peace and of justice in the world.[13]

So the *UN Declaration of Human Rights* formed the basis for the FRG.

For the history of human genetics after 1945, the publications of the UNESCO on racial issues were important:

Statement on Race	Paris	1950, July
Statement on the Nature of Race and Race Differences	Paris	1951, June
Proposal on the Biological Aspects of Race	Moscow	1964, August
Statement on Race and Racial Prejudice	Paris	1967, September

Those were published in two series *The Race Question in Modern Science* and *The Race Questions and Modern Thought*.[14] An overview gave the anthology *The Race Concept: Results of an Inquiry*, in that is the declaration *Statement on Race*. In that it was stated:

> 1. Scientists have reached general agreement in recognising that mankind is one: that all men belong to the same species, Homo sapiens. It is further generally agreed among scientists that all men are probably derived from the same common stock [. . .].
>
> 2. From the biological standpoint, the species Homo sapiens is made up of a number of populations, each one of which differs from the others in the frequency of one or more genes . . .[15]

This text was reported by the British-American anthropologist Ashley Montagu (1905–1999), while the text was revised, e.g. by the human geneticists Hermann J. Muller (1890–1967) and Curt Stern (1902–1981). 96 biologists and geneticists were asked to give a comment on this statement of race, of which 69 answered. Replies came from heredobiologist Fritz Lenz (1887–1976) and the anthropologists like Eugen Fischer (1874–1967), Karl F. Saller (1902–1969) and Hans Weinert (1887–1967). Most of the work was done by the (human) geneticists Hans Grüneberg (1907–1982), Tage Kemp (1896–1967), Ernst Mayr (1904–2005), Jan G. F. Mohr (1921–2009), Hermann J. Muller (1890–1967), Lionel S. Penrose (1898–1972), Sheldon C. Reed (1910–2003) and Curt Stern (1902–1981).[16] One year later the declaration was modified: "*1. Scientists are generally agreed that all men living today belong to a single species, Homo sapiens, and are derived from a common stock, even though there is some dispute as to when and how different human groups diverged from this common stock.*"[17] This revised version was also

[13]"Das Deutsche Volk bekennt sich darum zu unverletzlichen und unveräußerlichen Menschenrechten als Grundlage jeder menschlichen Gemeinschaft, des Friedens und der Gerechtigkeit in der Welt."

[14]The Race Question in Modern Science. Paris 1959; The Race Question and Modern Thought, Vol. 1–6. Paris 1953–1970.

[15]UNESCO 1952, 92–97.

[16]UNESCO 1952, 92–97.

[17]UNESCO 1952, 350.

signed by Hans Nachtsheim (1890–1979) and was reported by Leslie Clarence Dunn (1893–1974).[18]

These statements were fundamental and valid for all sciences. Because of those the scientific discussion about characteristics and qualities of races came gradually to an end. In aspects of heredity, this term "race" lost its important role in general.

This statement reflects the discussion in the beginning in the 1950s. By those statements all considerations on race or racial hygiene came gradually to an end. In occupied FRG, there was no way to continue the discussion on these topics regardless of one's own opinion.

3 The Term "Human Genetics" in German

In 1949, the human geneticist Curt Stern, who had worked at the *Kaiser-Wilhelm-Institute for Biology* in Berlin-Dahlem till 1933, wrote:

> Human genetics is a young science. When, in 1900, the modern study of inheritance began, plants and, soon, animals formed its material.[19]

Very soon the subject of this science became the general rules of heredity in man. Without the experiments of Gregor Mendel (1822–1884), this would not have been possible.[20]

In German-speaking countries the heredity of characteristics and qualities was named "human heredity" (Menschliche Erblichkeitslehre oder Erblehre) or "heredobiology of man" (Erbbiologie des Menschen). So Fritz and Widukind Lenz (1919–1995) stated in 1968.[21] They continued that for some years, but beginning in England and America, the name "human genetics" became internationally common.

In German, the term was published for the first time in 1940. The biologist Günther Just (1892–1950) wrote in his handbook of hereditary biology of man (*Handbuch der Erbbiologie des Menschen*[22]) that this was a summary of the knowledge and reflects the present state of research in human genetics, "*im gegenwärtigen Forschungsstande der Humangenetik*".[23] But, he just named the term and gave no definition or further explanations. Vogel said about the book that it hardly contains traces of Nazi ideology.[24]

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[18]Dunn 1975, 343–354.

[19]Stern 1949, 3.

[20]Stern 1949, 1.

[21]Becker 1968, 1.

[22]Just 1940.—Published from 1939 to 1940. Just 1940.

[23]Just 1940, V.

[24]Vogel 1999, 410.— "Auch das ‚Handbuch der Erbbiologie des Menschen' [. . .] enthielt kaum Spuren von Nazi-Ideologie. "

In 1947, Laurence Hasbrock Snyder (1901–1986) stated in *The Principles of Heredity* the tasks of human genetics:

> These applications are first: genetic prognosis, that is, the furnishing of genetic advice in prospective marriages and prospective families; second, diagnosis, on the basis of genetic data, of conditions difficult to identify by other means; third, instituting of preventive measures against certain diseases and abnormalities, on the basis of specific genetic backgrounds; fourth, determination of nonpaternity, and other medico-legal problems, on the basis of test characters, particularly the blood agglutinogens; and fifth, recommendation of eugenic programs for the protection and improvement of society, a problem which can be scientifically approached only with a broad understanding of the interrelations of heredity and environment.[25]

The book of Snyder was translated into German by the physician and anthropologist Wolfgang Lehmann (1905–1980) and therefore had influence in the FRG.[26] In the same way argued both the human geneticists Otmar von Verschuer (1886–1969) and Widukind Lenz in their German handbooks: genetics of man (*Genetik des Menschen. Lehrbuch der Humangenetik*) by Verschuer (1959) and medical genetics (*Medizinische Genetik*) by W. Lenz (1961).[27] At that time also the handbook of human genetics (*Lehrbuch der allgemeinen Humangenetik*) by Vogel (1961) was published, but also *Principles of Human Genetics* by Curt Stern and *Human genetics* by Victor McKusick (1921–2008) were translated into German.[28] There was no difference in the content of the different books, except that some focused more on the theory and others more on the application of human genetics.

At that time, in every published textbook there was a chapter on eugenics. From today's point of view, this might seem astonishing. The aim of eugenics was expressed by Friedrich Vogel (1925–2006) as follows:

> Ziel aller Eugenik ist es, zu bewirken, dass vorteilhaftere Erbanlagen in größerer Anzahl an die folgenden Generationen weitergegeben werden als schädliche.[29]

He stated that more advantageous hereditary factors should be transmitted to the next generation. For this, there are two ways open: on one hand, one can try to prevent that injurious genes are transmitted (negative eugenics) or, on the other hand, to increase the number of valuable hereditary factors. This is based on the definition given by Francis Galton (1822–1911) in his talk "Eugenics: its definition, scope and aims" in London 1904 and published in *Archiv für Rassen- und Gesellschaftsbiologie* in 1905.[30] This was regarded as the fundamental document of Eugenics in German-speaking countries[31] and defined as follows: "Eugenics is the science which deals with all influences that improve the inborn qualities of a

[25] Snyder 1947, 114.

[26] Snyder 1955.

[27] See Verschuer 1959, 4; Lenz 1961, V.

[28] Stern 1955; McKusick 1968.

[29] Vogel 1961, 628.

[30] Galton 1904 and 1905.

[31] Muckermann 1929, 7.

race; also with those that develop them to the utmost advantage."[32] His closing remarks were: "The first and main point is to secure the general intellectual acceptance of eugenics as a hopeful and most important study."

A similar definition had in German the word Race Hygiene (Rassenhygiene), defined for the first time by Alfred Ploetz (1860–1940) in his fundamental book on the ability of our race and the protection of the weak (*Die Tüchtigkeit unserer Rasse und der Schutz der Schwachen*[33]) in 1895. He saw this as the apprenticeship of conditions of the optimal preservation and perfection of the human race: "Rassenhygiene ist die Lehre von den Bedingungen der optimalen Erhaltung und Vervollkommnung der menschlichen Rasse."[34] This definition represented the smallest common denominator. Although there were differences, Eugenics and Race hygiene were regarded as the same before 1945 in Germany.[35]

After an analysis of the different German handbooks on human genetics, one can state that the reference point for eugenics is Francis Galton. His term was scientifically oriented and put inheritance in the centre, while in contrast the concept of race hygiene was oriented above all socially. This might be the reason that the term eugenics is used till today.[36]

In *The Return of Eugenics* (1988), R.J. Neuhaus stated that Eugenics is a movement with the attitude to improve or perfect man (the human species) by engineering and that "the horror of the Third Reich may have effected but a momentary pause in the theory and practice of eugenics". The ideas of prolonging life, prevention and healing diseases as well as improvement of physical and mental characteristics have been developing throughout history.[37] And Michael J. Sandel stated in 2007, "Eugenics was a movement of large ambition—to improve the genetic makeup of the human race".[38]

This meaning of the term Eugenics is considered to have no political correlation, but only a scientific one. Therefore, it is still part of human genetics in Germany and elsewhere.

At the middle of the twentieth century, scientists regarded the genetic programme of inheritance as a major difference between the world of life and that of inanimate objects, according to Ernst Mayr (1904-2005).[39] And Penrose added: "Human genetics is an applied science. It makes use of techniques of all kinds as they become available for the study of the hereditary processes in man."[40]

[32]Galton 1904, 1.—In German: Galton 1905.

[33]Ploetz 1895.

[34]Gütt et al. 1934, 237.

[35]Petermann 2005.

[36]See on the difference of the terms eugenics and race hygiene Petermann 2005; 2009.

[37]Neuhaus 1988.

[38]Sandel 2007.

[39]Mayr 1984, 505.

[40]Penrose 1959, 9.

4 The Beginning of Human Genetics at Universities

After the war life in the USA went on as usual, but meanwhile in Europe and especially in Germany nearly everything had to be rebuilt, from buildings to infrastructure and institutions. At German universities at all faculties, there was extensive destruction, as also at the medical centres. Institutes and even hospitals were destroyed. Rebuilding of those started with departments of internal medicine and surgery that were essential for all people. Other faculties and fields followed continuously.

All institutes for human heredity that were founded from 1933 to 1945 were closed for political reasons. Till the end of World War II, most of those institutes were closely intertwined with the political system of the Third Reich e.g. with the system of compulsory sterilisation because of hereditary diseases (Table 1).[41]

But there was a need for research on human genetics and institutes had to be established. Therefore, well-known people in the field of human heredity were allowed to carry on their research in human genetics: 1946 Fritz Lenz in Gottingen, 1951 Otmar von Verschuer in Muenster, in neurologic and psychiatric research supported by Gerhard Koch (1913–1999), and 1952 Wolfgang Lehmann in Kiel but also Hans Nachtsheim in Berlin. All of them have in common their relationship to the *Kaiser-Wilhelm-Institute for Anthropology, Human Heredity and Eugenics* (KWI), as director (Verschuer), head of department (Lenz, Nachtsheim), members (Becker, Lehmann) or guest for research (Koch).[42]

Already in 1946 Nachtsheim was given a professorship for genetics at the university of East Berlin, but left this in winter 1948/49. At that time, the *Freie Universität* was founded in West Berlin with a chair for genetics at the mathematical-natural-scientific faculty and his department of experimental heredopathology became first part of the *Deutsche Forschungshochschule* and later of the *Max-Planck-Institute* as department of comparative heredobiology and heredopathology (*vergleichende Erbbiologie und Erbpathologie*).[43] He was regarded as unhampered by reproach for being involved in the Nazi system. At that time, there was nothing public about his experimentation on children.[44]

4.1 Göttingen: Fritz Lenz[45]

On January 1, 1946, the dean of the medical faculty, *Georg-August University Gottingen*, wrote to the head of province of Hannover referring to the establishment

[41] Vogel 1999.

[42] See Schmuhl 2005 and Vierhaus 1990.

[43] Nachtsheim 1961 und Kröner 1998, 1–2.

[44] See Schwerin 2004.

[45] UAG: Kur. PA Lenz, Fritz, and Kur. 1053.

Table 1 Institutes for Heredobiology and Race Hygiene at the Universities before 1945

Place	Institute at the Medical Fakulty	Years	Chair
Berlin	KWI for Anthropology, Human Heredity and Eugenics	1927–1942 1942–1945	Eugen Fischer Otmar von Verschuer
	KWI Abt. Eugenik	1933–1945	Fritz Lenz
Düsseldorf	Associate professorship for Hereditary Hygiene and Race Health	1934–1940	Friedrich E. Haag
Frankfurt	Institute for Heredobiology and Race Hygiene	1935–1942 1943–1945	Otmar von Verschuer Heinrich W. Kranz
Gießen	Institute for Hereditary and Race Health	1934–1942 1943–1945	Heinrich W. Kranz Herman Boehm
Hamburg	Institute for Anthropology (Ordinariat 1933)	1924–1965	Walter Scheidt
Kiel	Anthropological Institute	1923–1935	Otto Aichel
Köln	Institute for Heredobiology and Race Hygiene	1939–1945 1941–1945	Ferdinand Claussen (V) Wolfgang Bauermeister
München	Institute for Race Hygiene	1923–1933	Fritz Lenz
		1933–1936	Lothar G. Tirala (Amtsenthebung)
		1936–1944	Ernst Rüdin (komm.)
Tübingen	Institute for Anthropology and Racial Science Heredobiological Institute (1938) Anthropological Institute (1945)	1934–1968	Wilhelm Gieseler
Würzburg	Heredobiological Institute	1937–1941	Ludwig Schmidt-Kehl (†)
		1941–1942	Friedrich Keiter (komm.)
		1942–1945	Günther Just

This table only names universities that were later situated in the Western part of Germany (FRG)

of a professorship for human heredity. They stated that Fritz Lenz, professor for human heredity in Berlin, would come to Gottingen, if he were given a professorship. Then it was added that he is the well-known author, together with Erwin Baur (1875–1933) and Eugen Fischer (1874–1967), of the basic textbook on human heredity.[46]

In November 1946, Fritz Lenz arrived in Niedersachsen as a refugee from Berlin (*Ostzonen-Flüchtling*). Soon after his arrival, he had applied at the University of Göttingen. On December 2, 1946, the dean of the medical faculty wrote a letter in favour of this application for establishing an institute of human heredity (*Institut für menschliche Erblehre*). This became part of the budget for the years 1947/48 at the University of Göttingen. A decree of July 24, 1946, said that a professorship for

[46]Baur et al. 1923.—Up to the year 1944, five editions were published. The first part was translated into English in 1931.

human heredity will be established. On September 28, 1946, this was approved by the British university control officer. Since November 21, 1946, Fritz Lenz was extraordinary (*außerordentlicher, a.o.*) professor in Göttingen and was a part-time employee. His job was named as paternity testing, without any further explanation.[47] Receiving this, Lenz asked for one office worker, assistant student and assistant lecturer, but also for 2–3 rooms next to the Institute for Hygiene.

In his personal record, there is a statement that he had been professor in Berlin since November 1, 1933. According to Article 131 of the constitution of the FRG (Grundgesetz, GG), it was said that persons like him should get a comparable position, if denazification is not inconsistent with this appointment. Finally, on July 14, 1949, the denazification of Lenz was held in written procedure with the result that he could work without any restrictions (*ohne jede Beschränkung*).[48] This decision was based on a personal statement of Lenz, on his professional record and on a list of his publications. It is remarkable that all articles in political magazines of the Third Reich like "Aim and Way" (*Ziel und Weg*) as well as "National socialist Monthly Reports" (*Nationalsozialistische Monatshefte)* were not named.

In the above named letter of the medical faculty, it was stated that he has argued in propagandistic publications only in a scientific but not in a political way: *"Er hat zu vielen in den vergangenen Jahren propagandistisch entstellten Fragen seines Faches wissenschaftliche eine Stellung eingenommen, welche seine Integrität erkennen lässt."*[49]

So argued Fritz Lenz also in the questionnaire and he named as witnesses inter alia the three human geneticists Hans Nachtsheim, Paul Popenoe (1888–1979) and Tage Kemp.

On August 8, 1952, Lenz was given the title "ordinary professor" for the time he belonged to the teaching staff (*Lehrkörper*) of the University of Göttingen.[50] During his professorship, he gave lectures on human heredity, medical genetics as well as marriage and hereditary counselling. At the end of winter semester 1954/55, he became professor emeritus, but represented himself till winter semester 1956/57. His successor was Peter Emil Becker (1908–2000) in 1957 (till 1975), whom he knew from the KWI in Berlin. For Becker also § 131 GG was valid, because he had been at the KWI by the end of the war.

Fritz Lenz was the first professor for human heredity in FRG after 1945 in spite of all doubts. In his personal records (*Personalakten*) at the university archive, there are newspaper reports that argued that Fritz Lenz was not the right person for this job.[51] But this did neither influence the British occupying power nor the University of Göttingen.

[47]UAG: Kur. PA Lenz, Fritz.

[48]Entnazifierungs-Hauptausschuss / Göttingen, 15.07.1949 / Kategorie V (5)—ohne jede Beschränkung.

[49]UAG: PA Lenz, Fritz (1): fol. 8v.

[50]UAG: PA Lenz, Fritz (1): fol.112.

[51]Ibid.

4.2 Münster: Otmar von Verschuer

Before 1945, there was no professorship or institute for race hygiene or human heredity at the university in Muenster.[52] In the year 1950, it was decided that an institute for human heredity and anthropology should be established at the *Westfälische Wilhelms-University* of Münster, medical faculty.[53] The Jesuit Hermann Muckermann (1877–1962), who was head of a department at the KWI in Berlin till 1933, was recruited to hold the chair as a temporary replacement.[54] On October 25, 1950, the medical faculty suggested that Otmar von Verschuer should be nominated for this professorship. It was said to be urgent, because he might be appointed in Tübingen.[55] On February 22, 1951, there was signed an appellate agreement: Verschuer should take over the chair for human genetics (human heredity), starting April 1, 1951, with his own secretary.[56] His appointment was dated March 17, 1951. In this, there were named 60 boxes of material that would come to Muenster and had its origin in the KWI Berlin.[57]

The denazification of Verschuer took already place in 1946: to him was sent an atonement reply (date: 11,7,1946) with the information that he was judged as a sympathiser (*Mitläufer*) and had to pay 600 RM till December 7, 1946, or as replacement 14 days imprisonment. The fees for this were 2.038 RM, to pay within 8 days. Verschuer accepted this verdict. This meant—probably—that he wanted to pay the money. But Robert Havemann (1910–1982) contradicted this, and till 1949, it was a pending lawsuit, before it ended without any other result. Because of this Verschuer did not get professorships in Frankfurt and Tubingen.

In the archive records of the University of Frankfurt, there is a sheet, where Verschuer confirmed in 1937 that he has no Jewish ancestry and also that he was not a member of the NSDAP. Included is a letter of the Race-political office (*Rassenpolitisches Amt)* of the NSDAP, dated July 20, 1937, saying that Verschuer could not be named a National socialist, because he was objective and mainly scientific and apolitical. His appointment was essentially justified and might be helpful in a propagandistic way towards disbelieving circles.[58]

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[52]To the situation in Münster before 1945 see Kröner 1998 and 2012

[53]UAM: Bestand 052, Nr. 357: fol. 6.

[54]Ibid. fol. 7.

[55]Ibid. fol. 9.

[56]Ibid. fol. 15: Verschuer should get a salary of 13.600 DM plus housing benefit and family allowance; also he should get his removal costs.

[57]UAM: Bestand 052, No. 357.—This material is partly still available at the University of Muenster.

[58]UAM: Bestand 10, No. 3561: Personal record O. v. Verschuer, University Frankfurt a.Main, enclosed.

Verschuer continued his research in Münster, while using the library[59], the collection of reprints[60] and the other things he had brought from Berlin with him in 60 boxes.[61] His twin research was still basically intact and often cited. Also § 131 GG could be applied for Verschuer, because he was at the KWI in Berlin till 1944.

On February 14, 1951, Verschuer agreed to come to Muenster. His plans were to get a *Max-PIanck-Institute* (MPI) in Muenster, but he did not succeed. He was appointed professor on March 17, 1951.[62]

In 1958, Verschuer described the institute for human genetics in Muenster.[63] At this time, members of the staff were professor Otmar von Verschuer, lecturer Heinrich Schade (1907–1989) and assistant lecturers Gerhard Koch and Karl-Heinz Degenhardt (1920–1994). The material came from the *Kaiser-Wilhelm-Institut für Anthropologie, menschliche Erblehre und Eugenik* (Berlin): an archive with data from 5000 pairs of twins and also a longitudinal survey, special series of twins with tuberculosis and cancer for follow-up and genealogy database. Important were comparative hereditary biology and pathology as well as studies on the population in Westphalia. A new field was the examination of radiation injuries of the hereditary factors and their influence on the mutation rate. At the end, Verschuer stated that the outlined scope of duties makes institutes for human genetics at all universities necessary.

In which way von Verschuer was involved in research according to the aims of the National socialist regime is a subject that is still not answered definitively.[64] His publications show a strong national and conservative attitude since the 1920s, and this did not change during the Third Reich. Problematic is the relationship of the KWI to the concentration camp in Auschwitz: human material was sent to Berlin for research.[65] At the time Verschuer was appointed in Münster, this was not general knowledge. His scientific work, especially his twin research, was till the 1970s cited e.g. by German and Anglo-American authors and was fundamental for the questions of heredity. For 10 years, the institute in Muenster was the largest and most influential one in Germany.[66]

The next one appointed for human genetics at universities was Wolfgang Lehmann, colleague of Verschuer at the KWI in Berlin. He became lecturer at

[59]The library of the KWI was the stock of the institute of human genetics in Muenster.

[60]There are two collections of reprints, one he collected during his time in Frankfurt and Berlin and the other one was build up in Münster. A supplement is a collection of publications of the staff of the different institutes at the institute of ethics, history and theory of medicine, University Münster.

[61]UAM: Bestand 10, No. 3561.

[62]UAM: Bestand 10, Nr. 3651: fol. 16.

[63]Verschuer 1958.

[64]Publications on Otmar von Verschuer: Kröner 1998 und 2012; Schmuhl 2005; Weiss 2010.

[65]Physician Josef Mengele (1911–1979) was doctoral candidate of Verschuer in Frankfurt and was involved in the selection at the ramp in Auschwitz-Birkenau. See Sachse 2003.

[66]Vogel 1999, 410.

the *Christian-Albrechts-University* of Kiel already in 1948, but in 1956 this was changed to an extraordinary professorship and finally a professorship in 1962. It appeared that he was first employed on probation, while he was working as a primary care physician. His appointment in 1956 showed that he had proven himself as a human geneticist.

But human genetic research was not limited to institutes for human genetics; also those for anthropology were involved, like that in Munich. Its director since 1948 was Karl Saller and genetic research was done by Helmut Baitsch (1921–2007) since 1951 till 1958, when he became curator of the anthropological state collection (*Anthropologische Staatssammlung*).

In 1953, there was a breakthrough in the therapy of hereditary disease by the paediatrician Horst Bickel (1918–2000). He reported for the first time the treatment of a child with phenylketonuria by phenylalanine-reduced diet.[67] In the same year, James Watson (b. 1928) and Francis H.C. Crick (1916–2004) published their model of Deoxyribonucleic acid (DNA), about which Max Delbrück (1906–1981) gave a talk at the *Harnack House* in Berlin. Both discoveries did not influence the establishment of human genetics in the FRG: it was the threat by nuclear power and the influence of ionising radiation since it was known that those could cause mutations.[68]

5 The First International Congress on Human Genetics: Copenhagen 1956

> The knowledge of the conditions affected by heredity makes it possible to follow and control their development and fluctuation in the population and to ascertain the behaviour of hereditary diseases down through the ages.

So congress president Tage Kemp stated at the opening of *The First International Congress of Human Genetics*.[69] The congress was organised by a Danish Committee, supported by 14 national committees, including UK, the USA and France but also FRG. Members of the German committee were Otmar von Verschuer, Fritz Lenz, Hans Nachtsheim and Otto Ullrich (1894–1957).[70] While Nachtsheim, Lenz and Verschuer held professorships for (human) genetics, Ullrich was at the paediatric clinic in Bonn.

At this congress, 12 German scientists gave talks and presented them in their native language like all the other participants. From today's view, this is astonishing, as are the topics of their presentations. For example, Gerhard Koch (Muenster) presented the results of a preliminary reexamination of the Berlin twin research

[67] Bickel 1953.

[68] Vogel 1999, 41.

[69] The congress was supported by the Rockefeller Foundation and WHO.

[70] Otto Ullrich described in 1930 first the characteristics of the Turner Syndrome.

series.[71] This research was based on his work at the KWI in Berlin before 1945. Other speakers, besides those already mentioned, were e.g. Ernst Rüdin (1874–1952, München), Karl Saller (München), Friedrich Keiter (1906–1967, Hamburg), Peter E. Becker (Tuttlingen), Karl-Heinz Degenhardt (Bonn), Widukind Lenz (Hamburg) and G. Gerhard Wendt (1921–1987, Marburg).

The First International Congress was important, because it was the first chance after 1945 to meet many scientists dealing with questions of human heredity from different countries and also from Germany, as Vogel stated.[72] But it also showed that German scientists were not isolated after World War II and were part of the scientific community.

In the personal record of Koch, one can find the fees for the congress: those were 150 Kronen (at that time 91 DM).[73] Koch and maybe also other participants had to pay about the half of their travelling costs on their own: that meant they must have had a great interest.

At the opening the Danish minister of Education stated: *"Human genetics and its importance in preventing the occurrence and spread of hereditary diseases have just become of intense . . . interest by the perspective arising from the peaceful use of atomic energy."*[74] This threat was the stimulating element for human genetics.

> In turning to the past we note that the importance of our Congress lies in the fact that Human Genetics has finally reached the status of a branch of the Biological Sciences, with its own characteristics that not only allow, but also require a specific, and at times, autonomous treatment.[75]

This was stated by the congress president Luigi Gedda (1902–2000) in the Proceedings of the second congress 1961 in Rome. As before, there were scientific committees of different countries. Part of the German one were again Becker and Verschuer, Wolfgang Lehmann (Kiel) and Karl Saller (München) but also Wilhelm Gieseler (1900–1976, Tübingen) and Ernst Kretschmer (1888–1864, Tübingen).

The number of German speakers had increased to 24. In addition to the above mentioned speakers from the first congress also contributed e.g. Helmut Baitsch (München), Heinrich Schade (Münster), Walter Fuhrmann (1924–1995, Berlin), Hartwig Cleve (1928–1994, New York) and Friedrich Schwarzfischer (1921–2004, München). These contributions showed that the field of human genetics has developed in the FRG and as previously in Copenhagen their presentations were in German.

At this congress, the participants again emphasised the fear of ionic radiation that was present, and a resolution was passed with the requirement that all governments should condemn nuclear power as a weapon. But also they should support

[71] Koch 1972. "Ergebnisse aus der Nachuntersuchung der Berliner Zwillingsserie nach 20–25 Jahren."

[72] Vogel 1999, 41.

[73] UAM: F2/1, Nr. 1872b.

[74] Proceedings 1956/57.

[75] Proceedings 1963, 17.

research on its influence on the genetics of man and establish controls for its peaceful use.[76]

Verschuer stated that human genetics as an independent field of medical sciences has been established by the First International Congress in Copenhagen 1956. Furthermore, that the term human genetics was asserted internationally:

> "Ihre Selbständigkeit als eigenes Wissenschaftsgebiet kam durch den 1. Internationalen Kongress für Humangenetik, Kopenhaen 1956, zum Ausdruck. Die anglo-amerikanische Bezeichnung 'Human Genetics' hat sich international durchgesetzt."[77]

6 The Establishment of Human Genetics

In 1955, the ministry for questions on nuclear power (*Bundesministerium für Atomfragen*) was founded in 1955. Verschuer saw these as his chance and reported to them on the mutations induced by radiation. At the end, Verschuer got funded for 15 years his project on registration of all hereditary diseases in the region of Muenster. Also Nachtsheim was given money for recording of selected genetic diseases. Both projects were the start for research on mutations in Germany but also for the establishment of institutes for human genetics at universities.[78] This was the stimulus for the establishment of human genetics in the FRG.

In 1946, Karl Jasper stated that universities should develop to the best and highest level. "*In the diversification of the whole the new one will be the whole, as life produces life*."[79] This leads to the point that the best performance will be a specialised one.

In 1957, the German Council of Science and Humanities (*Deutscher Wissenschaftsrat*) was founded by the Federal Governments and those of the federal states. As Konrad Adenauer (1876–1967), first Federal Chancellor of the Federal Republic of Germany, at the signing of the Administrative Agreement said of it, it was *"the first time than an institution has been created on German territory which is intended to provide an overview of scientific work in the Federal Republic o Germany [with regard to the advancement of science."*[80] This was the first advisory board for science policy in Europe.

The *Wissenschaftsrat* published in 1960 its recommendations for the further development of universities.[81] It stated that new professorial chairs at universities should only be

[76]Proceedings 1963, 38.

[77]Verschuer 1959, VII.

[78]Vogel 1999.

[79]Weber 1991, 11.

[80]www.wissenschaftsrat.de/history.

[81]Wissenschaftsrat 1968.

(a) for fields that were still in evolution and where it might be expected that there would be a chair in the future, and
(b) in special cases for the permanent support of small special fields.

In medicine, a chair for Genetics at every Medical Faculty was regarded necessary, supported by a chair for anthropology. Because of lack of scientists this was hard to fulfil. Therefore, for a short time the existing institutes should be extended to force the training of young professionals.

The *Wissenschaftsrat* considered it also desirable that the following special fields should be established: serological genetics, cytological genetics, biochemical genetics and clinical statistical genetics.[82] In all, the German Scientific Council postulated 35 chairs for genetics, 11 of these at Medical faculties (Table 2).

In consideration of the recommendations, Nachtsheim said that in Würzburg there should be an associate professorship, but there was nobody. In Tübingen, they believed that one associate professorship for anthropology should be enough and that Freiburg is not mentioned seems to be a mistake.[83] And he summarises: in Germany available candidates for this professorships, however, are not more than half dozen that could be suggested for a professorship or associate professorship.[84]

The lack of scientists was an important factor, not only in human genetics. But the political authorities wanted to rebuild all necessary institutions, in order to prevent a new dictatorship: if the people had everything needed they would not start a revolution.

The knowledge about the fact that always, in spite of all, a new beginning is possible, if one focuses the aim, is maybe the biggest consolation, the best strengthening which history is able to provide for us. Such was the comment of the former president Richard Weizsäcker on the responsibility of science.[85] This can also be seen as a description for the establishment of human genetics.

As already described, in 1960 there were only three institutes for human genetics at medical faculties (Göttingen, Kiel and Münster). According to the recommendations of the *Wissenschaftsrat* till 1975, the number increased to 24. Under those were Heidelberg and Freiburg (1961), later then Erlangen (1965). These three institutes stand for the different ways how human genetics was established at universities.

[82]Wissenschaftsrat 1968, 22.

[83]Nachtsheim 1961, 6.

[84]Dito. "Überblickt man aber die heute in Deutschland für diese Lehrstühle vorhandenen Anwärter, so last sich nicht einmal ein halbes Dutzend nennen, das man mit gutem Gewissen für ein Ordinariat oder auch nur für ein Estraordinariat vorschlagen könnte."

[85]Weizsäcker 1986, 1083. "Das Wissen darum, dass immer, trotz allem, ein Neuanfang möglich ist, wenn man das Ziel im Auge behält, ist vielleicht der größte Trost, die beste Stärkung, die uns die Geschichte zu vermitteln vermag."

Table 2 Human Genetics at the Medical Faculties at Universities in the FRG, 1945–1975

Place	Institute at the Medical Faculty	Years	Chair
Bonn	Institute for Human Genetics	1964–1980	Heinz Weicker
Düsseldorf	Institute for Human Genetics and Anthropology	1965–1974 1975–1996	Heinrich Schade Günther Röhrborn
Erlangen	Institute for Human Genetics and Anthropology	1965–1979 1979–1999	Gerhard Koch Gerhard Pfeiffer
Essen	Institute for Human Genetics	1977–2001	Eberhard Passarge
Frankfurt/M.	Institute for Human Genetics and Comparative Heredopathology	1961–1983	Karl-Heinz Degenhardt
Freiburg/Br.	Institute for Anthropology and Human Genetics (1965)	1961–1970 1970–2001	Helmut Baitsch Ulrich Wolf
Giessen	Institute for Human Genetics	1967–1992	Walter Fuhrmann
Göttingen	Institute for Human Heredity Institute for Human Genetics (1962)	1946–1955 1957–1975 1977–2014	Fritz Lenz Peter Emil Becker Wolfgang Engel
Hamburg	Institute for Human Genetics	1962–1965 1967–1993	Widukind Lenz Werner Goedde
Hannover	Institute for Genetics	1972–1990	Gebhard Flatz
Heidelberg	Institute for Anthropology and Human Genetics	1962–1993	Friedrich Vogel
Homburg/Saar	Institute for Human Genetics	1973–2003	Klaus Dieter Zang
Kiel	Institute for Human Genetics (1962)	1956–1970	Wolfgang Lehmann
Lübeck	Institute for Human Genetics	1973–1978	Rudolf A. Pfeiffer
Marburg	Institute for Human Genetics	1963–1985	Gerhard Wendt
München	Institute for Anthropology and Human Genetics (1958)	1949–1969 1969–1973 1973–1994	Karl Saller Gerfried Ziegelmeyer (komm.) Hartwig Cleve
Münster	Institute for Human Genetics	1951–1964 1965–1985	Otmar von Verschuer Widukind Lenz
Tübingen	Institute for Anthropology and Human Genetics (1962)	1955–1968 1969–2001	Wilhelm Gieseler Horst Ritter
Ulm	Dept. Human Genetics Dept. Medical Genetics Institute for Human Genetics (2000)	1975–2000 1978–2010	Winfrid Krone Walther Vogel

This table is based on Koch 1985, Vogel 1999 and files of the University Archives in Erlangen, Freiburg, Göttingen, Heidelberg and Münster. There are differences in the year of establishment, because data in the archive records differ from those that were published

6.1 Heidelberg: Friedrich Vogel

The Medical Faculty of the *Karl-Rupprechts-University* of Heidelberg had decided in its meeting of February 2, 1961, to apply for a full professorship for anthropology

and human genetics at the state ministry. In this application, the recommendations of the *Wissenschaftsrat* were named and that those quote a professorship in Heidelberg. Before somebody was appointed professor for human genetics in Heidelberg, other institutes were asked to suggest possible candidates. Besides Friedrich Vogel there were named e.g. W. Lenz (Hamburg), G. G. Wendt (Marburg), H. Schade (Münster), K.-H. Degenhardt (Münster) and F. Keiter (Würzburg). The suggestions were made not only by human geneticists but also by anatomists, psychiatrists and anthropologists. This was followed by a discussion in the Medical Faculty with arguments pro and contra the named candidates, but finally the decision was made in favour of Friedrich Vogel.

In 1953, Vogel had applied for a job at the Max-Planck-Institute (MPI) for comparative genetics (vergleichende Genetik). The head of the department Nachtsheim was quite surprised that someone was still interested in the field of human genetics. For Vogel, this was a groundbreaking decision for his life. His research dealt with retinoblastoma and also the electroencephalogram. On January 18, 1957, he received the appointment as lecturer (venia legendi) for human genetics.

In March 1962, the professorship for anthropology and human genetics was established at the medical faculty of the University in Heidelberg and on October 9, 1962, the physician Friedrich Vogel was appointed professor after a controversial discussion.[86] Also an institute was set up and supported by a sufficient number of scientists, medical technicians and secretaries.

As also happened at the University of Heidelberg, Friedrich Vogel was asked and made suggestions for professorships, when he was asked. Of those, who were named by him, G. Koch was appointed in Erlangen, W. Lenz in Münster, H. Schade in Düsseldorf and Hans-Werner Goedde (b. 1927) in Hamburg.[87] It can be stated that Friedrich Vogel was an influential person by the institutionalisation of human genetics in the FRG.

6.2 *Freiburg: Helmut Baitsch*

In 1954, the *Albert-Ludwigs-University* in Freiburg stated that the Institute for Anthropology was a total war loss. Two years later, the anthropologist Johann J. E. Schäuble (1904–1968) became the professor, but had no rooms at all. Since December 1st, 1956 Kurt Gerhardt[88] (1912–1993) was provisionally deputising, while Schäuble went to Kiel. Gerhardt was given time off for research at the university in Muenster, so he could take his job.

[86]UAH: Acc. 5/02.

[87]UAH: Acc. 5/01.

[88]Kurt Gerhardt was suggested by Otmar von Verschuer.

On April 8, 1961, the physician Helmut Baitsch was asked whether he was interested in an extraordinary professorship for anthropology and he was appointed on July 21, 1961. In 1958, Baitsch qualified as a university lecturer at the faculty for natural sciences and mathematics with a work on the objectification of the paternity test. Then he became curator at the anthropological state collection (*Anthropologische Staatssammlung*) in Munich. On May 26, 1962, Helmut Baitsch gave his introductory lecture on new results of research on chromosomes and was named extraordinary professor for anthropology as well as director of the anthropological institute. He gained the extraordinary professorship on July 21, 1961. In 1965, the name of the institute and extraordinary Professorship were changed to *Institute for Anthropology and Human Genetics*.[89] In 1966, Helmut Baitsch was given a personal and then in 1967 an ordinary professorship.[90] Kurt Gerhardt belonged since 1961 to the anatomical institute and since 1971 to the philosophical one. During all this time, he was also a member of the medical faculty.[91]

In 1969, Baitsch was granted time off for the organisation of a DFG special research field (Sonderforschungsbereich) for two years. Then, since July 20, 1970, Baitsch became rector of the University of Ulm for his lifetime.

On September 17, 1970, Ulrich Wolf (b. 1933) was appointed temporary head of the department. In 1972, he became director of the institute and gained the professorship.

6.3 Erlangen: Gerhard Koch

At the institute for human genetics in Muenster Gerhard Koch had his own research project as specialist for neurology and psychiatry as well as human geneticist since 1952. Half-time he worked at the clinic for neurology. He was appointed lecturer in June 1954 and 1960 extraordinary professor for human genetics.[92] For this, expert advice was given by Verschuer, Becker (Göttingen) and Lehmann (Berlin, Kiel).

In October 1942, Verschuer had written a letter of confirmation that Koch should proceed with his important research for 3 months at the KWI in Berlin, for which he had gained already money by the German research foundation (*Deutsche Forschungsgemeinschaft*, DFG).[93] So Koch could work at the institute since 1943 as a guest and did not have to fulfil his military service.

[89]UAF: B0124: 14. März 1965.
[90]UAF: B0124: 26.7.1967.
[91]UAF: B0124.
[92]UAM: Bestand 53, Nr. 29,1.
[93]Koch 1993, 99–100.

Gerhard Koch was imposed a fine of 1.500 RM in a denazification process on political cleaning with no further restrictions. This took place in the French occupation zone, in Baden, and is dated on May 20, 1947.[94] He was engaged to Anne-Marie Cudell from Portugal and the marriage was forbidden by the Reich office of genealogy (Reichssippenamt) Berlin in August 1943. His research was on heredity of symptomatic epilepsy.[95] It may be that this was the reason that he was not regarded as a follower of the national socialist regime.

After the war, he had worked as a specialist for neurology and psychiatry (1948) in Tübingen and since 1949 at a clinic in Lisbon, before he started to work in Münster.

On November 12, 1965, he was appointed professor for human genetics and anthropology at the *Friedrich-Alexander-University* in Erlangen.[96] In the winter semester, 1971/72 Koch became dean of the medical faculty.

On October 1, 1974, the institute for human genetics and anthropology became a clinical institution according to the Bavarian University Law (*Bayerisches Hochschulgesetz*, BayHSchG) of December 21, 1973.[97]

One of the tasks of the institute was genetic counselling. This started in 1966 with 13 and increased up to 314 consultancies in 1976.[98] Koch summarised that genetic counselling had become part of the life of families. For him the aim had to be a positive family planning so that parents with a high genetic risk can have healthy children.[99]

In a letter, dated August 13, 1969,[100] Gerhard Koch wrote that chromosome diagnostics became more and more important for clinical diagnostics and therapy as well as for eugenic counselling. Anthropologic and heredobiologic expert opinions were an integrating component of judgement in affiliation proceedings. In March 1972, the institute gained the right to bill the national health insurances and in 1973 the private ones. This was important to get reimbursement of costs. And in May 1974, there was a conference of all health ministers of the federal states. Their vote was positive on the topic of genetic counselling. So genetic counselling has been established as a task of institutes for human genetics.

On September 19, 1978, Rudolf A. Pfeiffer (1931–2012, Lübeck) became successor of Gerhard Koch in Erlangen.

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[94]FAU: F2/1—1872a.

[95]Koch 1993, 159.

[96]UAE: F2/1—1872a.

[97]Bayerisches Hochschulgesetz. Bayerisches Gesetz- und Verordnungsblatt (BayGVBl) 1973 (26): 679–708.

[98]Koch 1977, 20.

[99]Koch 1977, 1938.

[100]UAE: object record human genetics (unsigned).

6.4 Münster: Widukind Lenz

On October 1964, Verschuer had reached the retirement age and started to represent himself as director of the institute. Soon it became clear that Widukind Lenz would be appointed professor for human genetics. Therefore, Verschuer handed over the leadership of the institute on April 7, 1965. This was before Lenz was appointed on April 19, 1965.[101]

Also Widukind Lenz had also to undergo the process of denazification and was judged "unaffected" (nicht betroffen).[102] He became paediatrician in 1961 and gained his lectureship on July 16, 1958, in Hamburg and in 1961 the professorship for human genetics. His inaugural lecture on July 26, 1962, was on the influence of heredity and environment on the person.

The examples of these institutes show that different ways led to the establishment of human genetics at medical faculties of universities: anthropological institutes added human genetics and later also the focus of their work; others were newly founded as institutes for human genetics. The *Recommendations of the German Scientific Council* had a great influence on the establishment of human genetics.

Since the 1960s, there was "genetics of man" at the faculty of natural sciences and "human genetics" as a field at the medical faculty, also named "medical" or "clinical genetics". There was still anthropology as a field at the medical or natural scientifically or philosophical faculty. This depended on the persons that worked in this field. A general differentiation cannot be made.

The development is comparable to that of anaesthesiology: because of the recommendations new professorships were established. It can be stated that at all those had a great influence on the evolution of medicine.

7 The Professionalisation of Human Genetics

All fields of medicine go through different states by professionalisation. Paul Unschuld named for this different strategies like manipulation of knowledge, foundation of scientific organisations and establishment of a scientific journal.[103]

With the first handbook on human genetics in German, there was also created specialised knowledge. Most of the family doctors after 1945 did not know much about heredity of qualities and characteristics. So they could not follow the discussion in the scientific community of human genetics.

After 1945, the German Human Geneticists were members of either the society for research of constitution (*Gesellschaft für Konstitutionsforschung)* or the German society for Anthropology (*Deutschen Gesellschaft für Anthropologie*). Both

[101] UAM: Bestand 052, Nr. 357: fol. 5.

[102] Ibid.: Denazification main committee, region Hildesheim: 18.12.1950.

[103] Unschuld 1974 and 1978.

unified in 1965 to the society for Anthropology and Human Genetics (*Gesellschaft für Anthropologie und Humangenetik*). In 1987, the German Society for Human Genetics (*Deutsche Gesellschaft für Humangenetik*) had separated.

The third step was the foundation of the journal *Humangenetik* in the year 1964, since 1976 *Human Genetics*. Editors were Baitsch, Becker, Vogel, Arno Motulsky (Seattle) and Gerhard Wendt (Marburg). Its predecessor was the Journal of Human Heredity and Constitution (*Zeitschrift für menschliche Vererbungs- und Konstitutionslehre)*. This was edited by Verschuer, who was forced to resign. The new name marks a change like Weingart postulated.[104] *Medizinischegenetik* was first published in 1989 and is since 2006 edited by the *Deutschen Gesellschaft für Humangenetik* (GfH).

For the professionalisation of Human Genetics in Germany, it was important that human genetics had been recognised as a new field of medicine. Besides the establishment of human genetics at universities, the handbook and journals for human genetics in German and the foundation of a society were also important. The same is valid also for other fields of human genetics.[105]

8 The Tasks of Human Genetics

In the archive records of Erlangen, Freiburg, Göttingen, Heidelberg and Münster, the first tasks for the new established institutes for human genetics were named as paternity tests and genetic counselling, without any further explanation.

When institutes were established at universities, research completed the tasks of human geneticists. German scientists were involved in discovering genetically determined diseases like Pätau Syndrome (trisomy 13) or Becker Muscular Dystrophy.

8.1 Paternity Tests

After the end of World War II in Germany (Deutsches Reich), the situation for women was difficult. Some of them lost their husbands or did not know where they were till 1955. In this year, the last prisoners of war returned to their homeland or

[104] Weingart et al. 1988.

[105] See Petermann 2012.

home towns (Rückkehr der Zehntausend). On the other hand, there were a lot of soldiers, German and those of the occupying power, and there was the threat of rape by those.[106] These facts might be an explanation why paternity tests were the first jobs for human geneticists. But also institutes for forensic medicine did these.

The pathologist Karl Landsteiner (1868–1943) had identified in 1900 und 1901 the blood groups of man. Soon the discussion on heredity of those began. The physician Fritz (Friedrich) Schiff (1889–1940) caused a stir of interest of forensic scientists to use the heredity of blood groups for paternity testing. Since 1926, this method was established in the Deutsches Reich, when Schiff published his basic book on this topic.[107] The first verdict based on blood group testing was in November 1927.[108] Included in handbooks for forensic medicine[109] and by the German health council (Reichsgesundheitsrat) in 1930[110] this method was accepted. During the Third Reich, it was used for proving the Aryan ancestry. But this method was still available and proven after 1945 and could be used by the institutes of human genetics or forensic medicine. By different courts of law, paternity testing was advised and also paid for. At that time, cost bearing was an important factor.

8.2 Genetic Counselling

Since the beginning, the unsolved problem was the question of fees or reimbursement for genetic counselling up to the 1970s. Another aspect was that only physicians were given the admittance for medical advice according to genetic diagnosis.

From the historical point of view, genetic counselling can be seen in the tradition of the necessary given advice before 1945, because of the regulations of the Marriage health Law (*Ehegesundheitsgesetz*) of 1935. But the aim of counselling changed after 1945 from social to individual aspects.

The problems were at all time the same: despite all the increasing knowledge, there are still imponderables in interpreting the results of genetic diagnostic.

One reason for counselling was the fear that ionic radiation might have caused mutations. The nuclear bombs on Japan and the nuclear testing raised the question of the influence on the genetics of man. Therefore, human genetics gained the

[106] See Gebhardt 2015. The author made a projection of datas mostly based on those of cities. But these can not be taken for the countryside, where social control is more established.

[107] Schiff 1926. After his emigration, he published it in English: Blood grouping technique. A manual for clinicians, serologists, anthropologists, and students o legal and military medicine. New York: Interscience Publishers 1942.

[108] Geserick 2011, 41.

[109] E.g. Hofmann 1927.

[110] Geserick 2011, 42.

interest of public policy. In the German nuclear commission (*Deutsche Atomkommission*), there was a working group (VI/4) named radiobiology (Strahlenbiologie). Human geneticists were part of the discussion as guests. One topic was e.g. child examinations at school to get more information about diseases like e.g. haemophilia and muscle dystrophy.[111]

When prenatal diagnosis was introduced, starting in 1966 with Amniocentesis, genetic counselling was regarded necessary and paid by the insurances. In 1959, Jérome Lejeune (1926–1994) and colleagues had discovered that mongolism has three members of the 21st pair of chromosomes instead of two. With this, human genetics was brought to the attention of ordinary physicians, who were beginning to accept its necessity. The increasing knowledge about diseases helped to establish human genetics more and more.

In 1955, *Counseling in Medical Genetics* by biologist Sheldon C Reed (1910–2003) was published.[112] This book influenced the development in the FRG and Koch classified it as the first book on this topic. The German publication on genetic counselling was summarised in genetic family advice (Genetische Familienberatung) by Walter Fuhrmann (1924–1995) and Vogel in 1969. At this time, there were 15 institutes for this. In the following years, human geneticists of the FRG made further publications, but as a standard book Koch stated the one by Regine Witkowski on genetic and hereditary syndromes and congenital malformations (Genetik erblicher Syndrome und Missbildungen) and published in the GDR in 1974. This shows two things: genetic counselling became more and more a task of German human geneticists and in the science there was no iron curtain between the two parts of Germany.

At the beginning, the question of payment was unsolved and lack of money was characteristic. The main problem for the institutes for human genetics was to get paid for genetic counselling. The health insurers said that only physicians could bill those and not the anthropologists. They pay in general for medical services and those must strictly be performed by doctors. In Freiburg already in 1952, the anthropologists were also no longer allowed to make paternity testing.[113] This was one reason why the anthropologists lost their influence in the institutes for human genetics more and more. Also the statement of the German scientific council determined human genetics as a field of the medical faculty.

The federal government of FRG initiated a model test for genetic counselling in Frankfurt and Marburg and stated that the necessity of genetic advice centres was recognised.[114] Reasons for counselling were epilepsy, schizophrenia, spina bifida,

[111]UAH: Acc. 5/02: Meeting of May 6, 1959, in Bonn-Bad Godesberg at the federal ministry for nuclear energy and water management (Bundesministerium für Atomkernenergie und Wasserwirtschaft, BMAt).

[112]Reed 1955.

[113]UAF, B0124: 14.01.1952.

[114]Bundesministerium 1979.—"Die Notwendigkeit der Einrichtung genetischer Beratungsstellen ist erkannt worden."

Chorea Huntington as well as older couples, intermarriage and general consultation before marriage. In the period from 1972 to 1976, there were 3.387 consultations in Frankfurt and 1.390 in Marburg. For comparison, in Erlangen there were 1.078 consultations in the period from 1966 to 1976 with increasing number since the institute was paid for it.[115]

What might have been the consequences of the genetic counselling? At least, it was only to inform the mother of the unborn child or the couple. Up to the year 1975, abortion in Germany was only allowed for medical reasons that meant if there was a medical risk for the mother. This penal code (Strafgesetzbuch) of the German empire (Deutsches Reich) was set up in 1871.[116] Since then, in § 218 abortion was forbidden and a criminal act; no exceptions were made. In 1927, the Supreme Court (Reichsgericht) said that an abortion was possible for medical reasons. Nevertheless, abortion was a problem because of the economic situation of women.[117] In the first amendment of the Act to prevent unfit offspring (*Gesetz zur Verhütung erbkranken Nachwuchses*, GzVeN) of June 26, 1935, it is said in § 10: if the sterilisation of a women is in accordance with the law and she is pregnant, an abortion is allowed up to 6th month of pregnancy.[118] But, since March 1943 an abortion of Aryan women was sentenced with death. The GzVeN was not regarded as a national socialist law and therefore not abolished by the military government in 1948. On December 13, 2006, the German parliament (*Deutscher Bundestag*) passed the condemnation of this law and stated it as no longer valid.[119]

From a juridical view, it was clear that after 1945 § 218 was still valid in an unchanged manner: abortion was not at all allowed. Still accepted was abortion because of medical reasons, but there was an ongoing discussion. In 1971, the reform of § 218 started, when 371 women stated that they had an abortion.[120] In 1975, a law of changing § 218 was enacted and for the first-time results of genetic analysis were accepted for abortion. But, it said that abortion is a criminal act and punishable with imprisonment of three years or fine. But there were exceptions: abortion is not punishable, if there was a consultation more than three days before the procedure and no more than 12 weeks since the conception. Other reasons were because of criminal act (e.g. rape), medical ones and also eugenics or embryopathic reasons before the 20th week of pregnancy. This law was valid until the German unification in 1991. Therefore, abortion was only allowed since 1975 in the Federal Republic of Germany.

[115]Koch 1977.

[116]Reichs-Gesetzblatt (RGBl) 1871, No. 19 (08.05.): 95.

[117]Liepman 1927, 5. In 1926, there were 1.313.625 births but 875.750 abortions.

[118]Gütt 1936, 80.

[119]Deutscher Bundestag: Drucksache 16/3811: 13.12.2006. The law was not applied after 1945 any more.

[120]*Stern* 1971, Nr. 24: cover. (11.06.1971).

9 Conclusion

After 1945, the German human geneticists had tried to put their science into a positive light. This was relieved by the fact that eugenics was e.g. in Anglo-American and North European countries had no negative touch. In the focus was still the work on discovering the mechanism of heredity in general and especially of hereditary diseases.

Contrary to this, there was a break with regard to the institutes dealing with questions of heredity and human genetics. New institutes had to be founded.

But continuity was represented in the person of the scientists like Hans Nachtsheim, Otmar von Verschuer or Fritz Lenz, though their appointments were according to the existing law at that time. Because of the lack of scientists the universities had not really a choice. Today, as we know more about the facts of the Third Reich, those decisions might be analysed in a different way.

The scientific questions of heredity of characteristics and qualities started at the beginning of the twentieth century and still go on. In Germany, the main difference between human heredity before and of human genetics after 1945 is that the focus of interest changed from social or population aspects to individual interests: genetic counselling and prenatal testing gained more and more importance. There was continuity in science and no difference between the FRG and e.g. Anglo-American countries. Human genetics as a field of medicine was and is an applied and translational science. That human genetics could establish was very much depending on the political circumstances.

Changing the point of view means to look first at the contemporary conditions before judging them.

At the beginning, it was a question of good descent and best characteristics. With the increasing knowledge on hereditary and acquired qualities, the perception changed to illusions. The possibilities of human genetics and reproductive medicine offer new possibilities. The desire to produce a baby designed according to the wishes of the parents or to defeat diseases for a long life are the new challenges, as David Goldstein stated in 2011:

> There could be unexpected consequences if greater understanding of disease genetics gives parents more choice in what they pass to their children.[121]

[121] Goldstein 2011.

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F2/1, 1872c: Personal record Gerhard Koch
53, Nr. 29,1: Personal record Gerhard Koch
Sachakte Humangenetik (unsigniert)

UAF: University Archive Freiburg
B0124: Institut für Humangenetik und Anthropologie (1951–1999)

UAG: University Archive Göttingen
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UAH: University Archive Heidelberg
Acc. 5/01: stock human genetics
Acc. 5/02: estate Friedrich Vogel (114 files, partly with blocking period)

UAM: University Archive Münster
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Herbert Bach (1926–1996): One of the Pioneers of Human Genetics in East Germany (GDR)

Jörg Pittelkow

Abstract Herbert Bach was one of the initiators of the human genetic counselling service in East Germany (GDR). He believed in the causal connection between anthropology and genetics. As director of the Institute of Anthropology at the Friedrich-Schiller-University Jena, he established human genetics with his major parts in teaching, diagnosis and genetic counselling in the 1960s. In 1974, one of the first offices of genetic counselling was opened here. So the institute was a member of the cooperative project of the Ministry of Health to established human genetics in the GDR. Bach's special orders were the making of concepts for implementation and the coordination of the genetic counselling service. In 1978, he also became the chief of the Centre of the Genetic Counselling Service of the GDR. In addition, Bach had a great influence of the ethical discourse about human genetics.

Keywords Herbert Bach • Anthropology • Genetic counselling • Jena • GDR

1 Introduction

In 1966, a national expert group for research planning came to the conclusion that human genetics should become one of the main future research projects in the German Democratic Republic (GDR).[1] The very next year, the Minister of Public Health started a programme to establish human genetics as a distinct field of study. At the Academy of Sciences of the GDR, an interdisciplinary research group for human genetics ("Forschungsgemeinschaft Humangenetik") was founded. This

[1]For a general view of human genetics in the GDR, see Weisemann et al. 1997.

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research group was the origin of the later project to expand human genetics by the Ministry of Public Health ("Humangenetik-Projekt"). Although human genetics was established late as an official scientific discipline, it did not start out of nothing. In various departments of research and teaching, human genetics was an important subject, such as medicine (e.g., Alwin Knapp (1918–1995) in Greifswald, Regine Witkowski (b. 1934) in Berlin, Bernhard Wittwer (1936–1989) in Magdeburg), serology (e.g., Otto Prokop (1921–2009) in Berlin), genetics (e.g., Elisabeth Günther (b. 1925) in Greifswald, Paula Hertwig (1889–1983) in Halle, Hans-Albrecht Freye (1923–1994) in Halle, Rudolf Hagemann (b. 1931) in Halle, Jörg Schöneich (b. 1934) in Gatersleben) as well as physical anthropology (e.g., Herbert Bach (1926–1996) in Jena, Hans Grimm (1910–1995) in Berlin). Schulz called these hidden activities human genetics in the underground.[2] One of these workers in the underground was Herbert Bach, an anthropologist and biologist at the University of Jena.

2 Biographical Profile

Herbert Bach was born in the Thuringian town Gotha on 14 March 1926. After attending school, learning a technical profession and participating in the Second World War, he studied biology at the Friedrich-Schiller-University in Jena from 1948 until 1952. Around that time, the discussion about Lysenko's doctrine was intensifying. In Jena, the biologists and geneticists Otto Renner (1883–1960), Jürgen Harms (1885–1956) and Hans Wartenberg (1900–1972) were in direct confrontation with Georg Schneider (1909–1970), one of the most faithful defenders of Lysenkoism in the GDR. The debate deeply influenced Bach's scientific belief and his love for genetics. After graduating, he worked as a scientific assistant for few years, first at the Ernst-Haeckel-Haus (the Institute of History of Science) and then at the Institute of Anthropology and Ethnology. During this time, he was trained in the field of physical anthropology by Bernhard Struck (1888–1971). In 1957, he was awarded a doctorate, and he received his postdoctoral qualification (habilitation) in the field of physical anthropology in 1962. From 1960 to 1993, he served as the director of the institute. In 1963, Bach became an assistant professor for physical anthropology; in 1974, an adjunct professor; and in 1981, full professor for human genetics. In 1993, Uwe Claussen (1945–2008) succeeded him as director and in 1994 Bach retired (emeritus professor). He died in Jena on 12 July 1996 (Fig. 1).

Bach was an internationally well-respected scientist in the field of prehistorical anthropology and human genetics, especially regarding genetic counselling and ethical discourse. He renewed anthropological sciences in Jena by fostering prehistorical anthropology, e.g. reconstructing the biologic composition of historic populations, and by studying changes in biologic traits, e.g. through regular surveys in school children. He wanted to discover the (genetic) causes of human

[2] Schulz 2007, 1289.

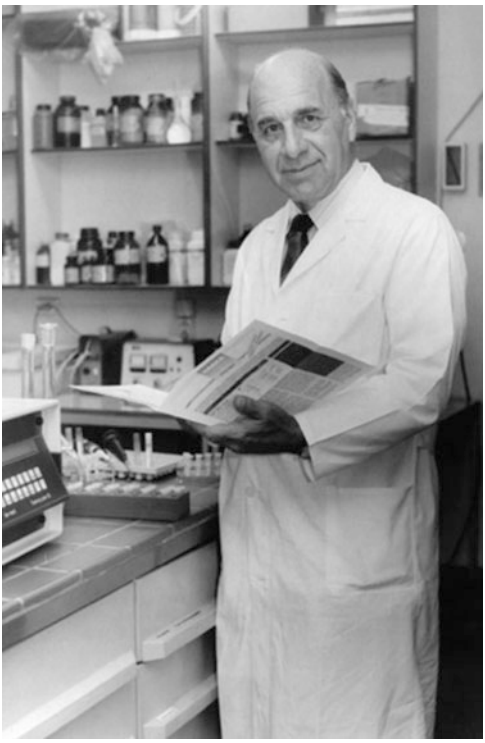

Fig. 1 Herbert Bach in the laboratory, 1993 (With permission of Anne Günther, Friedrich-Schiller-Universität Jena)

biodiversity by exploring the nature-nurture interaction. All anthropological research was based on the theorem of population genetics. Bach regarded physical anthropology and human genetics as associated sciences, as two sides of the same coin. While anthropology studied the non-pathologic variability of man, human genetics are investigated the causal determination of differences and the evolution of somatic characteristics.

For human genetics in the GDR, Bach notably founded the first counselling centre and coordinated the implementation of a genetic counselling service. In addition, he fought for formalised guidelines on counselling and popularised the benefits of counselling.[3]

2.1 Development of Human Genetics in Jena

Two anthropological institutes existed within the GDR: first institute, founded in 1930, at the university in Jena and a second institute at the Humboldt University in

[3]For an overview on life and work of Bach, see Pittelkow 2015.

Fig. 2 The former Institute of Anthropology and Human Genetics, today Institute of Human Genetics (Photo: Jörg Pittelkow)

Berlin, founded in 1955. A department of anthropology existed at the German Academy of Physical Culture and Sports (Deutsche Hochschule für Körperkultur und Sport) in Leipzig. Anthropologists were employed at varying medical institutions and prehistoric museums. Focus of research was prehistoric anthropology and anthropometric studies. In contrast to other nations, there were no scientific racial studies within the GDR (Fig. 2).[4]

As director of the Institute of Anthropology and Ethnology since 1960, Bach rebuilt the anthropological institute by shifting its focus from ethnology to genetics and expanding the scope of research and teaching. Staff levels were increased. The institute was to become a service provider to the medical department of the university with a laboratory for genetic and chromosomal analysis. In addition, he offered counselling on family planning. From 1964 onwards, the archival records of the institute document the topic of human genetics, including lectures on human genetics as well as research on the heritability of any somatic characteristics. The institute participated in the central researching group for human genetics with studies about heredity of phenotypes.[5]

These decisions resulted both from recent developments within the scientific policy of the GDR and as well as from academic considerations.

[4]Designations for the institute in Jena varied: Institute of Social Anthropology (1930–1936), Institute of Anthropology and Ethnology (1936–1969), Institute of Anthropology (1969–1974) and Institute of Anthropology and Human Genetics (1974–1993).

[5]See Universitätsarchiv Jena, Best. BC, Nr. 123, Best. N, Nr. 173, Best. BC, Nr. 150.

Concluding with the "Dritte Hochschulreform", reforms demanded an explicit commitment of all scientific institutions towards economic objectives.[6]

As a consequence of the work of Eugen Fischer (1874–1967) during the beginning of the twentieth century, research on human genetics had already been established as a field of inquiry within physical anthropology. Bach followed the growing tendency of closer structural and institutional fusion of anthropology and human genetics. In the FRG, a similar development occurred, e.g. in Munich by Karl Saller (1902–1969).[7]

An essential step to realise the idea of connecting both sciences in Jena and to comply with political demands was the beginning of the cooperation with the clinical department. In 1967, together with Wolfgang Plenert (1921–2000) and Niels Sönnichsen (b. 1930), two distinguished physicians of the university, Bach proposed a concept to establish human genetics as one of the major venues of research in Jena. This concept was accepted by university leadership and realised at the end of the 1960s. As a result, the institute is part of the department of medicine from 1968 until today.[8]

In 1971, the Ministry of Public Health formally started the "Humangenetik-Projekt". The ministry demanded application-oriented research under low ideological influence. In contrast to culture and education, public health was mainly based on hard research, because it aimed at an optimised health care for the populace. It was affected by economic circumstances and the state of application-oriented medical, biological and pharmaceutical knowledge. Within the latter, even persons that were not members of the communist party (SED) could hold positions of influence.

One of its members was the anthropological institute in Jena. The main task was to implement a national genetic counselling service. It addressed logistic considerations and improvements of diagnostics. The institute in Jena was involved in two fundamental ways: the installation of the counselling network and research about chromosomal diagnostics. The latter included amongst others a diagnostic key for Down Syndrome, procedures for prenatal diagnosis and automated analysis of chromosomes.[9]

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[6]Goals of the so-called *Dritte Hochschulreform* during the late 1960s were an effective, application-oriented research and the affirmation of SED influence on universities by curbing the last vestiges of academic democracy. Traditional faculty structure was abolished and reorganised into specialised departments (Sektionen), e.g. Sektion Biologie or Sektion Physik). University leadership was reduced to two levels, the university rector and department directors. Each university was assigned a main focus in research and teaching (e.g. physics, technology and medicine in Jena). For an overview, see Stutz 2007.

[7]For the history of anthropology and their relations to human genetics, see Hoßfeld 2016, Schwidetzky 1988 and Ziegelmayer 1987.

[8]See Universitätsarchiv Jena, Best. BC, Nr. 100.

[9]See the planning documents of the human genetics project (Bundesarchiv Berlin, DQ 1/11043 and DQ 1/11043) and the yearly reports on the activities of the Institute of Anthropology and Human Genetics (Universitätsarchiv Jena, Best. S/II, Nr. 281–283, 286 and 297). Also see Bach et al. 1969 and 1979.

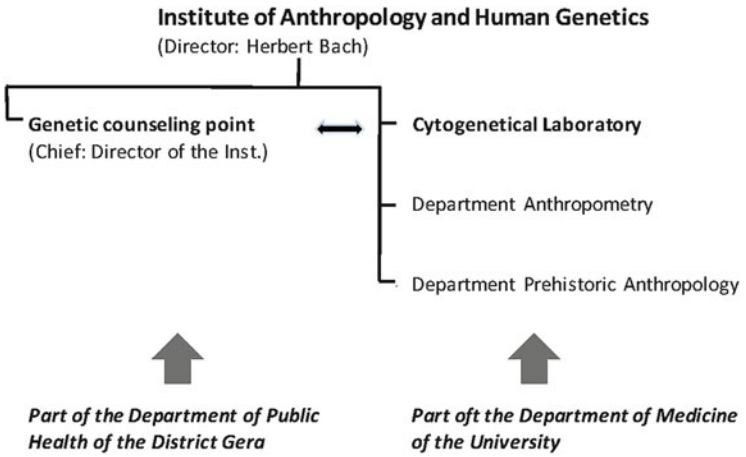

Fig. 3 Structure of the institute and the counselling centre

2.2 The Genetic Counselling Service Centre at Jena

The directives of the anthropological institute for the second half of the 1960s demanded a provision of genetic counselling. The strategy for 1968 made a proposal to install a counselling centre in conjunction with the Department of Public Health of the District of Gera. In October 1971, Bach presented a comprehensive concept for such a "joint venture" institution. Financial reasons and ongoing debates about management and funding delayed the implementation of the idea. However, on 8 January 1974, the district physician and the director of the department of medicine of the university finally signed the contract about the counselling centre. Specifically, it addressed: The centre should be a part of the public health-care system of the District of Gera. It was managed by the director of the anthropological institute and supervised by the district physician. The full-time staff were public employees. The institute provided space and laboratory analysis. Two positions were government financed and two and a half by the university. In 1982, the counselling centre became a full part of the department of medicine of the university. On 20 February 1974, Bach informed the university management that the counselling centre was active since 1 January 1974, because the geneticists of the institute were providing counselling for quite some time. Additionally, he discussed the possibility of counselling via media (Fig. 3).[10]

In summary, genetic counselling was classified as highly specialised medical service. This service included a comprehensive counselling interview encompassing both the concerns of the counselee and an analysis of the familial background. If it was necessary, prenatal diagnosis was provided. The counselees came of their own accord or by medical referral. A major aim was the assessment of

[10]Universitätsarchiv Jena, Best. S/II, Nr. 219, Best. BC, Nr.150, Best. L, Nr. 714, Best. S/II, Nr. 17.

the risk of hereditary diseases in the context of family planning or during pregnancy.[11]

2.3 Coordinating the Genetic Counselling Service in the GDR

The experiences made in Jena and Magdeburg, the place of the second counselling centre, were providing a basis of the proposal for installing the national counselling system. Bach exerted considerable influence on its design and implementation.[12] The international state of research and medical care as well as the equivalent level and potential in the GDR were analysed and compared. The research objects, the equipment of the counselling centres and the demand of advanced training for the staff were based on this analysis. The general conclusion was genetic counselling is a new kind of highly specialised prophylaxis outside of the traditional structures of the public health. The following points were proposed:

- The counselling centres should be integrated in the medical departments of universities, in medical academies or in central hospitals of the districts.
- In every district of the GDR, a counselling centre including the necessary laboratories should be established. These individual units should form a network, which is coordinated centrally.
- The duties of the centres are:
  - Determination and interpretation of the clinical (genetic) evidence
  - Recommendation for further diagnosis and therapy
  - Registration and counselling of families with an increased risk of hereditary diseases
- The array of methods should include the diagnosis of chromosomal aberrations and metabolic defects, prenatal diagnosis as well as (genetic) screening to identify mucoviscidosis or phenylketonuria and other genetic defects. It should provide tools to scan for mutagenic substances in the environment.

The Ministry of Public Health decided the agenda "Human Genetics Counselling System" in 1977. In form and content, the minister acted on the suggestion of Bach and the other geneticists. Following this decision, the district counselling centres

[11] See Janitzky 1990.

[12] See "Komplexes Überführungsprogramm Humangentischer Beratungsdienst", Bundesarchiv Berlin, DQ 1/26482, Teil 2, "Konzeption zur schrittweisen Einführung der genomischen Diagnostik in die humangenetische Forschung und hochspezialiierte Beratung", Bundesarchiv Berlin, DQ 1/26482, Teil 1 and "Analyse ´Personelle, materialle und organisatorische Voraussetzungen zur Überwindung der Uneinheitlichkeit des Auf- und Ausbaus der Humangenetischen Beratung in den Bezirken´", Bundesarchiv Berlin, DQ 1/26482, Teil 1. See also Bach o. J., 1983, 1984/85, 1986 and Steinbicker 1977.

were founded. The centre in Jena additionally played the role of a "headquarter", called "Genetic Counselling Centre of the GDR ("Humangenetisches Beratungszentrum der DDR"), under the direction of Herbert Bach. This was also a compliment to the performance of the team in Jena.

In the middle of the 1980s, the counselling service was completed and operated at a comparable level to other states in the East and West. The ministry of health supported any activities to establish innovative methods, e.g. for genomic and especially prenatal diagnosis. In the wake of the international "Genome Project" that began in the USA, the ministry looked at the possibility of international collaboration. In this situation, Bach demanded to pool the capacity of laboratories and to fund highly specialised laboratories. Hindrances were, as always, the lack of material and staff.

During the implementation of the counselling system, various activities were initiated to give public and professional information about its structure and benefits. In Bach's eyes, the initial acceptance of the genetic counselling by physicians and in the general population was alarmingly low. In consequence, he considered the potential, the benefits and also the limits of genetic counselling even more thoroughly. In 1974, he organised an international meeting about the problems of counselling in Mühlhausen/Thuringia.[13] It raised the recognition of human genetics and initiated the founding of the East German scientific Society of Human Genetics ("Gesellschaft für Humangenetik"). One of the tasks of this society was the professional training in genetics. Another result of the conference was an increased involvement of politicians in this development.[14] The congress' influence on the development of genetic counselling and medical genetics was comparable to that of the Forum Philippinum "Genetik und Gesellschaft" (Genetics and Society) at Marburg in 1969.[15]

3 The Headquarter of the National Counselling System

In 1981, Bach assumed the role of the leading manager of the counselling system in the GDR.[16] His major task was to secure a comparably high level of counselling at all centres. The following tasks were determined[17]:

– Coordinating collaboration and research

[13]For the conference in Mühlhausen, see Bach 1975a.

[14]In 1976, the party platform of the SED identified and explicitly promoted human genetics as an important field of research.

[15]For the conference in Marburg, see Wendt 1970.

[16]See Bundesarchiv Berlin, DQ 1/13732.

[17]See Bundesarchiv Berlin, DQ 1/13732 und DQ 1/26482, Teil 1 und 2; Universitätsarchiv Jena, Best. S/II, Nr. 409 & 416.

– Organising periodic working meetings with the staff of the centres
– Giving information about international trends and editing readers
– Giving expert opinion, organising staff training and overseeing the professional registry

One of Bach's duties was the regular inspection of all counselling centres and laboratories. The visits resulted in a list of demands to the Ministry of Public Health. He demanded more staff, adequate equipment, microscopes, international literature, chemical supplies, ultrasonic devices, enlarged facilities and also more publicity of the benefits of counselling. He advised not to establish multiple counselling centres and laboratories in the same town.[18]

3.1 Ethics of Genetic Counselling

One of the tasks of the human genetics project was to start an ethical discourse to find an answer to some essential questions, such as:

– Is genetic counselling a kind of official eugenics?
– Who should specify the extent of counselling and diagnosis?
– What is the relationship between the personal autonomy of the counselee and the medical responsibility of the counsellor?
– How to secure the privacy of counselling data?

The progress of the US Genome Project raised new and serious questions also in other various countries. The geneticists were afraid of a so-called eugenics from below.[19] Uwe Körner (1988) and Bach (1990a, b, 1990/91) identified an undue expectation that human genetics, acting in combination with diagnosis and induced abortion, could allow for perfectly planned children.

The argument distinguished between an "ethics of counselling" and an "ethics of decision". The former encompassed the responsibility of the counsellor regarding scope and extent of counselling in recognition of both situation and personality of the client as well as the risk of individual procedures. The latter covered the recommendation given by the counsellor and the final decision by the client. Opinions were mixed, especially regarding the subject of abortion.

Bach's opinions were substantial arguments in the discourse. His view was based on science and not on Marxian philosophy or ideology. In interdisciplinary discussion as well as scientific literature, Bach constituted an explicit authority; even for Marxian academics insofar, they were engaged in ethical questions

[18]See Bundesarchiv Berlin, DQ 1/26482, Teil 1.

[19]This refers to the warning made by Bishop 1996 that genetic dispositions could lead to exclusions from insurance or employment. The term "eugenics" was used very unevenly within the GDR and, in contrast to the Soviet Union, fell gradually out of use. There was a universal rejection of direct governmental influence.

regarding genetic counselling.[20] The central point of Bach's thinking was the person and his claim to live and to be successful—but also in the clash of the interests of the parents and the unborn, potentially disabled child. He postulated that, in general, genetic counselling is individual preparedness without obligation to decision. He strictly dismissed eugenic ideas.[21]

Bach developed his ethical principles by active counselling, public discourse and his struggle for obligatory standards of counselling. At the end of the 1980s, an acceptable state was achieved. In 1990, the standards were published under the title "Orientierung für die humangenetische Beratung", first in West Germany. They included tasks, targets and principles of counselling, indications and conditions as well as the documentation of diagnostic findings. The authors demanded an especially sensitive use of the quickly evolving potential of genetics and consideration of the implications for the individual and society.

4 Conclusion

Even though the first counselling centre of the GDR was established in Jena, the development of the field of human genetics and of a distributed counselling service in the GDR was the result of a focused if sometimes uneasy collaboration of diverse scientific and medical institutions with the Ministry of Public Health. The process was substantially shaped by the individuals involved. Bach excelled in organising academic activity and as an important mediator within human genetics. As in research and in teaching, he argued from a scientific medical position and contributed to an objective-/fact-based ethical discourse.

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[20] A good example for this is Dietl 1977.

[21] See f.e. Bach 1974, 1975 and 1990a.

The introductory words of the article are clear in their ethical objective: "Die vielfältigen Aspekte der Humangenetik erfordern einen besonders sensiblen Umgang mit den sich rasch entwickelnden Möglichkeiten der Genetik und den sich daraus ergebenden Konsequenzen für den einzelnen und die Gesellschaft. Die vorliegende "Orientierung" soll auf der Grundlage des gegenwärtigen Erkenntnisstandes allen an der humangenetischen Betreuung Beteiligten eine grundsätzliche Hilfe für die verantwortungsbewußte Erfüllung ihrer Aufgabe sein und dazu beitragen, daß Fehlentwicklungen, die dem Wohl der von genetisch bedingten Problemen Betroffenen entgegenstehen, möglichst vermieden werden" (Bach et al. 1991: 1077).

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Concise History of Prenatal Diagnostic Service in Russia

Vladislav S. Baranov

Abstract Evolution of *prenatal diagnostic service* (PDS) in Russia is briefly outlined. It has started in the middle of 1960s, when it was mainly treated as a part of medical genetic counselling (period 1). In 1993 PDS was officially approved by the *Federal Ministry of Health Care* as a new clinical service. For many years, 1993–2000 (period 2), the infrastructure of PDS, as a part of medical genetics, corresponded to territorial and administrative structures, including 72 local, 16 regional and 7 federal medical genetic centres. Each of those had specific duties including medical genetic counselling of pregnant women, their biochemical and ultrasound screening, foetal tissue sampling and laboratory analysis. The main goals of PDS at the beginning included elaboration of screening programmes and the mastering of new laboratory methods. At this period, major improvements in PDS concerned foetal examination with ultrasound (US) and foetal tissue sampling. Original chromosome preparations from chorionic villi provided high efficacy of prenatal karyotyping. Methods for molecular diagnostics of severe monogenic disorders as well as biochemical testing of embryonic protein markers in maternal blood at the first and second trimesters have been applied since the early 1990s. Conspicuous contributions of scientific and clinical staffs from Saint Petersburg in elaboration of diagnostic and screening methods paving the way to PDS in Russia are emphasized. The third period could be attributed to the federal law in 2000 with its order on prenatal US testing of all pregnant women on the 11–14, 18–21 and 31–34 weeks of gestation (w.g.). The next major step corresponds to the transition of PDS from the second to the first trimester of pregnancy. This decisive shift should be attributed to a new early prenatal or combined screening (EPS/CS) initiative approved by the *Ministry of Health Care* in 2010. EPS relies on combined US and biochemical screenings supplemented with automated risk assessment of

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foetal chromosomal disorders in the women on their 11–14 w.g. The efficacy of EPS/CS has been repeatedly proven within the next few years in many prenatal centres and becomes a mainstream of PDS in Russia. Meanwhile, starting from 2012, implication of new highly productive molecular and molecular cytogenetic methods of PDS has progressively increased. Though rather efficient at present, PDS needs further modifications stemming from array CGH and non-invasive prenatal diagnosis (NIPD). Necessity for elaboration of some sophisticated compromise between EPS and NIPT in Russia is now very urgent.

Keywords Prenatal diagnostic periods • Russia • New technologies

1 Introduction

Huge territory inhabited by 149 million people belonging to almost 130 different nations and ethnic groups with two million newborns annually creates many serious problems on its way to properly balanced and efficiently regulated *prenatal diagnostic service* (PDS) in Russia. Nonetheless, some conspicuous achievements and unambiguous positive trends in this quickly expanded area of medical service are now quite obvious. To my knowledge the history of PDS in Russia has never been reviewed. It was reflected in rather fragmentary and scanty report just within the country saying nothing to English-speaking people abroad. Therefore this review should be treated as the first attempt to fill this annoying gap.

2 Five Periods of PDS History in the USSR and the Russian Federation

As a chief of laboratory for prenatal diagnosis of inherited and inborn disorders at FSBI "The Research Institute of Obstetrics, Gynaecology and Reproductology named after D. O. Ott", involved in PDS for almost 30 years, this provided me with an ample opportunity to trace the history of native PDS, which started almost 40 years ago in the former Soviet Union (USSR) and now makes progress in modern Russia. The USSR lasted till 1991, and then the *Russian Federation* was established.

With the focus at PDS history of Russia, five poorly confined periods can be distinguished. Each is full of curious cases, famous personalities and interesting adventures, but these should be omitted in favour of more objective consideration of major events corresponding to the principal steps of PDS evolution in our country. Numbers of these periods and their tentative titles are given in Table 1.

Table 1 Basic periods of the prenatal diagnostic service (PDS) in Russia

BN	Periods	Title	Brief description
I	1969–1993	Initiation	Medical genetic recovery and PDS initiation
II	1993–2000	Methods, tools	Elaboration and implementation of new technologies
III	2000–2010	Active invasion	Alliance of medical genetics and obstetrics
IV	2010–2015	Combined screening	Early prenatal screening programme
V	2015–till now	New technologies	Molecular invasion into PDS

3 Medical Genetics as a Scientific and Practical Background of PDS

Prevention of inherited and inborn disorders remains a principal objective of medical genetics. Before the PDS era, medical genetic counselling of high-risk families with inherited and inborn disorders was a basic means of prediction and prevention of genetic disorders. Reproductive genetic counselling played a significant role in prevention of chromosomal disorders as well. Three feasible ways to prevent inherited disorders are tried. They include preconception genetic counselling (I), prenatal diagnostics (PD) (II) and phenotype corrections (normocopy) of affected newborns (III). Evaluation of inherited risk is a principal goal of preconception counselling. The second one (PD) got its start in Russia at the 1990s being supported by ultrasound, invasive sampling and available laboratory tests. Prevention of clinical manifestation of inherited or inborn disorder is the main aim of the third root. Thus born by medical genetics, PDS should be always treated as one of its most efficient practical branches devoted to prevention of inherited and inborn disorders.

The awful tragedy of Russian genetics in the mid-1930s caused by devastating activity of Lysenko lasted in Russia for decades and completely ruined genetic science and also medical genetics. Fortunately, this unbearable situation came to the end in the mid-1960s. Owing to many prominent human geneticists of the former USSR such as professors Sergey N. Davidenkov (1880–1961), Vladimir P. Efroimson (1908–1989), Alexandra A. Prokofieva-Belgovskaya (1903–1984), Solomon A. Neyfakh (1909–1992) and Helene E. Pogosyanz (1914–1990), the medical genetics survived and gave a rise to PDS later.

Gradual recovery of genetics occurred in 1961 when the first new medical genetic laboratory appeared in Leningrad (today Saint Petersburg), initiated by the member of *USSR Academy of Medical Sciences* S.N. Davidenkov (1880–1961) and his wife—well-known geneticist—Eugenia F. Davidenkov (1902–1996). In 1969, the first *Institute of Medical Genetics* was founded in Moscow with prominent geneticist and member of *USSR Academy of Medical Sciences* Prof. Nikolay P. Bochkov (1931–2011) as its head.

In line with medical genetics in the USSR, PDS at that time had three levels of organization with about 84 local medical genetic centres (LMGC) distributed throughout the country affiliated in paediatric and outpatient clinics. They were responsible for the initial first level of PDS including primary genetic counselling and ultrasonic examination. The second level belonged to 16 regional medical genetic centres (RMGC) responsible for all kinds of PDS including biochemical screening of pregnant women and foetal karyotyping. The PDS pyramid was crowned by seven federal medical genetic centres affiliated in main medical genetic and obstetric institutes of Moscow, Saint Petersburg (former Leningrad) and Tomsk. Their activity included molecular diagnostics and elaboration of new methods of mutation detection and allelic polymorphism studies in the causative genes involved in severe genetic disorders. Detailed cytogenetic analysis of complex chromosomal rearrangements was also included. More details concerning the history of molecular, cytogenetic and biochemical methods in PDS of the USSR are given in the next section.

It should be mentioned that even on these early days, some minimal set of PDS was already carried out and regulated by the order of the *Federal Ministry of Health Care*. One of them (no. 1120 on 31.10.1979) postulated organization of 80 local MG counselling services (LMGCS) in the USSR with 43 of them in Russia itself. It has also declared the organization of three basic centres responsible for supervision and governing of all medical genetic service (MGS). One of them was the *centre for prenatal pathology in laboratory of medical genetics* at the *Institute of Mother and Child Care*, which played a prominent role in elaboration and implementation of original methods for invasive chorionic and placental villi sampling and subsequent chromosomal analysis of the foetus. The first successful chorionic villus sampling (CVS) supplemented with relevant chromosome studies was reported in 1980.[1] Simultaneously, ultrasound became a part of the national health-care system, and the oldest prenatal centre in Moscow was established in 1979.[2] The spreading of prenatal US testing in obstetrical clinics working in collaboration with LMGC or RMGC made a great impact in development of PDS in Russia. Official approval of PDS came in 1985 with an order no. 787 from the Federal Ministry of Health Care to the RMGC which were recommended to use non-invasive as well as invasive techniques including CVS, amniocentesis and even cordocentesis in PD of foetal chromosome disorders. Accepted as highly stimulating impulse, its practical application was however hardly possible at that time. Invasive sampling was tried only in a few clinics, and chromosome preparations of good quality could be made from cord blood lymphocytes only. Cytogenetic analysis of chorionic villi or cultured amniotic cells was still not efficient as no reliable methods were available so far. Nonetheless several hundreds of invasive prenatal diagnoses were performed by 1985 at the *Institute of Mother and Child Care* and *Institute of Medical Genetics* both in Moscow. Cytogenetic analysis for foetal sexing and chromosome disorder

[1] Rozovsky et al. 1980.

[2] Medvedev and Elena 1998.

studies as well as some biochemical analysis of amniotic fluid samples for the search of rare disease biomarkers and AFP test for neural tube defects (NTD) were tried. The onset of invasive prenatal diagnostics was also declared at the *Medical Genetics Institute* in Tomsk. First successful attempts of invasive prenatal diagnostics of chromosomal and common monogenic diseases (like cystic fibrosis, haemophilia A, phenylketonuria) were also reported at the end of 1980s from D.O. Ott's Institute of Obstetrics and Gynaecology in Saint Petersburg.[3]

It should be taken for granted that PDS as a part of health care in Russia appeared in the early 1990s. More precisely, its date of birth was December 30, 1993, which corresponded to the issue of the order no. 316: "To the further development of medical genetics service of Ministry of Health Care of Russian Federation". Mainly involved in genetic practise, the order is also devoted to detailed description of PDS facilities. According to its regulation, RMGC were selected as a principal part for PDS responsible for all basic activities of prenatal centres such as ultrasound testing, biochemical screening, foetal tissue sampling and karyotyping.[4] RMGC staff, its occupations and basic equipment were annotated. Up now in spite of many changes and amendments which will be surveyed later, the order no. 316 still remains a solid legislative background as a starting point of PDS in our country.

4 Elaboration and Implementation of New Technologies in 1990–2000

The main problem of PDS is its complexity. Its efficacy depends on precise and equilibrated activity of many specialists from different scientific and practical areas, such as medical genetics, obstetrics, ultrasound, biochemistry and cytogenetic and molecular biology. PDS stimulated coalescence of all these areas to provide efficient screening of pregnant women for risk of inherited and inborn disorders in their foetuses complemented with safe foetal tissue sampling at different stages of pregnancy as well as with quick and reliable diagnostic tools to manage different types of foetal pathology. Before and after prenatal diagnostics, genetic counselling is also necessary. According to the order no. 316, the basic components of PDS should include:

1. Primary medical genetic counselling of pregnant woman
2. US testing in the second trimester of pregnancy
3. Biochemical screening for AFP and HCG in the second trimester
4. Foetal sampling (chorionic or placenta biopsy, amniocentesis, cordocentesis)
5. Laboratory diagnostics (cytogenetic, molecular, biochemical)
6. Concluding medical genetic counselling on results of PD

[3] Baranov 1993.

[4] Baranov 1997.

All these items were already in use in a number of PDS centres throughout the world before 1990, but most of them needed serious revision and special adjustment to be operative in Russia. Brief histories of their implementation in PDS of Russia are included.

4.1 Medical Genetic Counselling (MGC)

Stemmed from clinical genetics, this division was the most advanced part of PDS in Russia. Meanwhile it was always different from routine genetic counselling. The specificity of the primary (before PD) and the secondary (after invasive prenatal diagnostics) MGC was repeatedly declared and substantially improved by Prof. Vladimir I. Ivanov (1932–2010) (member of Russian Academy of Medical Sciences) and Prof. Vera L. Izevskaya, both from the Institute of Medical Genetics in Moscow. Substantial impact in elaboration and specification of medical genetic counselling in PDS in Saint Petersburg has been made by Victor G. Vakharlovsky (1940–2010) then a collaborator of our laboratory. Legislative and ethical principles of MGC recommendations are regularly revised in line with the progress of options of PDS due to invasion of new methods in prenatal diagnostics.

4.2 Ultrasound Testing

Since the early 1960s, ultrasound examination has been the prerogative of physicians and remains the most popular non-invasive prenatal diagnostic method in our country so far. More than 500 sonologists work in the field of obstetrics and gynaecology. Each Russian administrative region has several outpatient clinics, maternity units and one or two prenatal centres. Before the millennium routine obstetric ultrasound was usually performed twice during pregnancy in outpatient clinics. Screening criteria were regulated by guidelines drawn up by the *Association of Ultrasound in Perinatology and Gynaecology* founded in 1987. According to known Russian authorities in prenatal US testing Prof. Michael V. Medvedev and Prof. Helen V. Ioudina (both from Moscow prenatal centre), more than 80% of pregnant women had at least one ultrasound scan during pregnancy. More than 90% of women were screened in the mid-1990s. Two levels of US screening were recommended.[5] The patient was first examined between 18 and 24 weeks of gestation to identify foetal malformations and ultrasound markers of chromosomal abnormalities. The second scan was carried out at 30–34 weeks of gestation to assess foetal well-being and to exclude intrauterine growth restriction. If an advanced level examination was necessary, the patient was referred to the regional

[5]Medvedev and Elena 1998.

prenatal centre. The analysis of 1247 cases of prenatal karyotyping performed in Moscow Regional Prenatal Center before 1998 demonstrated increased detection rate of gross chromosomal abnormalities (trisomy, triploidy, unbalanced translocations) from 2.2% in 1990 to 14.1% in a high-risk group.[6]

4.3 Biochemical Screening for Foetal Anomalies

AFP testing was initially tried in the middle of 1980s mainly for detection of foetal neural tube defects (NTD) at the *Institute of Mother and Child Care* (Moscow). In 1987, it was also tried in our lab in Saint Petersburg. Biochemical screening of maternal serum AFP by radioimmunoassay was initially applied almost simultaneously in the lab for prenatal diagnostics (Saint Petersburg) and medical genetic laboratory of Moscow.[7]

Double biochemical test (AFP + HCG (human chorionic gonadotropin)) was tried in 1993 by Victoria N. Gorbunova and Tatiana K. Kascheeva (Saint Petersburg). Firstly risk of foetal chromosome anomalies was evaluated by means of special age risk tables and supplemented with likelihood ratios of MOMs (multiples of the median) values for AFP and HCG. Original software for automatic Down's risk assessment in the second trimester foetuses have been elaborated, applied since 2000 and is still in use.[8] Total second trimester screening of pregnant women with double biochemical test (AFP + HCG) was launched in Saint Petersburg in 1997, when the first original ELISA test system elaborated by national private company became available.[9] The biochemical test initially included AFP and HCG but was occasionally supplemented with unconjugated estriol and inhibin A later in the 2000s. Pilot project of early (first trimester) combined screening for PAPP-A and free β-HCG coupled with US testing have been tried in Saint Petersburg since 2003.[10] High efficacy of this study has further stimulated the expanding of early combined screening throughout Russia. More about early prenatal screening (EPS) is given in Sect. 5. Contingent screening in first and second pregnancy trimesters supplemented with total risk calculation for the both according to original formula that has been used since 2008.[11] Over 30.000 pregnant women (50% over 35 ages) have been tested by combined *first trimester screening* with overall Down syndrome detection rate about 96% and false positive rate 7.4%.[12] Further progress in technology and the rate of biochemical testing of foetal proteins in maternal blood

[6]Medvedev and Elena 1998.

[7]Gorbunova et al. 1991, Dubinina and Irina 1990.

[8]Kostyuk et al. 1992.

[9]Vakharlovsky et al. 1995, Baranov 1997.

[10]Nekrasova et al. 2007.

[11]Kascheeva 2008.

[12]Kascheeva et al. 2010.

enabled short-term analysis and have provided a chance for OSCAR service (One-Stop Clinic for Assessment of Risk) in the D.O. Ott's Institute of Obstetrics, Gynaecology and Reproductology since 2013.

4.4 Foetal Tissue Sampling

As already mentioned the first CVS in Russia was performed in 1980. Since that it has been repeatedly carried out in many other clinics with variable success. Different ways of sampling included transvaginal (needle or forceps) or transabdominal routes. Unfortunately the risk of pregnancy loss after villi withdrawal was initially so high (up to 30%) that this operation was officially forbidden in the late 1990s by known genetic Prof. Nicolay P. Bochkov as a vice president of Academy of Medical Sciences at that time. Nonetheless experienced obstetricians proceeded foetal sampling under US control with gradual decrease of complications to 1–3%, with its highest value as 3% for foetal blood sampling by means of cordocentesis in the early 2000s. The first chorionic and cord blood sampling in Ott's Institute (Saint Petersburg) were carried out by Vladimir M. Lebedev and Anton V. Mikhailov, respectively, in 1988.

Transabdominal chorion and placenta biopsy on 10–19th w.g. was found the safest since 1989 and was widely used throughout the country so far. Amniocentesis is safe and also very common operation in many prenatal centres of Moscow and Tomsk regions. According to our data, the current risk of foetal sampling irrespective of invasion type implicated (including cordocentesis) is less than 1%. In view of gradual transition of PDS to the first trimester of pregnancy (see Sect. 4), the proportion of foetal cord blood sampling progressively decreases and is officially considered as a drawback of PDS.

4.5 Laboratory Diagnostics

No regular prenatal diagnostic laboratory service existed in Russia before 1990. Some sporadic cases of invasive PD with application of cytogenetic, molecular or biochemical techniques were occasionally reported before the mid of 1980s (Sect. 2).

4.6 English Trace

In 1985 by lucky chance, the review's author as WHO (World Health Organization) student had training courses in the UK and succeeded to visit many advanced scientific and prenatal diagnostic centres in London, Glasgow, Edinburgh, Oxford

and Cardiff. During the visit I had a privilege to be acquainted with many outstanding British scientists such as Malcolm Ferguson-Smith, Anne McLaren, John Edwards, Peter Harper, Kay Davies, Tony Monaco, Bob Williamson, David Brock, Martin Bobrow and others. Besides very stimulating discussions, I was generously gifted with many original DNA probes—markers of relevant genes which mutations resulted in such inherited disorders as cystic fibrosis, haemophilia A and B, Duchenne muscle dystrophy, phenylketonuria, etc. The molecular technologies including Southern's blot and RFLP analysis for molecular genetic diagnosis were also provided. Valuable information on prenatal testing of foetal AFP both in maternal blood and amniotic fluid for prenatal detection of neural tube defects in the foetus as well as technology of foetal intestine microvilli enzymes study for biochemical testing of cystic fibrosis were provided as well. Real value of these probes and technologies has become evident and highly appreciated within a couple years in the first Russian laboratory of prenatal diagnostics of inherited and inborn disorders founded in *Ott's Institute of Obstetrics and Gynaecology* Russian Academy of Medical Sciences in Saint Petersburg in 1987. We will come back to details of this topic further, but substantial impact of above-mentioned UK scientists in the history and advance of PDS in Russia especially in prenatal molecular diagnostics of common genetic disorders should be always recognized with gratitude. Figure 1 shows the group of the scientists from the laboratory of Ott's Institute whose professional abilities and administrative gifts contributed a lot into development of PDS in Russia.

It should be also mentioned that from its start the activity of our lab was running in a close professional collaboration with municipal medical genetic centre (RMGS), founded in 1961 by academician S.N. Davidenkov as the first medical genetic laboratory (see Sect. 2). This unique alliance between Federal Scientific Institute named after Ott and municipal medical genetic center (RMGS), governed by Prof. Olga P. Romanenko, turned to be exceptionally fruitful. New methods and technologies elaborated at the laboratory for prenatal diagnostics were transmitted quickly and tested in the RMGS.

4.7 Cytogenetic Studies

First attempts of sexing and karyotyping of intrauterine foetus in Russia were undertaken in the early 1970s at the Institute of Mother and Child Care and also at the Institute of Medical Genetics, Moscow.[13] Both institutes are also known as the centres where first chorionic and amniotic cells were cultured for chromosome preparations and prenatal diagnostics of chromosomal and genetic disorders were performed.[14] Limited access to US equipment of proper quality, shortage of

[13]Zolotukhina 1972, Bakharev 1976.

[14]Zolotukhina 1980, Tsvetkova et al. 1983.

Fig. 1 The staff of the laboratory for prenatal diagnostics of inherited inborn disorders founded on 5 May 1987 in the Ott's Institute of Obstetrics, Gynecology and Reproductology, Saint Petersburg (Russia). *First row* (*left* to *right*): Tatyana Kascheeva (Dr. Sci.), well known in biochemical Screening; Tatyana Ivaschenko (professor), major impact in molecular diagnostics including Prenatal; Victoria Gorbunova (professor), initiator of biochemical and molecular studies; Tatyana Kuznetzova (Dr. Sci.), great contribution into cytogenetic studies. *Second row* (*left* to *right*): Michael Aseev (PhD), the first molecular PD of haemophilia A in 1989; Vladislav S. Baranov (Corresp. member of Russian Academy of stress out Sciences), the chief of the lab since 1987 till now (Photo: Vladislav S. Baranov)

specialists in chorionic and cord blood sampling, high risk of foetal loss after sampling at its start, the problems with cell culture mediums for amniotic cell growth as well as with chromosome staining from chorionic villi (CV) cells significantly hampered progression and spreading of PDS throughout the country. Conspicuous impact into foetal karyotyping was made almost simultaneously in Moscow and Saint Petersburg at the end of the 1980s. Short-term culture technique was elaborated and successfully tested at the Institute of Medical Genetics by the group of experienced cytogeneticist Prof. Tatyana V. Zolotukhina.[15] Fast direct method of chromosome preparations from chorionic villi was born in our laboratory. Suggested method was actually a modification of the original technique used for many years in experiments with laboratory mice at the embryology department of Institute of Experimental Medicine (Saint Petersburg).[16] This reliable and cheap method also known as "shaking-blotting technique" was adopted for prenatal karyotyping of chorionic and placenta villi samples.[17] The method gained much more credit when relevant chromosome preparations were treated by modified Hoechst 33258/actinomycin D method elaborated by Prof. Tatyana

[15]Zolotukhina 1988.

[16]Dyban and Vladislav 1987.

[17]Baranov 1989, Baranov et al. 1990a, 1990b.

V. Kuznetzova. This fluorescent staining resulted in a very clear chromosome banding and made possible reliable identification all human chromosomes and their rearrangements. The methods provided 99% successful rate of CV cell karyotyping on direct and semi-direct slides at the 9th through 20th weeks of gestation with average level of banding 400–500 bphs (bands per haploid set).[18] With some minor modifications, the same method could be adjusted for chromosome studies of any embryonic tissue containing dividing cells.[19] Since the early 1990s and up to now, the method is still in use in many prenatal centres throughout Russia. In 1995 it has been officially approved for prenatal diagnostics of chromosomal anomalies in the first and the second trimesters of pregnancy.[20] Over 12 thousand prenatal karyotypes prepared for almost 30 years by means of shaking-blotting technique only in our lab have unanimously proved its high efficacy in prenatal diagnostics of chromosome disorders. Comparing to short-term culturing of chorionic cells, the direct method has some obvious advantages. Coupled with cord blood lymphocyte and amniotic cell cultures, this method covers all important stages of invasive prenatal diagnostics of chromosome disorders and in conjunction with other cytogenetic techniques has a substantial impact in PDS of Russia.

Starting as early as 1990s, classic cytogenetic prenatal analysis were often supplemented with FISH method which was especially useful for precise identification of chromosome translocation products as well as other rearrangements including small marker chromosomes and provide additional convenience for chromosomal mosaicism studies.[21]

Attempts of non-invasive prenatal diagnosis of chromosomal aneuploidy using foetal cells floating in maternal blood were also repeatedly tried but failed to show its efficiency.[22]

4.8 Molecular Genetic Testing

Molecular studies and diagnostics of inherited diseases have been repeatedly tried in many institutes of Russian Academy of Medical Sciences and also at the institutes of Russian Medical Academy. Only biochemical and immune-cytochemical methods were sporadically tried in PD of monogenic disorders (see Sect. 3). Sudden breakthrough occurred at the start of the 1990s XX in laboratory of prenatal diagnostics of Ott's Institute in Saint Petersburg where within 5 years prenatal molecular diagnostic of the common genetic diseases was launched. At

[18]Kuznetzova et al. 1998.

[19]Baranov et al. 1995, Baranov and Tatyana 2007.

[20]Baranov et al.1995.

[21]Baranov et al.1995, Baranov and Tatyana 2007.

[22]Zolotukhina et al. 1999.

least two major reasons give a credit to this quick start: already available DNA probes specific for particular genetic disorders, gifted from the UK (1) and enrolment of Russia in already running International Human Genome Project which resulted in substantial financial support of our government to molecular genetic studies (2). As a participant of Human Genome Project, our laboratory was responsible for elaboration and implementation of molecular prenatal diagnostics of inherited disorders. By 1990 many causative genes of common genetic disorders and their mutations were already known, but there was no information on their actual frequencies in Russian populations, no technologies for their use in PD. Implementation of recently invented polymerase chain reaction (PCR) was one other lucky chance of success in our endeavour. It is worth mentioning that PCR in Russia was originally introduced in 1988 by Prof. Eugene I. Schwartz (1940–2003) here in Saint Petersburg.[23] All these coincidences favoured advancement of molecular diagnostics of common severe monogenic disorders in Russia. The first of them was cystic fibrosis. Getting started with the biochemical analysis of foetal enzymes in amniotic fluid, we soon elaborated and tried a nondirect molecular diagnostic method for this disorder and after CFTR gene discovery complemented it with direct detection of common ΔF508 mutation.[24] In 1989 the laboratory got the status of the state centre for prenatal diagnosis of CF and for at least next decade remained a single centre dealing with the families at risk of CF and provided its prenatal diagnostics. Over 6000 families at risk of CF have been genetically tested, and about 950 PD were carried out only in our centre so far.

It should be stressed that all molecular genetic studies of CF in Russia including PDS were launched in 1987 by Prof. Victoria N. Gorbunova and fruitfully proceeded by Prof. Tatiana E. Ivaschenko. Population studies over 20 mutations of CFTR gene including some novel ones (1677delTA) paved the way to reliable identification about 75% of affected chromosomes in the families at risk of CF and contributed a lot to its efficient prenatal diagnostics. Within the next few years, molecular diagnostics of CF spread throughout the country and is now available in many prenatal centres of Russia.

Other monogenic disorders amenable for molecular diagnostics elaborated in our laboratory in Saint Petersburg since the early 1990s included Duchenne muscular dystrophy, haemophilia A and B, phenylketonuria, myotonic dystrophy, Huntington chorea, fragile X-syndrome, spinal muscular atrophy and congenital adrenal hyperplasia (CAH).[25] Mutation analysis and population studies complemented the implementation of relevant prenatal molecular genetic diagnosis. They were mandatory prerequisite of PDS for each of the disorder mentioned above as well as for other prenatal molecular diagnostics. Actually we have realized that any monogenic disorder needs elaboration of its own quite specific algorithm of

[23] Shwarts et al. 1989.

[24] Baranov et al. 1991, Ivaschenko et al. 1991.

[25] Aseev et al. 1989, Baranov 1993; Baranov et al. 1990c, 1991, 1992.

PDS respective to the mutation patterns, methods of their identification, genetic facilities, peculiarities of clinical manifestation and term of pregnancy.

In the early 1990s, active molecular genetic studies of inherited monogenic and multifactor disorders started in many laboratories of federal medical genetic counsel (FMGC) and relevant institutes in Saint Petersburg, Moscow, Tomsk and others. Substantial impact in prenatal molecular diagnostics was made by Prof. Eugene I. Schwartz (Institute of Nuclear Physics, Saint Petersburg)[26] and by Prof. Oleg V. Evgrafov known for his studies in Duchenne muscle dystrophy and Huntington's disease (Institute for Medical Genetics, Moscow).[27]

Introduction of novel methods of DNA sequencing (NGS) and new options in molecular cytogenetic including array CGH in recent decades has substantially increased the efficacy of PDS and will be briefly reviewed in Sect. 5.

4.9 Biochemical Testing

The first biochemical testing of α-fetoprotein (AFP) was in amniotic fluid made by the electroimmunodiffusion technology by Prof. Victoria N. Gorbunova and Irine Elgart in 1987 in Saint Petersburg. AFP testing in amniotic fluid was complemented with concomitant detection of isoforms of acetylcholinesterase (AChE) for detection of opened NTD.

Biochemical test for prenatal diagnosis of cystic fibrosis was also practically important. Though two to three times rarer than in the West European population, CF poses a serious medical problem in Russia so far. Foetal intestine enzyme testing in amniotic fluid at the second trimester of pregnancy initially suggested in the UK by David Brock was initially tried in Saint Petersburg at the Institute of Experimental Medicine and the same year in Ott's Institute. First successful prenatal diagnostics of CF by means of biochemical method was reported by our group in 1989.[28] Before molecular diagnostics of CF became available and also in genetically noninformative families at risk of CF, prenatal biochemical testing was at the use for more than 10 years with over 400 families at risk of CF subjected to prenatal biochemical testing.

A special programme for the diagnosis and prevention of lysosomal storage diseases (LSD) both pre- and postnatal was initiated by Prof. Xenia D. Krasnopolskaya (1937–2000), then the chief of the *Department of Inherited Metabolic Diseases* (DIMD) at the *Research Center of Medical Genetics* (Moscow).[29] The work began in 1982 using standard as well as newly developed biochemical techniques for diagnostics of mucopolysaccharidoses (MPS),

[26] Shwarts et al. 1989.

[27] Evgrafov et al. 1990.

[28] Gorbunova et al. 1989.

[29] Krasnopolskaya et al. 1993, 1999.

mucolipidoses, glycoproteinoses, sphingolipidoses and other LSD. Prenatal diagnostics in the 1990s included MPS (types I, II, IIIA and IIIB, VI), Tay-Sachs disease, Sandhoff disease, GM1-gangliosidosis, metachromatic leukodystrophy, mannosidosis, Gaucher disease and multiple sulphatidosis. After 2000 at the same institute, the programme for diagnosis of rare metabolic diseases was significantly extended by Ekaterina Yu. Zakharova—the pupil and successor of Prof. X. D. Krasnopolskaya. Owing to molecular approaches, it now covers over 200 metabolic disorders and provides diagnostic service including prenatal, for many patients and families at high risk of metabolic disease throughout the country.

5 Active Implementation of PDS in Russia (2000–2010)

All facilities for PDS expansion including US and biochemical screening, genetic counselling, sampling technologies and efficient laboratory methods have become available since the beginning of the twenty-first century. Out of all pregnant women at the second trimester subjected to US testing in this period, inborn foetal disorders were identified only in 55 %, on average compared to 80–85% reflecting of current data.[30] Detection values of these disorders varied significantly in different prenatal centres being on average only 10–20% in LMGC and increased to 70–90% in RMGC. In spite of already existing ultrasound and biochemical screening programmes, the woman aged after 35 was still considered a risk factor indicative for invasive prenatal diagnosis (IPD). The other indications for IPD at this period included recurrent miscarriages, malformations or chromosomal aberrations in previous child, ultrasound markers of chromosomal diseases and the abnormal levels of AFP and HCG in maternal plasma. Altogether about 30% of all pregnant women at the second trimester were enrolled in the group of high risk of chromosomal anomalies and suggested for IPD. According to our data corresponding to average ones in Russia, the chromosome aberration detection rate was about 10–12% in the foetuses of the first and about 5–6% at the second trimester.[31]

Only about 20% of all IPD in Russia were carried out at the first trimester, and the rest 80% were at the second with high proportion of cord blood sampling—almost 30% of total IPD. Original programme of automatic selection of the women at risk of Down syndrome in their foetuses was at use since 2001 in our laboratory.[32] Similar automatic computer programme has been also suggested in Moscow.[33]

Further development of PDS was supported by Federal Ministry of Health Care order no. 457 issued on 31 December 2000. It has officially approved two levels of

[30]Medvedev and Elena 1998.

[31]Baranov and Tatyana 2007, Zhuchenko et al. 2014.

[32]Kascheeva et al. 2002

[33]Markova et al. 2005.

PDS with Maternal Consulting Cabinets (MCC) as first and RMGS as second level. It has also postulated as obligatory three consecutive US testing of all pregnant women at 10–14, 20–24 and 32–34 weeks of gestation. The biochemical screening shift from the second to predominantly the first trimester with a new set of embryonic marker proteins such as pregnancy-associated plasma protein A (PAPP-A) and free β-unit of HCG has been implicated. Thus gradual transition of PDS to earlier stages of pregnancy was officially approved.

Some more important positive events happened in PDS of Russia during this period. One of them concerned the first preimplantation genetic diagnosis of chromosomal and later genetic diseases in conjunction with clinic for extracorporeal fertilization.[34]

The last but not the least, one was implementation of molecular technology for prenatal diagnosis of the most frequent chromosomal diseases (trisomy 21, 13, 18, imbalances of sex chromosomes). The method called quantitative fluorescence PCR was initially introduced in our laboratory in 2004.[35] It has been recognized as efficient tool for PDS in Saint Petersburg in the middle of the 2000s and has expanded throughout the country after 2010.

In spite of these obvious achievements of PDS after the millennium, the overall efficacy of PDS of the foetuses with chromosomal aberrations still remained very low. For trisomy 21 (Down syndrome), it was equal to 30% and thus was almost twice less respecting to observe in European PD clinics. Only one fourth of all chromosomally abnormal foetuses were diagnosed at the first, while the rest were picked up at the second trimester of pregnancy.[36]

Thus by the end of 2010, the efficacy of chromosomal diseases PD in the foetuses still remained rather merger, and cordocentesis was carried in almost 20% of invasive sampling. All these results looked rather disappointing and witnessed for low efficiency of two-step PDS.

6 Combined Screening (Early Prenatal Screening Programme) (2010–2016)

Since 2010 a new one-stage programme has been tried for PDS in three territories. The latter was similar to already known international PDS programme. It included US testing with accurate measurements of nuchal translucency on 11–14 w.g. (1), analysis of PAPP-A and free β-HCG (2) and relative foetal Down syndrome risk calculation by means of software “Astraia” or “View Point” (3). Positive results of this pilot test substantiated its spreading throughout all 77 administrative regions in the country.

[34]Ivanov et al. 2008, Bazanov et al. 2009.

[35]Demin et al. 2008.

[36]Vakharlovsky et al. 2007.

It should be noted that early prenatal screening (EPS) approved by the Ministry of Health Care was actually initiated by Prof. Ludmila A. Zhuchenko (Moscow), and so far it is running under her supervision.[37] The project was officially approved by the order no. 572 (12 November 2012) of the Health Care Ministry. It was chiefly addressed to the medical staff participating in PDS especially to ones involved in early prenatal screening. According to its regulations, all PDS including terminations of pregnancy with affected foetus should be accomplished before 22 w.g. Pregnancy terminations after this term could be admitted after special permission from professional committee including US specialist, obstetrician, neonatologist and geneticist.

The results of the EPS summarized at the end of 2015 proved substantial improvements of all basic indexes of PD efficacy. Total number of pregnant women subjected to EPS in 2014 was about 1 million and thus close to the annual number of births. The selected group of the pregnant women at risk of Down syndrome decreased from 30% to almost 2% and thus became equal to this one according to international value used by FMF criteria. Proportion of the foetuses with chromosomal aberrations (main efficacy index of EPS) in 2015 increased over five times compared to this one in 2007–2009 (5.9% and 28%, respectively). Almost half of the recovered aneuploid foetuses had Down syndrome. Thus, implementation of EPS in PDS of Russia had unanimously positive effects and got full recognition at the federal level. Domination of EPS in PDS of current Russia was unanimously approved at the special meeting of Russian parliament (Duma) on February 25, 2016.

More progress of molecular PDS in 2010–2016 was mainly concerned with implementation of molecular methods for PD of common chromosome deletions (BACs-on-Beads technology, PerkinElmer) and whole genome structural chromosome rearrangements analysis. Comparative genome hybridization (array CGH) was also tried in some advanced PDS laboratories.[38]

Molecular genetic tests such as real-time PCR (RT-PCR) and multiplex ligation-dependent probe amplification (MLPA) were also used in PD of genetic and chromosome diseases by Prof. Alexander V. Polyakov (Moscow).[39] Non-invasive methods of foetal sexing in case of X-linked diseases as well as foetal Rh factor determination in Rh(-) pregnant women were also elaborated and tried in some PDS centres though their application did not become common as being not much in demand.

[37] Zhuchenko et al. 2014.

[38] Zolotukhina et al. 2005 and 2012, Skryabin et al. 2015.

[39] Polyakov 2014.

7 New Technologies Since 2015: Molecular Invasion in PDS

The most important revolution in PDS at the world scale and in Russia concerned the implementation of non-invasive prenatal diagnostics/testing technology (NIPD/NIPT) which became practically real after 2006 due to new generation sequencing technology (NGS). It was officially approved by International Society on Prenatal Diagnostics since 2014.

There are now several (3–5) private companies in Russia, which advertise NIPD, and according to some non-official information, over 4000 NIPD tests have already been done for pregnant women in Russia. Actually the NIPD tests were carried out somewhere abroad most probably in some US or European diagnostic centres with blood samples of pregnant women mostly illegally transferred abroad for testing.

Comparative NIPD conducted by two native molecular laboratories, one at the State Institute of Mother and Child Care and the other at the state university named after M.V. Lomonosov (both in Moscow), was done, and their combined results were submitted for publication this year.[40] Altogether about 700 NIPD tests were carried out by both groups and resulted in detection of 23.7% of chromosomally abnormal foetuses in 259 women of high-risk group and only 4.3% of foetuses with aneuploidy in cohort of 279 nonselected pregnant women. The sensitivity of NIPD in both groups was close to 100%; diagnostic value of false positive results was about 99% and thus corresponded to the NIPD indexes reported in the relevant literature. The data were submitted to the Ministry of Health Care for official approval as a manual for PDS. Though still rather preliminary and not sufficient for issue of official regulations, the problem of practical implication of NIPT/NIPD stands crucially for PDS in Russia. First, there are still just a few sequencing machines of sufficient DNA reading power in Russia. Second, the price of NIPD is still rather high and it will be hardly ever covered by the government. Third, there is a high probability that NIPD implementation will produce serious interference to already existing PDS and thus to rather efficiently operating at present time EPS. The necessity in the relevant specialists of high qualification in NIPD and confirmatory prenatal testing, medical geneticists and clinicians dealing with PDS is now especially urgent to make the transition from conventional invasive PDS to its new non-invasive era as smooth and cogent (see Sect. 4).

To our mind this transition is very time-consuming and will take unpredictably a long time. But even if some other days non-invasive PD becomes officially approved as a part of PDS in Russia, it should be integrated very smoothly and gradually not to ruin already existing algorithm of PDS in Russia.

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[40]Sukhikh et al. 2015, Pantiukh et al. 2016.

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Foundation of the International Federation of Human Genetics Societies: The Catalyst

Karen Birmingham

Abstract The International Federation of Human Genetic Societies was founded in 1996 by a group of American and European geneticists, who had become increasingly dissatisfied with the International Congress of Human Genetics, the only worldwide forum for geneticists at that time. The Congress, founded in 1956, was run by a self-styled "Permanent Committee", regarded by many as dysfunctional. In collaboration with the World Health Organization, two of the Permanent Committee co-authored a highly controversial set of guidelines on ethics and service provision, which Professor Marcus Pembrey used to catalyse the rapid instigation of the International Federation. This aspect of the Federation's foundation was unlikely to have come to light had it not been revealed during a brief interview with Professor Pembrey.

Keywords Oral history • Human genetics • International Federation of Human Genetic Societies • International Congress of Human Genetics • Ethical guidelines

Marcus Pembrey (b. 1943), Emeritus Professor of Paediatric Genetics at the Institute of Child Health, University College, London, was interviewed in his home in Mersea Island, Essex, UK in February 2013. The following description of the events leading to the Foundation of the *International Federation of Human Genetic Societies* is taken from this interview and is Professor Pembrey's personal view.

Although accounts of the establishment of the International Federation of Human Genetic Societies can be found in a variety of official papers,[1] it appears

[1] American Society of Human Genetics Minutes 1995; International Congress on Human Genetics Minutes, 1996.

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Fig. 1 Marcus Pembrey during interview in his home in Mersea Island, Essex (UK), February 2013 (Photo: Karen Birmingham)

that the connection between the Federation's beginnings and a highly controversial publication by the World Health Organization [*Guidelines on Ethical Issues on Medical Genetics and the Provision of Genetic Services*][2] is probably only held in the memories of a few key individuals. Evidence of considerable controversy generated by the new Federation can be found in correspondence within the personal archives of at least one of these individuals, but what is not to be found in any of the formal or informal documentation is how and why the disputed document catalysed the formation of the Federation. Marcus Pembrey was instrumental in setting up the Federation, but this specific historical aspect of the Federation's creation only emerged whilst he was being interviewed on a different subject entirely. He was interviewed as part of a series of brief oral histories concerning the ethics of a well-established longitudinal birth cohort: the Avon Longitudinal Study of Parents and Children (ALSPAC).[3] ALSPAC's own Ethics and Law Committee, which is attached to the Study and continues to this day, was initiated in 1990, while the study was still being planned and piloted and Marcus Pembrey, as ALSPAC's Director of Genetics from 1989 to 2005, frequently provided essential information and advice to the Committee. In describing his background and the evolution of his ethical values, he mentioned a World Health Organization "ambush" and the consequent formation of the International Federation of Human Genetic Societies.[4] Subsequently, confirmation of the Federation's early history was found within his personal archive, now housed within the *Wellcome Library*, London, although no link was evident between the *World Health Organization* guidelines and the establishment of the Federation (Fig. 1).

[2]Wertz 1995.

[3]Avon Longitudinal Study of Parents and Children [website].

[4]Interview with Marcus Pembrey, recorded by Karen Birmingham 2013.

1 Background

The official account of the foundation of the International Federation of Human Genetic Societies can be found in the minutes of both the annual meeting of the American Society of Human Genetics (1995) and the Ninth International Congress on Human Genetics (1996).[5] Founded in 1996, the Federation was set up "to provide a transparent structure to facilitate communication throughout the international community of human geneticists".[6] Transparency was of the utmost importance as the international forum for geneticists at the time, the *International Congress of Human Genetics* founded in 1956 and run by the self-styled Permanent Committee, was considered by Marcus Pembrey and others as both impenetrable and undemocratic. Some members of the Permanent Committee were critical of their own committee, considering it to be "too large to be effective"[7] and "with no legal status".[8] Although the Congress and especially the Permanent Committee were considered defective, there were other well-established and well-run societies in place such as the American Society of Human Genetics and the European Society of Human Genetics.

Marcus Pembrey has been a member of the European Society of Human Genetics from 1989, President from 1994 to 1995 and Chair of the Society's ethical committee, the Public and Professional Policy Committee, from 1994 to 1998. During his interview, he describes how the Public and Professional Policy Committee tried to construct policies from their extensive experience of real clinical examples and then to harmonise the European policies. He emphasised the importance in this work of his "great ally" Ségolène Aymé (b. 1946), also a member of the Public and Professional Policy Committee, President of the European Society of Human Genetics from 1996 to 1997 and an esteemed medical geneticist. "She was very organised" when he felt himself to be "very disorganised" but more importantly "she could handle Brussels".[9]

2 Guidelines

In 1995, Marcus Pembrey and his European colleagues became aware of the "*Guidelines on Ethical Issues on Medical Genetics and the Provision of Genetic Services*".[10] The document was relatively long (approximately 90 pages) and attempted to address two separate issues: (i) ethical dilemmas raised by potential

[5] See EN 1.

[6] International Federation of Human Genetic Societies [website].

[7] Moreton 1997a.

[8] Moreton 1997b.

[9] See EN 4.

[10] See EN 2.

use of genetic tests and more controversially (ii) recommendations in terms of services to the community.[11] Marcus Pembrey relates that as far as he and others were concerned, the authority of the authors to write such a document was decidedly questionable: "When we saw a draft of this we fell off our seats. Bloody Hell! Who are these [. . .] people telling the world how to do their thing?"[12] The authors were Dorothy Wertz, John Fletcher, Kåre Berg and Victor Boulyjenkov although Marcus Pembrey only recalled Dorothy Wertz and Kåre Berg during the interview. Dorothy Wertz (1938–2003) and John Fletcher (1931–2004) were well-respected bioethicists, who had conducted a survey of medical geneticists in eighteen nations and considered the varied approaches to ethical problems in counselling, screening and prenatal diagnosis. This culminated in the publication in 1987 of *Ethics and Human Genetics: A Cross-Cultural Perspective*.[13] Kåre Berg (1932–2009) was a Norwegian Professor of Medical Genetics, a well-regarded genetic researcher and at the time the guidelines were written, an adviser in genetic diseases and medical ethics to the World Health Organisation. Victor Boulyjenkov (b. 1948) is a medical geneticist who was employed by the World Health Organization on the Hereditary Diseases Programme in the Division of Noncommunicable Diseases and had approached Wertz and Fletcher in the summer of 1993 concerning these guidelines, which were originally for use in developing nations.[14] Marcus Pembrey and colleagues' indignation stemmed particularly from the inclusion of service provision in the draft Guidelines. It was not entirely clear as to the exact status of the guidelines as regards the World Health Organisation; the cover states "in cooperation with the World Health Organisation", Dorothy Wertz cites the document in the footnote of an article published in 1997 as "WHO guidelines on Ethical issues in Medical Genetics and the Provision of Genetic Services, . . . (hereinafter WHO guidelines)"[15] but by 1999, after years of controversy, Dorothy Wertz stated "It is important to know that the monograph is not an official document of the World Health Organisation and represents only the views of its four authors."[16]

The publication of the 1995 document was described by Marcus Pembrey as "a bit of an ambush" and he and his European colleagues were "triggered into thinking globally" about guidelines in genetics. They decided that an international federation of human genetic societies should be formed to counter this unwelcome involvement but "the first thing to do was to persuade the Americans to come in because the Americans dominated everything".[17]

[11] Aymé 1997a.
[12] See EN 4.
[13] Wertz 1987.
[14] Nippert 1999, 168.
[15] Wertz 1997, 299–346.
[16] Fletcher 1999, 107.
[17] See EN 4.

3 Minneapolis Breakfast Meeting

During the interview Marcus Pembrey recounts how members of the European Society of Human Genetics frequently attended the much larger meetings of the American Society of Human Genetics and that he and Ségolène Aymé "had a couple of allies over there".[18] He did not identify the 'allies'. In October 1995, the American Society held its annual meeting in Minneapolis, Minnesota, USA. Marcus Pembrey recalls a breakfast meeting that was held at this conference, although he does not make clear the purpose of the meeting. He said that Maimon Cohen (1935–2007), President of the American Society of Human Genetics (1994), had suggested previous to this meeting that there could or should be an informal association of presidents of the human genetic societies, but nothing had come of it. Marcus Pembrey goes on to vividly describe how at a crucial moment during the meeting he produced the guidelines, of which the Americans were completely unaware, and the outrage that this document provoked. "They were incensed beyond belief" and within half an hour a resolution to form an international federation of human genetic societies had been passed. "I remember people coming out [of the meeting] said 'Bloody Hell, how did you pull that off?' and I said 'well, you know, it just happened'."[19] It was agreed that bye-laws should be drafted and considered at the International Congress on Human Genetics in Rio de Janeiro the following August. The first meeting of the International Federation of Human Genetic Societies was duly held in Rio prior to the Ninth International Congress on Human Genetics.

4 Fallout

The fallout from the establishment of the Federation was considerable. There were two distinct aspects: (i) the rivalry generated between the new Federation and the Permanent Committee of the well-established International Congress of Human Genetic Societies and (ii) the writing of new ethical guidelines for medical geneticists. These two issues were not unrelated as Kåre Berg and Victor Boulyjenkov were both members of the Permanent Committee and authors of the controversial guidelines. Albert Schinzel (b. 1944), President of the European Society of Human Genetics (1995–1996), when reporting on the meeting in Rio de Janeiro, wrote that "K Berg was not present but according to rumour not pleased at all about the activities. No wonder, he presents himself as a partner for the WHO [World Health Organisation] in genetic issues [...] ."[20]

[18] See EN 4.
[19] See EN 4.
[20] Schinzel 1996.

5 Rivalry

There is much evidence in Marcus Pembrey's personal archive of the antagonism between the Permanent Committee and the new Federation which rapidly took over the oversight of the five yearly meetings of the International Congress of Human Genetic Societies. Perhaps the most startling evidence is contained in a letter from one member of the Permanent Committee to another in May 1997 concerning membership of the Federation: "Its three full members have unpleasant racial overtones of blond beasts against the tinted folk that were not intended but extraordinarily insensitive."[21] The three full members were the founding, continental, human genetic societies: American, European and Australasian. This letter, to the author's credit, was copied to Marcus Pembrey and one of the other "blond beasts". He also wrote later that year that "As constituted the Federation has no legitimate claim to speak for human genetics, and its existence has not yet been recognised by any international congress."[22] This member of the Permanent Committee was not without criticism of his own committee; he had previously expressed concern to Marcus Pembrey at the "shambles of the Permanent Committee"[23] and to another colleague he wrote "It will be interesting to see how this is settled—preferably not as in the Middle Ages when there were two Popes."[24]

6 New Guidelines

In October 1997, Ségolène Aymé, by now the first president of the International Federation of Human Genetic Societies, wrote to Victor Boulyjenkov, one of the authors of the World Health Organization guidelines, informing him that the Federation had reviewed the guidelines as requested by him. She stated that:

> . . . neither the IFHGS [International Federation of Human Genetic Societies] nor any of the three full members [. . .] can endorse this particular document. [. . .] It seems to us that the present document cannot be revised because of concerns over both the content and the consultation process. We believe a document of such potential importance should make use of the experience of the standing committees of international professional societies representing those who directly provide medical genetic services, and which have made some progress in developing consensus views.[25]

Despite this, in December that year, fifteen World Health Organisation advisers from developed and developing nations met and revised the guidelines. The much shorter document (16 pages) was published in May 1998 as "*Proposed*

[21] Moreton 1997c.
[22] See EN 8.
[23] Moreton 1997d.
[24] See EN 7.
[25] Aymé 1997b.

International Guidelines on the Ethical Issues on Medical Genetics and Genetic Services".[26] Dorothy Wertz emphasised that the document represented "a 100% consensus (not a majority vote) among all those present" and stated that "The WHO advisers hope that the proposed guidelines will become the nucleus of an international code, similar to the Helsinki Declaration."[27]

Despite the controversy generated by the World Health Organization guidelines, Marcus Pembrey recalls that the new Federation only considered writing its own international guidelines some time later, when Ségolène Aymé "pointed out that there was no point in having the Federation unless it did something useful"[28] and offered to raise European money for such an endeavour. This she did and the Federation established its transparent and democratic approach to the creation of guidelines: "the purpose of developing professional Guidelines and Policy Statements is to share them on the IFHGS [International Federation of Human Genetic Societies] website and to garner endorsements from the Corresponding Member Societies."[29]

7 Conclusions

Marcus Pembrey is clear that the founding of the International Federation of Human Genetic Societies was catalysed by the sudden disclosure of the document *Guidelines on Ethical Issues on Medical Genetics and the Provision of Genetic Services*.[30] Recollections during his interview capture this historical aspect convincingly although confirmation in any written documentation has not been found. He also makes it clear as to why he thought it possible for the Federation to be formed so rapidly at that particular time; he along with many others felt that there was an urgent need for a change to the only worldwide forum for geneticists, specifically the International Congress of Human Genetics, as it was undemocratic and lacked transparency. Kåre Berg and Victor Boulyjenkov were pivotal in the process as not only were they members of the Congress' Permanent Committee, but also authors of the guidelines. Marcus Pembrey describes previous antagonism to Kåre Berg: "an autocratic Norwegian geneticist, who had single-handedly controlled the European Society for many years until Ségolène and a few gallant people—I was not involved in this revolution—took it over and rested it from

[26]World Health Organisation 1998.

[27]See EN 14.

[28]See EN 4.

[29]International Federation of Human Genetic Societies, 1999; International Federation of Human Genetic Societies Executive Committee, 2002; Corresponding Members of the International Federation of Human Genetic Societies are national organisations while Full Members are regional multinational organisations.

[30]See EN 2.

this oligarchy of male retired … [sentence unfinished]".[31] The "revolution" took place at the European Society of Human Genetics meeting in Corfu (1990). Margareta Mikkelsen, (1923–2004) ESHG President 1993, also had a crucial role.[32] Once the new Federation was set up, their own guidelines were not immediately forthcoming, suggesting that the World Health Organization guidelines were indeed just a catalyst and not the underlying reasons for this major transformation in geneticists' international forums.

Although not mentioned in his interview, Marcus Pembrey had further discussions with Ségolène Aymé and others which provoked memories of their extreme discomfort with the Permanent Committee's financial arrangements. The Committee rules had established that the benefits of the congresses were for the Permanent Committee and the losses, if any, for the local organisers. The meeting for 1996 was to take place in Brazil, a developing country, which they felt should not have to bear the burden of any losses. The Permanent Committee's financial arrangements, as with other aspects of the organisation, were not transparent, and there was some evidence that money was being misappropriated. Requests for clarification of the accounts were not forthcoming. This was considered another important factor in convincing their American colleagues that a new Federation should be established.[33]

Marcus Pembrey's recall of events leading to the establishment of the International Federation of Human Genetic Societies during his interview was an 'aside' from the main theme of the oral history, but it is of importance. It not only captures an undocumented facet of history but also the emotional elements that were significant in driving the creation of a more democratic and legitimate organisation for human geneticists. The surprise and indignation arising from the realisation that Kåre Berg, Victor Boulyjenkov and two ethicists were seeking to speak on behalf of all practising medical geneticists through the World Health Organization is unlikely to have been documented even if the crucial breakfast meeting had been minuted. It is perhaps only through interview with individuals who were involved at the time that the more elusive aspects of history such as these can be identified.

[31] See EN 4.

[32] Aymé 2016; Personal correspondence to Marcus Pembrey, 26.07.2016.

[33] See EN 32.

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Part V
Gene Mapping

The First Human Genetic Map 1936

Alan R. Rushton

Abstract The rediscovery of Mendel's law of heredity in 1900 fueled breeding studies of plants and animals, which demonstrated the independent segregation of genetic characters during meiosis. The co-segregation of grouped characters suggested to T. H. Morgan that this behaviour paralleled the behaviour of chromosomes during meiotic segregation. Rare crossover events implied a model in which the frequency of such recombination events was correlated with the physical location of genetic elements on specific chromosomes.

Human genetic studies did not progress because there were many human chromosomes in each human cell, and the likelihood of detecting co-segregation of two characters was minimal. The genes for different blood groups appeared to segregate independently and offered an opportunity to assess potential linkage with characters such as eye and hair colour or diseases such as haemophilia and Friedreich's ataxia. However, research groups in the USA and the UK found no evidence of human genetic linkage before 1935.

Julia Bell and J. B. S. Haldane from the Galton Laboratory in London then studied the segregation of two characters known to be associated with the X chromosome: haemophilia and colour blindness. Their pedigree analysis published in 1936 demonstrated close linkage of the two loci. Haldane then expanded the work to involve several other genetic characters associated with the X chromosome. Recombination frequencies were used to construct a genetic map of the human X chromosome with five defined loci. The concepts developed in this work provided the basis for linkage studies in the decades ahead until the advent of DNA technology.

Keywords Human gene mapping • Julia Bell • Haemophilia • JBS Haldane • Colour blindness • Galton Laboratory

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The Cambridge biologist William Bateson (1861–1926) became the champion of the application of the inheritance laws first elucidated by Gregor Mendel (1822–1884). Bateson began breeding studies after 1900 in both plants and animals which confirmed that the unit characters for inheritance typically segregated independently at meiosis.[1] Within a few years, however, exceptions to the rule became evident. In breeding studies of the primrose and the sweet pea, Bateson and his colleagues found unexpected inheritance ratios suggesting "coupling" of characters instead of independent segregation.[2] Arthur Darbishire (1879–1915) and W. Raphael Weldon (1860–1906) at Oxford observed the same phenomenon in their breeding studies of mice. The expected segregation patterns for eye and coat colour were evident, but the neurologic trait "waltzing" was not inherited in the proportions expected.[3]

A mechanism to explain this so-called linkage of inherited characters was developed by T. H. Morgan (1866–1945) and his colleagues at Columbia University after 1910.[4] They proposed that the unit characters were carried by the chromosomes, almost like beads on a string. Their massive breeding studies with the fruit fly *Drosophila melanogaster* showed that the hereditary characters from that organism fell into four aggregations of co-segregation termed linkage groups. This species also had four pairs of chromosomes. Other varieties of the fruit fly had different numbers of chromosomes. It became clear that the number of linkage groups correlated with the pairs of chromosomes characteristic for each species. The same phenomenon was observed in plants. The sweet pea has seven chromosome pairs and seven genetic linkage groups.[5] The *Drosophila* workers then observed that the characters within each linkage group were not always inherited together. They reasoned that a transfer of genetic material may have occurred during the synapse phase of meiosis when chromosome arms were aligned. A random break in chromosome arms then could result in the formation of daughter chromosomes with heterogeneous regions. The probability of such a "crossover" event appeared to be specific for each character. If the genes occupied a linear array on the chromosome arms, those near each other would cross over at a lower rate than those further away. The New York group reasoned that a physical map of the chromosomes could be developed using linkage studies and the frequency of such recombination events.[6]

Further linkage investigations of plant and animal species expanded dramatically in the years after World War I. But attempts to analyse human genetic linkage made no progress. Controlled breeding studies were not possible, and attempts to correlate the segregation of physical traits were expected to be difficult because of the large number of chromosomes in human cells. T. S. Painter (1889–1969)

[1] Olby 1987.

[2] Bateson 1905, Bateson and Gregory 1905.

[3] Darbshire 1904.

[4] For a general review of linkage studies, see Sturtevant and Beadle 1962, 63–92, and Mayr 1982, 754–777.

[5] Punnett 1923 and 1926.

[6] Morgan 1911, Dunn 1991, and Morgan 1922.

counted 48 chromosomes (23 autosome and 1 sex chromosome pair) in 1923.[7] The likelihood of finding pairs of co-segregating characters seemed to be miniscule at best. The independent segregation of the different human blood groups subsequently suggested a series of genetic markers that could be measured in a large human population and then correlated with the inheritance of specific human traits, both normal and pathological. For example, Philip Levine (1900–1987) from the Cornell Medical College in New York studied the association of the ABO blood group and sensitivity to allergies, but found no correlation in 1927.[8]

Laurence Snyder (1901–1986) from the Ohio State University Medical School advocated widespread application of such human linkage studies in the 1920s. He believed that the discovery of linkage between blood groups and human diseases such as migraine headache and schizophrenia could predict disease in children and then allow parents the ability to plan further reproduction. He suggested study of the co-segregation of several common traits:

1. ABO blood group
2. Eye colour
3. Hair on second digit joint
4. Occipital hair whorl direction
5. PTC tasting
6. Susceptibility to goitre
7. Susceptibility to diphtheria
8. Atopy
9. Polydactyly[9]

Early in the next decade, Felix Bernstein (1878–1956) from the University of Goettingen proposed a series of mathematical formulae to assess human linkage with blood group markers.[10] His work galvanised the thinking of Lancelot Hogben (1895–1975) at the London School of Economics. His 1932 book *Genetic Principles in Medicine and Social Sciences* was written as a "genetic manifesto", a call to action that stimulated human genetic research thereafter. He agreed that the human blood groups provided good markers to begin linkage studies, but he also argued against a piecemeal approach. Contributions from a large number of subjects were required for any statistical analysis of linkage data. He proposed the establishment of registrars in hospitals to collect family pedigree data:

> Little further advance will be made in the study of hereditary transmission in the human species through the work of isolated investigators [. . .]. Advances in the future will only be made if it is recognized that the further development of the subject presupposes the cooperation of large number of workers with facilities for obtaining the requisite data.[11]

[7] Painter 1923.

[8] Levine 1926.

[9] Snyder 1929 and 1931.

[10] Bernstein 1930 and 1931.

[11] Hogben 1932, 89–90.

His manifesto challenged investigators in this field:

> Today the prospects for advancing human genetics as an exact science are much better than they appeared to be 20 years ago. New methods of mathematical analysis for testing the applicability of experimentally established hypotheses to human data have been elaborated [. . .]. **It is now legitimate to entertain the possibility that the human chromosomes can be mapped**. (Emphasis added)[12]

Hence, this was the first Human Genome Project.

A review of Hogben's book in the *British Medical Journal* declared a "new spirit of hopefulness" in human genetics that had recently evolved as statistical methods were devised to analyse clinical data from human families, despite their small size and non-random breeding pattern.[13]

However, the hope that human linkages would certainly be discovered was soon disappointed as several extensive collaborative studies at this time revealed *nothing* about the organisation of the human genome. Hogben and his colleagues initially examined the segregation of several common human traits:

1. ABO blood group
2. Susceptibility to goitre
3. Susceptibility to diphtheria
4. Atopy
5. Haemophilia
6. Direction of occipital hair whorl
7. Migraine
8. Polydactyly
9. Eye colour
10. Feeblemindedness

No correlations were observed.[14] The same group next examined the segregation of the ABO blood group, tasting of PTC, brachydactyly and the neurologic disorder Friedrich's ataxia. No linked segregation was observed. Hogben noted sadly, "The yield of relevant information obtained in linkage studies confined to a few genes is very small [. . .]".[15]

A serology research unit was established in 1935 at the Galton Laboratory, University College London, to expand the number of blood group markers that could be utilised in human linkage studies. George L. Taylor (1897–1945) and Robert Race (1907–1984) collaborated with R.A. Fisher (1890–1962) in the hope of finding a link to inherited diseases such as Huntington's chorea which did not become manifest until middle age. Unaffected persons at risk then could be advised not to have children who also were likely to inherit the same devastating disease.

[12]Hogben 1932, 214.

[13]Anonymous 1932, 293–294.

[14]Hogben 1932, 83–84.

[15]Hogben and Pollack 1935.

The group collected family pedigrees from many London hospitals and attempted to correlate the segregation of the following characters:

1. ABO blood group
2. MN blood group
3. P blood group
4. PTC tasting
5. Eye colour
6. Hair colour
7. Freckles
8. Attached earlobe
9. Occipital hair whorl
10. Handedness
11. Hair on second digit

Unfortunately, no linkages were detected, and the results of the study were never published.[16]

About the same time, Alexander Wiener (1907–1976) at the Jewish Hospital in Brooklyn, New York, analysed family data segregating atopy, eye colour and the blood groups ABO and MN. Again, no linkage associations were detected.[17]

With the abject failure of these attempts to find any human genetic linkage relationships, Julia Bell (1879–1979) and J. B. S. Haldane (1892–1964) from the Galton Laboratory then examined the behaviour of several traits already known to be associated with one chromosome, i.e. the special case of sex-linked genetic characters. In 1930, Charles Davenport (1866–1944) from the *Eugenics Record Office* in New York had collected data on 14 human characters that appeared to be inherited in a sex-linked fashion and therefore should provide a fertile field for genetic analysis.[18] Haldane noted that this seemed to be a reasonable goal. "I am fundamentally a lazy man and like to see definite results when I do make an effort".[19]

Julia Bell had worked with Karl Pearson (1857–1936) at the Galton Laboratory for many years, first as a statistician and later as the general editor for the *Treasury of Human Inheritance* (THI). The *THI* was a massive multivolume compilation of pedigree data on normal and pathological human characters which was published over several decades. Bell collaborated with many physicians in the collection of family data and eventually qualified as a physician herself in 1920.[20] However, Pearson did not permit analysis of genetic mechanisms for the *THI* data. He had been angered by the biometric-Mendelian controversy between himself and Bateson before 1910[21] and chose to view the

[16]Mazumdar 1992, 239–240.

[17]Wiener et al. 1936/37, and Zieve et al. 1936/37.

[18]Davenport 1930.

[19]Kevles 1985, 202.

[20]Harper 2005 and Jones 2004.

[21]Rushton 2000 and 2009.

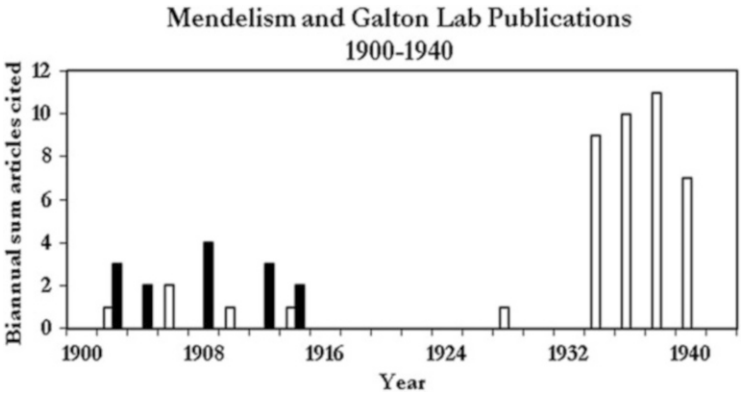

Fig. 1 Articles favouring biometry in *black* and those Mendelism in *white* (© A. Rushton)

work strictly as an "unbiased gathering of hereditary data".[22] It was to be assembled in a "theory-free fashion".[23] Pearson was convinced that the "mission of science is not to explain but to describe. . .".[24]

I have examined all the articles published before 1940 in the Galton publications *Biometrika* and *Annals of Eugenics*, as well as the *THI* volumes (see Fig. 1). Before World War I, there is evidence of give and take among the authors' discussions in these publications. However, once Pearson decided to declare his journals theory free, there are only rare exceptions when genetic analysis was permitted by the editor.

Pearson retired in 1933, and R.A. Fisher assumed control of the *Galton Laboratory* and its publications. Fisher noted in a 1934 editorial that he sought to elicit cooperation between the experts in genetics and statistics. His goal was to "prevent the perpetuation of one-sided 'biometrical' and 'genetical' standpoints on human problems".[25] Julia Bell was now permitted to assess the hereditary patterns of the pedigrees she had collected. In 1934, she declared Huntington's chorea to be a dominant character.[26] Subsequently, she discovered three different forms of peroneal muscle atrophy: recessive, dominant and sex linked. She also examined the segregation of PTC tasting in these families, but found no correlation of the characters.[27]

Mendelian-based studies certainly flourished at the *Galton* under its new director. Haldane occupied the chair of genetics at University College London after 1933. Previously he had been reader in biochemistry at Oxford and had broad

[22]Pearson 1909, i.
[23]Norton 1975 and Pearson 1938, 5.
[24]Porter 2004, 257.
[25]Fisher 1934/35, 1.
[26]Bell 1934.
[27]Bell 1935.

experience in genetics, statistics and physiology. The subsequent director at Galton, Lionel Penrose, recalled that Haldane typically functioned:

> to supply genetical and mathematical ideas, and he welcomed every opportunity of examining the material collected by others and using his own methods of analysis upon it. It was characteristic of him that in doing this he always gave generous recognition. . .to the contributions of his co-workers when results were published.[28]

Bell began by collecting pedigrees with specific sex-linked characters. Colour blindness and haemophilia were two well-known traits inherited by males. William Bulloch (1868–1941) of the *London Hospital* had assembled data from many bleeder families over the years and had worked with Bell to prepare a volume of the *THI* on this topic.[29] As early as 1928, Bulloch had noted to Fisher that some haemophiliacs were also colour blind. One bleeder had told Bulloch that he was colour blind and that he knew of other "bleeders" who also were unable to discern colours.[30] Genetic analysis revealed that the elements for colour blindness and haemophilia could be borne by only one X chromosome in unaffected females or the genes could each be located on one of the two X chromosomes in female cells.

About the same time, M. Madlener of Kempten Hospital in Germany reported a family that did show the coupling of the genes for haemophilia and colour blindness: this family pedigree demonstrated the co-segregation of the two characters, indicating that the two genes involved were coupled to only one of the two X chromosomes. Hence, this example clearly demonstrated human genetic linkage.[31]

Bell collected five pedigrees of individuals with red-green colour blindness and haemophilia from her London physician sources. She and Haldane examined this data as well as the Madlener pedigree. Approximately 30 male offspring were recorded, and one case of apparent recombination was documented. They calculated that the genes for colour blindness and haemophilia were tightly linked on the X chromosome with about 5% likelihood of recombination. The results of their collaboration were presented at the Royal Society in London on 27 February 1937.[32]

Julia Bell noted to a friend that the work was "really very exciting".[33]

Haldane then drew upon Bell's previous studies showing that certain human characters exhibited variable inheritance patterns in different families. A character might segregate in dominant, recessive or sex-linked fashion. He reviewed pedigrees of several disorders which he believed demonstrated what he termed "partial sex linkage". The frequency of crossing over in such families then could be used to calculate map distances along the X chromosome. He examined families with the skin disorder epidermolysis bullosa, a malignant skin disease xeroderma pigmentosum, colour blindness and two visual degenerative disorders Oguchi's

[28] Pirie 1966.

[29] Bulloch and Fildes 1911.

[30] P 647/5: Bulloch to Pearson 1928.

[31] Madlener 1928.

[32] Anonymous 1937, Bell and Haldane 1936 and 1937a,b.

[33] Kevles 1985, 202.

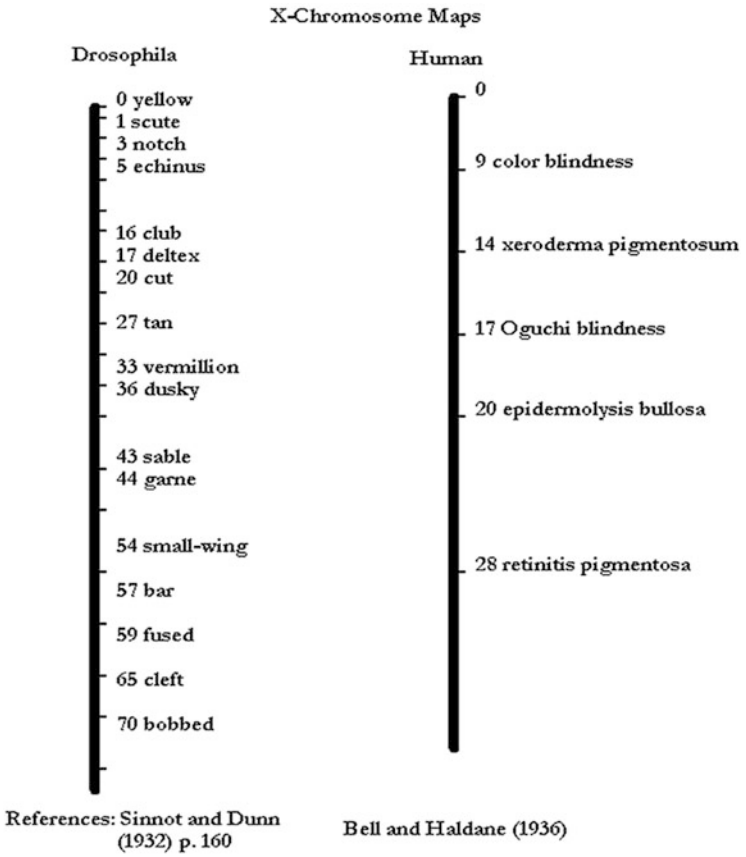

Fig. 2 Concurrent Drosophila and human X chromosome maps (© A. Rushton)

disease and retinitis pigmentosa without deafness. The calculated recombination rates resulted in the following "provisional map of a human chromosome" (Fig. 2).[34] The *Lancet* reported this mapping of a human chromosome to the wider medical audience in 1937.[32]

The "First Human Genome Project" successfully prepared a genetic map of one human chromosome. Work on detecting autosomal linkages resumed after World War II, again using blood group markers. Success began to slowly emerge in 1951 as Jan Mohr (1921–2009) from the University of Copenhagen found linkage between the Lutheran and Lewis blood groups.[35] J.H. Renwick (1926–1994) and S.D. Lawler (1922–1996) from the *Galton Laboratory* then found the first disease linkage between the ABO blood group and the nail-patella loci in 1955.[36]

[34]Haldane 1936 and 1936/37.

[35]Mohr 1951.

[36]Renwick and Lawler 1955.

The advent of recombinant DNA technology in the 1980s permitted researchers to directly analyse the intricate content of individual genes and gene neighbours residing on the same chromosome. The second *Human Genome Project* successfully mapped the entire human genome of 3.3 billion base pairs into 20, 500 discrete genes by 2003.

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Glasgow Contributions to Human Gene Mapping

Malcolm A. Ferguson-Smith

Abstract The classic approaches to human gene mapping using genetic linkage in human pedigrees (Renwick) and nondisjunction and deletion in patients with chromosomal aberrations (MAF-S) were established in 1959–1961 in Pontecorvo's Department. The genetic markers yielded several genetic linkages before Renwick left for UCL in 1968. The cytogenetics lab mapped a stature gene and others associated with the Turner phenotype to Xp, and TGF and XG were mapped in XX males with X-Y illegitimate recombination to just outside the pseudoautosomal boundary, leading to the discovery of SRY 24 years later. ACP1 to chromosome 2p was the first human gene to be mapped by deletion mapping. Others followed from the lab including the loci for ABO, AK1, HP, ADA, GALT, GOTS, XG, HPAFP, TS1 and HY. Heterozygosity at many other loci excluded them from the deleted regions enabling the construction of an exclusion map. A physical map of both the pairing and non-pairing regions of the Y was made using Southern blotting and DNA markers isolated from a Y chromosome library. The first reports of the successful localisation of single copy genes by isotopic in situ hybridisation came from the cytogenetics lab in 1980, specifically the regional localisation of the alpha- and beta-globin genes, followed by kappa light chain genes to 2p. Much of the Glasgow contribution to the human mapping project derived from a cytogenetic approach based on nondisjunction and deletions associated with chromosomal syndromes.

Keywords Linkage • Deletion • Exclusion • FISH • Haploidisation

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The Genetics Department at Glasgow University started with the appointment of Guido Pontecorvo (1907–1999) to a lectureship in Genetics in 1945, and this was followed by his promotion to a new Chair of Genetics in 1955. Ponte, as he liked to be called, had been a PhD student (1939–1941) with Herman Muller in Edinburgh and had moved in 1941 to a research post in the Zoology Department in Glasgow. Ponte's great contribution was his work on tetrad analysis, mitotic crossing over and haploidisation in *Aspergillus nidulans* that led to his finding of intragenic recombination and the discovery that the gene could be divided into smaller units, later known as cistrons, each recognisable by mutation. This work was reported just prior to the publication of the Watson and Crick model of the DNA double helix. His exploitation of what he termed the parasexual cycle (in distinction to the sexual or meiotic cycle) led Ponte to the notion that, if somatic crossing over could be studied in cultured human cells, the problems of human gene mapping could be overcome; these problems were the impossibility of experimental breeding, the small number of progeny and long generation times. His ideas were encapsulated in a series of lectures presented in 1956 at Columbia University and published in a monograph entitled *Trends in Genetic Analysis*.[1] Somatic recombination, chromosomal nondisjunction and haploidisation were the essential elements to be harnessed in the construction of a human gene map from cultured cells. Progress in achieving these strategies was hampered by a lack of analysable polymorphic markers, and Ponte's approach did not bear fruit by the time he left Glasgow in 1968 to join Michael Stoker at the ICRF. Within 2 years, segregation of human chromosomes in human/mouse somatic cell hybrids had emerged to substantiate Ponte's vision. Interspecies biochemical variants provided an abundance of polymorphisms for chromosomal assignment and gene ordering. Ponte joined in the research on somatic cell hybrids at ICRF, and his contributions included the discovery[2] that polyethylene glycol could be used instead of Sendai virus to induce man/mouse somatic cell hybrids.

I believe that the *first ever* contribution to human gene mapping came from Glasgow in 1937, well before the establishment of the Genetics Department. This came from Professor William John Brownlow Riddell (1899–1976), Head of the Department of Ophthalmology. One of his interests was colour blindness, and prompted most likely by Julia Bell (1879–1979), he looked for genetic linkage between the X-linked loci for haemophilia and colour vision defects. Three of the fourteen haemophiliac families he tested also segregated for colour blindness, and the first revealed the only recombinant included in the famous paper of Bell and Haldane[3] that described the first example of genetic linkage in humans. As Riddell's paper[4] on this family was published 3 months earlier than the Bell and Haldane paper, it can be said that Riddell was the first to report crossing over in humans. The initial rough estimate of 5% recombination between the two loci was reinvestigated

[1] Pontecorvo 1958.

[2] Pontecorvo 1975, 397–400.

[3] Bell and Haldane 1937, 119–150.

[4] Riddell 1937, 113–116.

by Haldane and Smith in 1947 in 17 families (including the three contributed by Riddell), and a new estimate of 9.8% recombination was reported.[5] While the first human linkage involved the X chromosome, the first autosomal linkage was reported by Jan Mohr in 1951 between the Lutheran and Lewis (including secretor) blood groups;[6] Mohr found a hint that the gene for myotonic dystrophy belonged to the same linkage group and this was confirmed in 1971.[7]

The complex work on somatic recombination in cultured cells was in progress in the Glasgow Genetics Department in 1959 when James H. Renwick (1926–1994) joined as senior lecturer to continue his work from the Galton Laboratory on human gene mapping by classical genetic linkage analysis, supported by a novel computer programme he was developing with colleagues in Baltimore. With Marian M. Izatt, he set up a marker lab to test common blood groups and other polymorphisms in serum and red cell enzymes that could be used to test for linkage in families with genetic disorders and dominant traits. Renwick felt that a word was needed to indicate that loci separated by 50% recombination could be on the same chromosome and so he coined the useful word "syntenic". Synteny is now a genetic term in common usage. During the 1960s blood samples from such families streamed into the marker lab for testing. Renwick's publications on linkage arising out of his 9-year work in Glasgow before moving to UCL in 1968 are listed in Table 1. They include follow-up studies on the famous ABO/nail-patella syndrome linkage[8] and the first gene assignment to a human autosome, namely, Duffy blood group to HSA1.[9] Other notable findings on the list are the assignment of haptoglobin to HSA16,[10] the linkage between Duffy and zonular cataract[11] and confirmation of the linkage between secretor and myotonic dystrophy.[12] It should be noted that all these linkages were with polymorphisms tested by the marker lab. Among disease families that failed to show linkage were several extensive pedigrees of multiple self-healing squamous epithelioma (MSSE).[13] MSSE was mapped later to HSA9q23[14] using DNA markers, but it took a further 17 years before mutations were found in TGFBR1.[15] Review in 1994 of the 1971 Glasgow linkage analysis revealed a low positive lod score with ABO. A number of other disease gene loci were mapped by genetic linkage analysis in Glasgow, including multipoint mapping of the DMD region in Xp,[16] the gene for hereditary persistence of alpha-

[5]Haldane and Smith 1947, 10–21.
[6]Mohr 1951, 339–344.
[7]Renwick et al. 1971, 407–416.
[8]Renwick and Lawler 1955, 312–311.
[9]Donahue et al. 1968, 949–955.
[10]Robson et al. 1969, 1163–1165.
[11]Renwick and Lawler 1963, 67–84.
[12]Renwick et al. 1971, 407–416.
[13]Ferguson-Smith 1934, 267–272.
[14]Goudie et al. 1993, 165–169.
[15]Goudie et al. 2011, 365–375.
[16]Wilcox et al. 1985, 365–375.

Table 1 J. H. Renwick: Glasgow papers on human gene mapping by genetic linkage, 1962–1971

1962	Elliptocytosis and rhesus	Bannerman and Renwick	Ann Hum Genet. 26:23–38
1963	Duffy and cataract	Renwick and Lawler	Ann Hum Genet. 27:67–84
1964	XG and colour blindness	Renwick and Schultz	Amer J Hum Genet. 16:410–418
1965	Intraepithelial dyskeratosis	Pollizer et al.	Amer J Hum Genet. 17:104–108
1965	Nail-patella: parameters	Renwick and Izatt	Ann Hum Genet. 28:369–378
1965	Nail-patella: recombination	Renwick and Schultz	Ann Hum Genet. 28:379–392
1966	Blood groups	Umansky et al.	Vox Sanguinis 11:450–459
1967	Linkage data: storage	Renwick and Bolling	Amer J Hum Genet. 19:360–367
1968	Retinal degeneration	Pearce et al.	Ann Hum Genet. 32:125–126
1968	Duffy blood group: chr 1	Donahue et al.	Proc Nat Acad Sci. 61:949–955
1969	Haptoglobin: chr 16	Robson et al.	Nature 223:1163–1165
1969	White sponge nevus	Browne et al.	Ann Hum Genet. 32:369–374
1969	Angiokeratoma: XG	Johnston et al.	Ann Hum Genet. 32:271–281
1971	Myotonic dystrophy: secretor	Renwick et al.	J Med Genet. 8:407–416
1971	Chromosome heteromorphisms	Wikramanayake et al.	Ann Génétique 14:245–256
1971	Chromosome variations	Renwick	Ann Hum Genet. 35:79–97.
1971	Multiple self-healing epithelioma	Ferguson-Smith et al.	Birth Defects Ser. VII, 8:157–163

fetoprotein to human chromosome 4q,[17] the Emery-Dreifuss muscular dystrophy gene to Xq28[18] and the gene for tuberous sclerosis type 1 to HSA9q33.[19]

In 1961 I returned to Glasgow after 3 years in Baltimore to take up a lectureship with Ponte and to work on human chromosomes. From 1956 to 1959, I had worked on sex chromatin with Bernard Lennox (1914–1997) at the Glasgow University Pathology Department at the Western Infirmary, and our buccal smear surveys had revealed for the first time cases of Klinefelter syndrome in 11% of severe male infertility and 1% of males with learning difficulty. My trip to Baltimore in February 1959 was to improve my attempts to make chromosome preps in these patients, and, as a fellow with Victor McKusick (1921–2008), I trained in medical genetics, ran a chromosome diagnostic laboratory and undertook research on sex chromosome abnormalities with Lawson Wilkins (1894–1963) and Howard W. Jones (1910–2015). We were able to map, by haploidisation, a stature gene and genes controlling the Turner phenotype to the short arm of the X and to

[17]Ferguson-Smith et al. 1985, 628.
[18]Yates et al. 1986, 587–590.
[19]Connor et al. 1987, 544–546.

homologues on the Y chromosome.[20] In other words, Turner syndrome was caused by haploinsufficiency of genes on the X that escaped inactivation in normal women and had active homologues on the Y. This helped to explain the Klinefelter phenotype on the basis of increased dosage of active X-Y-linked genes. Klinefelter patients with more than two X chromosomes had additional skeletal abnormalities and greater learning difficulties. On returning to Glasgow, I revisited and karyotyped our Klinefelter patients and, at the same time, tested their colour vision (and their parents) to determine the origin of nondisjunction; blood was also taken for XG blood group studies in collaboration with Ruth Sanger for the same purpose. Our XG blood group results in Klinefelter patients with a 46,XX karyotype suggested that illegitimate X-Y recombination beyond the pseudoautosomal boundary had resulted in the exchange of the testis-determining locus on the Y for the XG locus on the X and that these two loci must map just outside the boundary.[21] This was confirmed much later by FISH mapping[22] and by the positional cloning of the SRY gene.[23]

The chromosome diagnostic service work in Glasgow and our efforts to improve chromosome identification led to the identification of families with chromosome heteromorphisms potentially useful for gene mapping. This was reinforced by the report of the Duffy: HSA1 assignment.[24] A series of 84 such families were assembled and tested by the marker lab with largely negative results. One family segregating for a centric heteromorphism of HSA16 gave positive lod scores for haptoglobin[25] and this contributed to the haptoglobin assignment made by Robson and colleagues.[26] The remaining negative results were not entirely wasted as they enabled markers to be excluded from many chromosomal regions. The exclusion map was reported by Ferguson-Smith,[27] and additional exclusions from chromosomal deletions were added at later HGM Workshops.[28]

Chromosomal deletions and duplications detected by the cytogenetics lab as part of the West of Scotland Regional Genetic Service provided the raw material for deletion mapping by haploidisation of many of the blood groups and other polymorphisms investigated by the marker lab which I had continued after Renwick's departure to London in 1968. The first ever human gene to be mapped by deletion mapping was red cell acid phosphatase to the distal end of HSA2p.[29] Our patient was a child with developmental malformations and learning difficulties whose

[20]Ferguson-Smith 1965, 142–155.
[21]Ferguson-Smith 1966, 475–476.
[22]Ferguson-Smith 1988, 239–248.
[23]Sinclair Andrew et al. 1990, 240–244.
[24]Donahue et al. 1968, 949–955.
[25]Wikramanayake et al. 1971, 245–256.
[26]Robson et al. 1969, 1163–1165.
[27]Ferguson-Smith et al. 1975, 130–137.
[28]Aitken et al. 1976, 256–265 and Aitken and Ferguson-Smith 1978a, b, 613–617.
[29]Ferguson-Smith et al. 1973, 271–274.

Table 2 Glasgow chromosomal assignments by deletion/duplication mapping

Acid phosphatase	AcP	2p23-pter	*Nature New Biology* 1973, 243:271–274
Adenylate kinase 1	AK1	9q34	*Human Genetics* 1976, 34:35–43
Glutamic-oxaloacetic transaminase	GOTs	10q24–25	*Cytogenet Cell Genet* 1978, 22:468–471
Nucleoside phosphorylase	NP	14q12-qter	*Cytogenet Cell Genet* 1978, 22:490–492
Haptoglobin, alpha	Hpα	16cen-16q22	*Cytogenet Cell Genet* 1978, 22:513
Adenosine deaminase	ADA	20p11-qter	*Cytogenet Cell Genet* 1978, 22:514–517
5S ribosomal RNA	5S	1q42–44	*J Med Genetics* 1979, 16:246–253
Galactose 1-phosphate uridyltransferase	GALT	9p13-cen	*Cytogenet Cell Genet* 1982, 32:24–42
XG blood group	XG	Xp23-pter	*Cytogenet Cell Genet* 1982, 32:273–274
HY antigen	HY	Ycen-qter	*Development* 1987, 101 Suppl.: 157–161

father was a balanced 2p;5q translocation carrier and who was homozygous for the B allele at the AcP locus. The mother was homozygous for the A allele, and the homozygous state of both parents was confirmed by enzyme assay of AcP. The child typed for the A allele and was hemizygous on enzyme assay. It was concluded that the paternal B allele had been lost in the HSA2p23-pter deletion in the child and this interpretation was confirmed subsequently by mapping results by other groups. Additional chromosomal assignments of polymorphic loci followed from our cytogenetics and marker labs, and these are listed in Table 2. Most of these were presented at the Human Gene Mapping Workshops from 1973 onwards. For example, a series of unbalanced HSA9 translocations was highly informative in the precise mapping of both adenylate kinase (AK1) and galactose 1-phosphate uridyltransferase (GALT) to 9q34 and 9p13, respectively.[30] As the AK1 locus was closely linked to the ABO/nail-patella linkage, all three loci could be assigned to 9q34. One GALT family was particularly interesting as the mother with a balanced 9:11 translocation was homozygous for the Duarte variant allele and so had an enzyme activity 50% less than normal.[31] Her two offspring with duplications of 9p13 had GALT levels equivalent to normal resulting from the two maternal variant alleles and the single normal paternal allele. In two other interesting families, glutamate oxaloacetate transaminase (GOTs) was assigned to HSA10q24-25 from unbalanced translocations with breakpoints at 10q23 in one family and 10q26 in the other; a GOTs allele was deleted in the former but not in the latter.[32]

X-chromosome deletions discovered in Glasgow have also been informative for the human gene map. Our large series of Duchenne muscular dystrophy (DMD) families that contributed to the joint project that identified the dystrophin gene[33] included a DMD patient with short stature and learning difficulties who had a

[30]Ferguson-Smith et al. 1976, 35–43; Ferguson-Smith and Aitken 1982, 24–42.

[31]Ferguson-Smith et al. 1982, 24–42.

[32]Aitken and Ferguson-Smith 1978a, b, 468–471.

[33]Kunkel et al. 1986, 73–77.

6 megabase Xp deletion by flow cytometry.[34] In a similar case reported by Franke et al. (1985) and cited by Wilcox et al., the DMD locus was deleted together with loci for retinitis pigmentosa, chronic granulomatous disease and the McLeod syndrome. This deletion was also measured in Glasgow at approximately 6Mb. None of these disorders were found in the Glasgow patient, probably indicating different deletion breakpoints in each patient. In another Glasgow family, a male child born with ichthyosis due to steroid sulfatase (STS) deficiency proved to have a Xp22.3-pter deletion due to a maternal Xp23:Yq13 translocation; he had a normal Y chromosome. The mother was short in stature but had no other features of Turner syndrome. The child's maternal grandparents had normal sex chromosomes, and it seems that the X:Y rearrangement must have occurred at meiosis in the grandfather. This individual had an Xga+ve allele at the XG locus but had failed to pass this to the child's mother who, like her mother, was Xg-ve. This maps the XG and STS loci to within the Xp23-pter deletion in the mother and child.[35] The conclusion is confirmed by STS assay which shows no STS activity in the child and half the expected activity in the mother. The result is of special interest to the Glasgow lab as it supports our 1966 prediction from XX males that the XG locus maps to Xp just outside the pseudoautosomal boundary.[36] Further confirmation came the following year from studies that revealed that 12E7, a gene associated with XG, maps close to the XY pairing segments.[37]

The interest of the Glasgow cytogenetics lab in human sex chromosomes led to a long-term project to make a physical map of the Y chromosome. As crossing over occurs only in the small pairing segments, the map could not be made in the much larger differential segment by genetic linkage analysis, and so recourse had to be made to deletion mapping. Recombinant clones containing single copy Y sequences were isolated from several Y chromosome-specific libraries made from interspecific cell hybrids. These were mapped by Southern blotting to genomic DNA from patients with known Y chromosome aberrations, mostly deletions detected during infertility investigations, or from XX males in which X-Y interchange had transferred variable lengths of Yp to the X.[38] The order of 39 Yp probes was determined in 25 XX males that separated the Y short arm into 17 intervals. A similar map for the Y long arm was constructed with 37 DNA probes that ordered them into 14 separate intervals. Exceptions to the consensus order occurred in 23% of Yp aberrations and 12% of Yq aberrations due mostly to inversions. The resulting map has been useful in determining breakpoints in many patients with presumptive Y aberrations.

In 1969 Pardue and Gall made the first in situ hybridisations (ISH) when they mapped radiolabelled repetitive mouse satellite DNA to the paracentric regions of

[34]Wilcox et al. 1986, 175–180.

[35]Ferguson-Smith et al. 1982, 273–274.

[36]Ferguson-Smith 1966, 475–476.

[37]Goodfellow et al. 1983, 346–349.

[38]Affara et al. 1986, 5353–5373; Ferguson-Smith et al. 1987, 41–50.

mouse chromosomes.[39] An attempt to map the globin genes using labelled messenger RNA from rabbit reticulocytes in 1972 failed because the mRNA could not be made sufficiently radioactive to give a specific signal on human chromosomes. With the advent of recombinant DNA technology, a joint Glasgow project with Robert Williamson (b. 1938) from the Beatson Institute, and funded by the MRC, revisited the possibility of mapping single copy genes by ISH. We first developed the method using tritiated thymidine labelled ribosomal RNA from *Xenopus laevis*, cloned in a plasmid vector and detected on metaphase acrocentric chromosomes by autoradiography using liquid photographic emulsion.[40] This success was followed by making cRNA probes from cloned genomic DNA of both β- and α- globin genes which hybridised successfully to the short arms of HSA11 and HSA16, respectively.[41] These proved to be the first regional assignments of single genes by ISH. We then mapped the kappa light chain immunoglobulin gene locus to HSA2p later the same year.[42] Among the many applications of ISH, the mapping of cloned Y chromosome probes to the end of the short arm of one of the two Xs in XX males was particularly satisfying.[43] Radioactive labelling of probes was soon replaced by fluorescence labelling, and FISH then became the most widely used method for gene assignment.

In 1987 several of those involved in human gene mapping in Glasgow moved to appointments at the Cambridge University Department of Pathology and worked on a programme entitled the Molecular Pathology of Disease funded by the MRC. Research continued on human gene mapping, positional cloning of disease genes and comparative genomics. Contributions from Glasgow and Cambridge after this date are recorded elsewhere.

It may be of interest to note that many of the cytogenetic approaches described in this article depend on nondisjunction and haploidisation, two of the three elements advocated by Pontecorvo 60 years ago as likely to be the most productive in the construction of a human gene map. Glasgow can be proud of its contributions to this endeavour.

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[39]Pardue and Gall 1970, 1356–1358.

[40]Malcolm et al. 1977, 256–261.

[41]Malcolm et al. 1981, 135–141; Barg et al. 1981, 124 and 1982, 252–253.

[42]Malcolm et al. 1982, 4957–4961.

[43]Ferguson-Smith 1988, 239–248.

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Human Gene Mapping: The Mass Media Iconography of the Human Genome Project in the Most Popular Greek Newspapers

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Abstract The media serve as intermediaries between science and the public, framing social reality for their readers and shaping the public consciousness of science-related events. In this context, mass media iconography, for instance, the "genetic map," plays an important role in the public communications of science and technology. It facilitates the understanding of an often abstruse technoscientific discourse and a complex experimental methodology. Powerful and potent iconography can achieve the beautification of a technoscientific fact or can underscore widespread public concerns and open resistance to it more effectively than any words. As a part of a journalist's routine, iconography is used for the purposes of popularizing, concretizing, and dramatizing issues, in brief for making issues both newsworthy and interesting for the public audiences. Generally, the iconography in the mass media through its rhetoric and ideological charge often contributes to shaping public opinion (positive or negative) on a scientific fact or a new technology.

The Human Genome Project (HGP) is one of the most important scientific events covered by the media, and the public attention which it has received has helped to change the relationship between science and society. The purpose of this paper is to present a review of the results of a case study that focuses on the media iconography relating to the HGP and human genome sequencing in the most popular Greek newspapers and how its use has affected science and technology communication. In particular, examined here are a series of selected photographs, digital depictions, infographics, illustrations, and cartoons that accompanied and framed the publications which have contributed to the development of a specific public image for HGP.

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The research has showed that the HGP attracted intense coverage from the most popular Greek newspapers. The rhetoric and the framing of publications for HGP compared it to the greatest moments of social, artistic, and scientific developments, and the iconography was chosen by these newspapers to strengthen the "positive" media framing for HGP and shape a general "positive" public image for this. Furthermore, this paper suggests that the study of the mass media iconography for biosciences and biotechnology is a challenge for those interested in the effective communication of bioscientific developments. Researchers from disciplines like Science Communication (SciCom) and Science, Technology, and Society (STS) could contribute to the effectiveness of such efforts by turning a critical eye toward the functions, purposes, and effects of iconography in science communication.

Keywords Human Genome Project • Greek newspapers • Public image of science and technology • Science communication

1 Introduction

Scientific and technological developments have a fundamental importance and influence on the structure and organization of modern societies, being a constituent element of modern culture.[1] According to Bijker: "The stories we tell about [science and] technology reflect and can also affect our understanding of the place of [science and] technology in our lives and our society.[2]" In other words, technoscience is socially shaped, and society is technoscientifically shaped.[3] In this context, the mass media play a significant role in shaping the public image of science and technology, exerting influence on public support and the funding of research programs, and promoting the idea of a knowledge-based society. For the majority of people, the complex world of science and technology is a "black box." Because of this, as has been pointed out by Nelkin, author of the classic book *Selling Science: How the Press Covers Science and Technology*,[4] "the [mass] media serve as brokers between science and the public, framing social reality for their readers and shaping the public consciousness about science-related events. They are, for many readers, the only accessible sources of information about science and technology. Through their selection of news, journalists help to set the agenda for public policy. Through the information they convey about risks, they may affect stock market prices and influence product sales. And through the presentation of science news, the media influence public attitudes towards sci-

[1] See Sweezy and Magdoff 1991, 1–15.; Giddens 1990, 55–56; Castells, 1989, 12–15, 17–19, 28–32.

[2] Bijker 1995, 1.

[3] Bijker 1995, 288.

[4] Nelkin 1995.

ence.[5]" Although the mass media do not frequently tell us directly what opinions to have on a problem, they have a significant effect on telling us what the issues and topics are that we need to have an opinion on.[6]

From daily news reports concerning the space exploration in US newspapers and the TV broadcast of the "Apollo 11" Moon landing (in 1969) to the triumphant public presentation of Dolly, the first mammal cloned from an adult sheep cell (in 1997), and the announcement of the completion of the human genome "working draft" (in 2000), the interest of the mass media coverage of issues related to technoscientific developments and changes has increased in recent decades. It is indicative that the rapid developments in biosciences and biotechnology during the last 50 years have contributed to the flourishing of a wide public debate about gene therapy, stem cell research, genetic manipulation, DNA fingerprinting, cloning, and a number of other issues in which the main actors attempt to influence and dominate in terms of the expressions and the discourse that is articulated.[7] Furthermore, the biosciences and biotechnology play an increasingly dominant role within technoscience and have become a focal point of media attention when it comes to research initiatives that shape their course. As aptly noted by Kohring and Matthes: "Since it is not possible to acquire direct experience of biotechnology—genetically modified soya does not alter its shape and Dolly the sheep continues to be a sheep (and we cannot see or feel the difference in either)—media coverage plays an important and distinct role in shaping the public perception(s) of modern biotechnology.[8]" In this context, since the early 1990s, a large number of studies have focused on analyzing the characteristics and the formation of public debate, the media coverage, the public image, and the cultural history of bioscience and biotechnology.[9] An additional set of studies has focused on the power of the mass media to define public issues surrounding biotechnology and the role played by the selective framing of biotechnology stories.[10]

In conjunction, the iconography in the mass media acts as a powerful means of communication. A powerful and potent iconography can achieve the beautification of a technoscientific fact or can underscore widespread public concerns and open resistance to it more effectively than any words. It facilitates the understanding of an often obscure technoscientific discourse and/or an elaborate experimental methodology. Moreover, it portrays scientists at work or at the time of their scientific triumph, makes the microcosm visible, and depicts our biological structure. Generally, the iconography in mass media, through its rhetoric and ideological charge,

[5]Nelkin 2001, 205.

[6]McCombs 1991, 12.

[7]See Hansen 2006.

[8]Kohring 2002, 143.

[9]See van Dijck Jose 1998; Nelkin 1996, 2001; Turney 1998a, 1998b; Listerman 2006; Squier 2004; Conrad 2001. From the Greek bibliography, see National Hellenic Research Foundation, 1997, 1999.

[10]See Kohring 2002; Nisbet 2002; Ten Eyck 2003; Listerman 2010.

often contributes to shaping public opinion (positively or negatively) on a scientific fact or a new technology. Finally, the iconography is part of a journalist's routine and is used for the purposes of popularizing, concretizing, and dramatizing issues, in brief for making stories both newsworthy and interesting for their audiences. More particularly, the "genetic iconography" in mass media (representations of the double helix of DNA, the cloned Dolly, genetically modified plants and animals, the "Frankenfood," the "human genome map," etc.) and in public culture (science fiction books as *Brave New World*, *Oryx and Crake*, *Next*, etc.; films like *Gattaca*, *Blade Runner*, *Jurassic Park*, *Cloned*, etc.; comics like *Megalex*, Y*:* T*he Last Man*, *DNAgents*, *X-Men*, etc.; video games like *Bio-Attack*, *BioShock* etc.; and BioArt exhibitions) expresses the new horizons the biosciences has expanded to, but has also become a symbol of the dual-sided nature of biotechnology, the promises, and dilemmas.

The Human Genome Project (HGP) is one of the most important scientific events which have been covered by the media, and the public attention which it has received has helped to change the relationship between science and society.[11] The HGP emerged through a long accumulation of technoscientific changes. Well-known episodes in this accumulation are the rediscovery of Mendel's laws in 1900, the 1940–1970 studies that lead to certain knowledge regarding the structure of the DNA, the 1970–1990 development of techniques that allow for the creation of recombinant DNA and genome sequencing, and, more recently, the sequencing of the human genome (1990–2000) on the grounds of an appropriate articulation of scientific, political, and economic factors. On June 26, 2000 there was the triumphal announcement of the completion of the human genome "working draft." In February 2001, the scientific journals *Nature* and *Science* announced that the number of human genes was about 30,000, while in April 2003 the formally completed sequencing of the human genome was fully recorded. New programs for the genome sequencing of other organisms are taking place at a plethora of scientific institutes and organizations between 2000 and 2009.

In Science Communication (SciCom), a series of studies came to examine the extensive media coverage of human genome sequencing in the USA and North-Central European countries. Characteristic is the study by Henderson and Kitzinger, whose research was based on the analysis of news reports in the British media; this argued that the HGP announcement can be seen as "a valuable case study in which the worlds of science, media and policy came together in a common goal.[12]" According to them, the main characteristics of HGP media coverage can be summarized as follows: (a) the graphic of the coverage is not static but takes the form of a bell curve, the peak of which is located around 2000[13]; (b) journalists specializing in science-technology coverage were much more cautious in regard to

[11]For more about HGP, see https://www.genome.gov/10001772/all-about-the--human-genome-project-hgp/. Accessed 27 May 2016.

[12]Henderson 2007, 80.

[13]Henderson 2007, 79–80.

cultivating expectations from the HGP than other journalists[14]; (c) amidst overall positive coverage, especially during the period June–July 2000, there were reports that focused on the broader social, legal, and ethical issues arising from the sequencing of the human genome[15]; and (d) the main linguistic metaphors that portrayed the HGP had to do with discovering the book of life, the book of humankind, the genetic book, etc.[16] In another comparative study with broad reach, Costa researched how the mass media have handled HGP in four different countries: the USA, France, the UK, and Italy.[17] The study covered five newspapers—*The New York Times*, *Le Monde*, *The Independent*, *Corriere della Sera*, and *la Repubblica*—for the period 1990–2001. Quantitative analysis showed that there was an increase of articles on HGP in all these newspapers that reached a peak in 2000 before following a marked decline. Qualitative analysis revealed important differences in the coverage. O'Mahony and Shäfer, in their own study, compared German and Irish media coverage of human genome research in the year 2000, analyzing articles from the largest-selling national broadsheets: *Süddeutsche Zeitung*, *Frankfurter Allgemeine*, *Die Welt*, *The Irish Independent*, *The Irish Times*, *The Irish Examiner*, and *The Sunday Business Post*.[18] They found that even though there is a globally networked media system, which tends to produce uniform media coverage on the global implications of advances in the biosciences, for the global audience, several national or local actors can cause variations from this. One more suggestive study is by Gerhards and Shäfer, which examined how two normative models of science in the public sphere—the "science-dominated scientific public sphere" and the "contextualized scientific public sphere" model—may be applied to the understanding of the media coverage of human genome sequencing in selected German and US broadsheet newspapers: *Süddeutsche Zeitung*, *Frankfurter Allgemeine Zeitung*, *The Washington Post*, and *The New York Times*.[19] The study covered the 1999–2001 period. It confirmed that reporting on human genome research was extensive in both countries. Also, a special group of studies focused on the role of metaphors in journalistic discourse, especially metaphors used to popularize the complex nature and properties of the genome. This group of studies paid special attention to the role and the importance of linguistic metaphors used for the genome in journalistic discourse.[20] While several studies have explored the media coverage of the human genome sequencing and the HGP, there has been little discussion and few studies focusing on the iconography of HGP in the mass media. Also, very little attention has been paid

[14]Henderson 2007, 78.
[15]Henderson 2007, 67–68.
[16]Henderson 2007, 70.
[17]Costa 2003, 2.
[18]O'Mahony 2005.
[19]Gerhards 2009.
[20]See Hellsten 2002, 2005, 2008; Doring, 2005; Rödder, 2009; Nerlich 2004; Calsamiglia 2004.

to how the iconography of HGP in the mass media can be understood as part of the process of science communication for public audiences.

The aim of this paper is to present a review of the results of a case study that focuses on the media iconography used for HGP and human genome sequencing in the most popular Greek newspapers and how its use has affected science and technology communication. Specifically, examined here are a series of selected photographs, digital depictions, infographics, illustrations, and cartoons that accompanied and framed the publications which have contributed to the development of a specific public image for HGP. It should be noted though that this article is part of a wider research program into the public image of the HGP and the genome sequencing in the most popular Greek newspapers.[21] Greece is a valuable case study because it is a non-English-speaking country, European-Western but peripheral, and non-Catholic but Christian. It is a small country, where the transition from a predominantly rural to an urban industrial and service society occurred relatively late. Greek research institutes do not participate in human genome sequencing directly—one notable exception is the Laboratory of Molecular Medicine and Human Genetics at the University of Crete, which had been assigned the task of sequencing 15% of the human chromosome 10. Although technoscientific research in Greece is considered as providing an opportunity to develop the economy, it is extremely underfunded in both the public and the private sector.[22] As for the Greek mass media network, it is pluralistic, with many national and local media outlets that cover the whole of the political spectrum.[23] In this context, the media coverage of HGP in the Greek press has been inversely proportional to the country's limited participation in this research field.[24]

2 Data and Methods

For the purpose of this research, an extensive set of publications (textual and visual) were analyzed; these were obtained from the electronic databases of three of the most popular and largest-selling national Greek newspapers, *To Vima* (Το Βήμα), *Ta Nea* (Τα Νέα), and *I Kathimerini* (Η *Καθημερινή*). They are regarded as the Greek equivalent of newspapers like *Le Monde* in France, *The New York Times* in the USA, *El País* in Spain, *Corriere della Sera* in Italy, and *Süddeutsche Zeitung* in Germany. A series of photographs, pictures, digital depictions, diagrams and tables, infographics, illustrations, cartoons, and generally everything in iconographic material that has been published in these three newspapers have been studied.

[21] See Morfakis 2013.

[22] Arapostathis 2010.

[23] On the Greek media system and its coverage of general and special technoscience. See Mergoupi-Savaidou 2012, Tympas 2010.

[24] Morfakis 2013.

The whole of the newspaper, main layout and inserts, is looked into, except magazines distributed with the newspaper. All the different sections of the newspaper—political, international, world news, science/technology, health, culture, sports, economy, and developments—were researched. Publications containing the keywords "genome mapping" and "genome decoding program" were searched for about a quarter of a century, from 1986 to 2009. This corresponds to the years when the HGP was perceived (1986), implemented, and completed (2003). It also covers subsequent research into metagenomics and genomes sequencing by other organizations (up to 2009). The electronic archive of *To Vima* covers the period 1985–2006, of *Ta Nea* the period 1985–2009, and of *I Kathimerini* the period 2000–2003. The search resulted in a total of 313 publications, all of which were taken into account. To explore the role of media infographics in shaping a specific public image for HGP requires a multidisciplinary approach that leverages studies and tools from the fields of SciCom, framing analysis, content analysis, and semiotics.

SciCom studies provide us with the appropriate theoretical and methodological tools needed for the analysis of media coverage, media framing, and the public image of science and technology. As mentioned above, there are a series of studies that have examined the extent of the media coverage of human genome sequencing and HGP in several Western countries. The results of these studies are useful theoretical tools that are utilized in this paper.

Framing analysis has been proved to be especially appropriate for analyzing the public image of technoscience as shaped by the media.[25] To be sure, there are different and even competing definitions of the concept of a media "frame," just as there are of the "process of framing." According to Gitlin, using the concept of the "frame" "enable[s] journalists to process large amounts of information quickly and routinely [and to] package the information for efficient relay to their audiences.[26]" For Gamson and Modigliani, a frame is a "central organizing idea or story line that provides meaning" to events related to an issue.[27] Entman finds that framing means selecting "some aspects of a perceived reality and mak[ing] them more salient in a communicating text, in such a way as to promote a particular problem definition, causal interpretation, moral evaluation and/or treatment recommendation.[28]" For the purposes of this paper, we used the media framing typology for biotechnology established by Durant, Bauer, and Gaskell, which has been enriched by Nisbet and Lewenstein and several other scholars and which offers a good basis for comparison of global and regional media frames for biotechnology.[29] Based on these studies, a table is presented with a basic framing typology for biotechnology (Table 1).

Content analysis as well as semiotics was utilized in the elaboration and analysis of the iconographic material that was detected in the most popular national Greek

[25]See Entman 1993; Pan 1993; Scheufele 1999, 2000; Goffman, 1974; Gamson 1989; de Vreese, 2005; d'Angelo 2002.

[26]Gitlin 1980, 7.

[27]Gamson 1987, 143.

[28]Entman 1993, 52.

[29]See Durant 1998; Nisbet 2002; Ten Eyck 2003; Listerman, 2010; Kohring 2002.

Table 1 A framing typology for biotechnology

	Durant et al. (1998)	Nisbet and Lewenstein (2002)	Kohring and Matthes (2002)	Ten Eyck and Williment (2003)	Listerman (2006)
Media Frames	Progress	Progress	Agri-food: pros and cons	Progress	Utility
	Economic prospect	Economic prospect	Research in biomedicine	Economic prospect	Risk
	Ethical concerns	Ethical concerns	Biomedicine as moral risk	Nature/Nurture	Control
	Public accountability	Pandora's box	Profits of biomedicine	Public accountability	Fate
		Runaway [technology]	Regulation for economy	Ethical concerns	Morality
		Nature/Nurture		Runaway technology	
		Public accountability	Biomedicine for health	Pandora's box	
		Globalization	Agri-food regulation		
			Regulation of identity		
			Research as benefit		
			Regulation for economy		
			Economic prospects		

Sources: Durant et al. (1998), Nisbet and Lewenstein (2002), Kohring and Matthes (2002), Ten Eyck and Williment (2003) and Listerman (2006)

newspapers.[30] In particular, the classic essays of Barthes, "The Photographic Message" and the "Rhetoric of the Image," were useful methodological tools.[31] The collective volumes of Kress and van Leeuwen and van Leeuwen and Carey and the book of Gross are a rich methodological resource in investigating the visual representation and public image of socially significant issues.[32] Also, for the purposes of this paper, two important works were exploited. The first study is that by Jacobi and Schiele, which focused on the role of imagery in portraying science and scientists in *Science et Vie* and *La Recherche*, two French magazines.[33] In particular, they have elected to analyze a very specific type of illustration: the photographic portrait of the scientist. This case study is an example of imagery

[30]See Krippendorff 2008; Wimmer 2005.

[31]Barthes, 1977, 15–31, 32–51.

[32]Kress 1996; van Leeuwen 2001; Gross 1994, 1996.

[33]Jacobi 1989.

analysis. The other study, by Giarelli, examines how the popular mass medium of cartoons in the USA contributes to shaping a public image about some of the commonly held beliefs about cloning and stem cell research. She claims that "analyzing popular images can allow access to public understanding about genetic technology and evaluation of public beliefs, preconceptions, and expectations as the public is educated on the use and value of services.[34]"

3 The Mass Media Iconography of HGP in the Most Popular Greek Newspapers

The iconographic material of a publication contributes to more complete communication and framing of information about a technoscientific fact. It can condense the journalistic discourse in a "dynamic" image. Also, it can express scientific principles, experimental data, or innovations and can help the article to convey meaning or to clarify ideas. An effective iconography is a stupendous tool in the communication of science and technology to both experts and public audiences. Mass media iconography includes a diverse stylistic and technical range within a number of categories of icons. Photographs, pictures, digital depictions, symbolic notation, diagrams and tables, infographics, illustrations, and cartoons are among the choices from which the journalist may select in an effort to "show" rather than "tell" about the science and technology. Each form of this iconographic material carries its own conventions and potential for interpretation or misinterpretation.[35]

The analysis of publications revealed six general classification categories of iconography that accompany and frame articles concerning human genome sequencing and HGP in the most popular Greek newspapers. These categories are presented and analyzed in this section in order to show how the iconography in the media creates a particular public image of HGP.

3.1 HGP (Bio)scientists' Portraits

The first category of iconography includes "photographic portraits" of (bio)scientists who have a leading or important role in the history of human genome sequencing and HGP. According to Jacobi and Schiele, "scientists are never portrayed in primary scientific journals, and it is out of the question to publish photographic portraits. The reason for this is easy to understand: science is enunciated without reference to the enunciator. The author disappears behind an object that seems to speak for itself, or write itself out independently. [...] Popular

[34]Giarelli 2006, 61.

[35]See Trumbo 1999.

magazines, on the other hand, make the scientist come alive. They need heroes, not so much to make science understood as to make it appealing and attractive. Through photographic portraits, knowledge finds itself conveniently anchored and relayed. The anchor selected is that of the laboratory: a photograph graphically displays the paraphernalia of science. [. . .] Popularization [. . .] personalizes knowledge by attributing it to its inventor [. . .].[36]" They continue, saying that "the researchers agree to cooperate with the popularizers and pose for their photographers" because "their aim is a subtle combination of a desire to make their findings known to the scientific community, on the one hand, and the temptation to use less professional methods of publicizing and highlighting their findings in circles outside their own.[37]"

The analysis of a sample of the (bio)scientists' portraits in the most popular Greek newspapers gave four archetypes of the scientist: (1) the "scientist-hero"—the scientist who has made an important discovery poses along with a physical object that attests to his/her historic and "revolutionary" work; (2) the "scientist-academic"—the scientist leaves the laboratory and his special attributes as an experimenter behind and poses in his/her official office or classroom, transforming him/her into a scholarly professor; (3) the scientist as an "everyday person"—the scientist is photographed at home in a familiar, almost intimate pose; and (4) the "scientist-businessman"—the scientist is photographed at his/her business office or with investors and other entrepreneurs.[38]

In one of the most frequently used photographs, Craig J. Venter (Figs. 1 and 2),[39] one of the main actors in the human genome sequencing, as the company's chief Celera Genomics Corp., is wearing a lab coat and, looking toward the viewer, poses with his scientific background achievement, a "blueprint" of the genome, on which his shadow falls. The rhetoric of this slightly art photography attempts to induce in the readers the feeling that Venter is the "creator or ruler" of the "blueprint" of the human genome that it is he who is controlling the "secrets of life." Furthermore, in this photo, the "blueprint" of the genome is in the background in relation to the (bio) scientist, suggesting indirectly that Venter's personality overshadows the scientific achievement or stands in an equal relation with it. In this photo, more importance is given to the researcher than to the scientific fact of genome sequencing itself. In addition, the way in which this photo was taken makes it possible to create various associations of ideas in its viewers and readers. From another semantic point, the photography implicitly likens the "blueprint" of the genome to a sheet music and the shadow of the (bio)scientist to the shadow of a composer (Venter-"Mozart") in a way of performing the "life (evolution) *partitura*." It is noteworthy that this photo is used repeatedly in the accompanying articles on the human genome sequencing and

[36]Jacobi 1989, 750–751.

[37]Jacobi Schiele, 1989, p.751.

[38]For more about archetypes images of the scientist, see Jacobi 1989, 739–750.

[39]For more information about Craig J. Venter, see http://jcvi.org/cms/about/bios/jcventer/. Accessed 22 May 2016.

Δρ ΚΡΑΙΓΚ ΒΕΝΤΕΡ

Ο Πόλεμος των Γονιδίων

ΠΡΟΣΩΠΑ

Ο δρ Κραιγκ Βέντερ φιλοδοξεί να γίνει ο Μπιλ Γκέητς της γενετικής· και εξασφάλισε τα ιδιωτικά κεφάλαια για να το επιτύχει.

Δρ Βέντερ. Φιλοδοξεί να γίνει ο Μπιλ Γκέητς της γενετικής

ΕΠΙΚΕΦΑΛΗΣ του αμερικανικού Ινστιτούτου Γονιδιακών Ερευνών, ο δρ Βέντερ σχεδιάζει να αφιερώσει τα 92 δισεκατομμύρια δραχμές που συγκέντρωσε από ιδιωτικά κεφάλαια στην προσπάθεια να χαρτογραφήσει το γονιδίωμα του ανθρώπου – την έλικα του DNA, που είναι το σχεδιάγραμμα της ζωής. Θέλει να είναι ο πρώτος που θα ολοκληρώσει αυτή τη χαρτογράφηση και θα κατοχυρώσει με δίπλωμα ευρεσιτεχνίας όποια χρήσιμα γονίδια βρει, ώστε να μπορεί να τα χρησιμοποιεί όπως επιθυμεί. Ο Πόλεμος των Γονιδίων έχει ήδη κηρυχθεί.

Στην Αμερική, τα ανθρώπινα γονίδια με συγκεκριμένη λειτουργία και εφαρμογή μπορούν να κατοχυρώνονται με δίπλωμα ευρεσιτεχνίας και την περασμένη εβδομάδα το Ευρωκοινοβούλιο αποφάσισε ότι το ίδιο θα γίνεται και στην Ευρώπη. Υπάρχουν κάποιες ασφαλιστικές δικλίδες – ουδείς μπορεί να κατοχυρώσει ένα γονίδιο, έστω κι αν γνωρίζει τη λειτουργία του, αν δεν μπορεί να δείξει ότι σχεδιάζει κάποια εφαρμογή γι' αυτό, ενώ όλες οι ανακαλύψεις θα κατατίθενται στις σελίδες του Ίντερνετ. Ωστόσο, οι επικριτές της απόφασης επισημαίνουν ότι είναι η πρώτη φορά που δίδεται δίπλωμα ευρεσιτεχνίας για ανακάλυψη μάλλον, παρά για εφεύρεση και πως ο κάτοχος της «πατέντας» ενός γονιδίου θα μπορεί να εμποδίζει άλλους επιστήμονες να δουλεύουν μ' αυτό.

Ως την περασμένη εβδομάδα, ο δρ Βέντερ συμμετείχε σε μια εν πολλοίς αλτρουιστική διεθνή συνεργασία, με τη συμμετοχή της Βρετανίας, των ΗΠΑ, της Γερμανίας, της Γαλλίας και της Ιαπωνίας για τη δημιουργία μιας βάσης δεδομένων των 60.000 έως 100.000 γονιδίων που υπαγορεύουν τη διαμόρφωση και τη λειτουργία του σώματος. Τώρα προσπαθεί να φτιάξει αυτή τη βάση για τον εαυτό του, με τη βοήθεια της Perkin-Elmer, μιας αμερικανικής εταιρείας κατασκευής επιστημονικών εργαλείων. Τα μηχανήματα που διαθέτει, υποστηρίζει, μπορούν να χαρτογραφήσουν σε μόλις τρία χρόνια το 97% του ανθρώπινου γονιδιώματος που παραμένει αχαρτογράφητο. Ο δρ Βέντερ θα ξεχωρίσει και θα απομονώσει τα κερδοφόρα γονίδια – τους κωδικούς του διαβήτη, των γηρατειών και όλων των ενζύμων που κάνουν το σώμα μας να λειτουργεί – και θα κατοχυρώσει την ανακάλυψή τους ως ευρεσιτεχνία. Οι άλλοι επιστήμονες θα μπορούν να τα χρησιμοποιούν για καθαρή έρευνα, αλλά για εφαρμοσμένη έρευνα, όπως κλινικές δοκιμές, θα απαιτείται η άδεια του δρος Βέντερ. Αν, για παράδειγμα, βρει ένα γονίδιο που να προδιαθέτει σε κάποιας μορφής καρκίνο, όλος ο κόσμος θα πρέπει να του πληρώνει δικαιώματα για να το χρησιμοποιεί.

Όπως περίπου συνέβη στον τομέα της πληροφορικής με τον Μπιλ Γκέητς, τη Μάικροσοφτ και το MS-DOS.

ΕΠΙΜΕΛΕΙΑ: **Γιώργος Αγγελόπουλος**

Fig. 1 Source: Angelopoulos Giorgos, "O polemos ton gonidion. Dr. Craig Venter" (The war of genes. Dr. Craig Venter), Ta Nea, May 19, 1998, 48

thereby becomes dominant in the collective imagination as a public image of HGP. However, the depiction of a scientist posing with the object that certifies his/her work and his/her contribution to society is one of the classic images of scientists.[40]

Simultaneously, another photography shows Venter in a (bio)scientific laboratory (Fig. 3). This is a typical portrait of a scientist in his/her workspace. He wears a white lab coat and represents a modern (bio)scientist. But this focuses on a single person has, as a consequence, to ignore or overlook the contribution of a plurality of scientists who have worked behind the lights of the camera in the scientific laboratories. This is especially true when we refer to the research about the human genome sequencing and the HGP, which is the outcome of a team of several cooperating scientists. Nevertheless, the futuristic style of this photo of Venter, named the "Bill Gates of the genome," according to the article, might remind us of snapshots of the

[40] Jacobi 1989, 739.

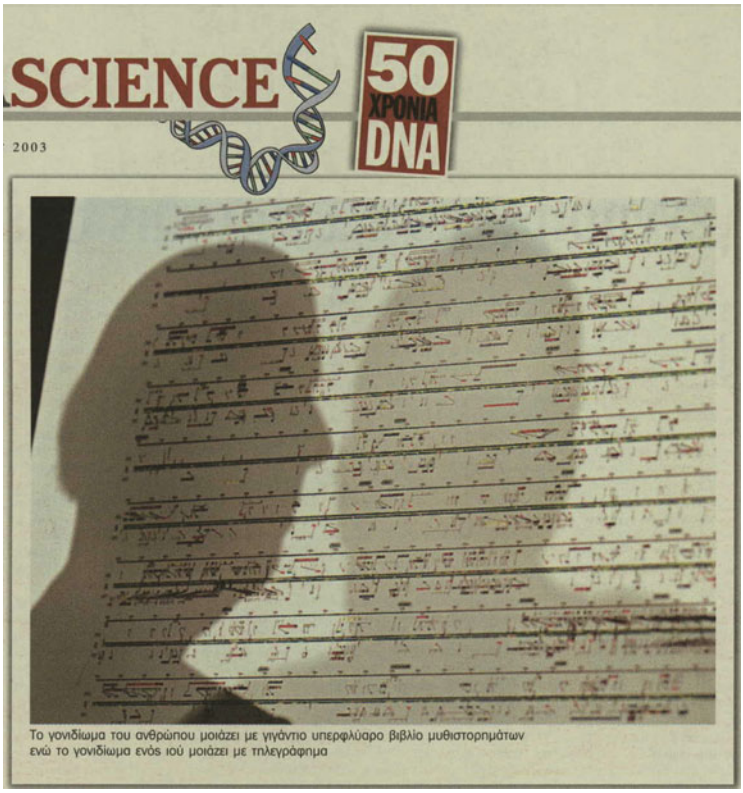

Fig. 2 Source: Tsaftaris Athanasios, "I grammatiki tou DNA" (The DNA grammar), To Vima, March 2, 2003, 8 (VimaScience) and Alachiotis Stamatis, "Ti krivi to Vivlio tis Zois" (What hides the Book of Life), To Vima, February 18, 2001, 65

television science fiction series *The X-Files*, giving to Venter a "mysterious aura" as a researcher into "life secrets." Another reference to science fiction is that in the article in which the photo is published implements a comparison between Venter and "Darth Vader," thus likening the "*Star Wars*" to the "Genome War."

In one more photo (Fig. 4), Venter is depicted with his collaborator Hamilton O. Smith.[41] In this portrait he appears as a jovial and sociable man, more casual, perhaps relaxing in his office at the company Celera Genomics Corp. The ordinary and intimate poses shape our public image of the scientists as a common public figure. These photos have no connection to the laboratory-type photographs. As pointed out by Jacobi and Schiele, a photo of this type "is looking for another angle, striving to portray the person not merely as a scientist, but as an everyday human being with tastes, sensations, preferences, a way of living and of loving. [. . .] The

[41]More information about the American microbiologist and Nobel Laureate Hamilton O. Smith http://jcvi.org/cms/about/bios/hsmith/. Accessed 22 May 2016.

Fig. 3 Source: Galatsatos Panagiotis, "Craig Venter," Ta Nea, April 22, 2000, 18–19

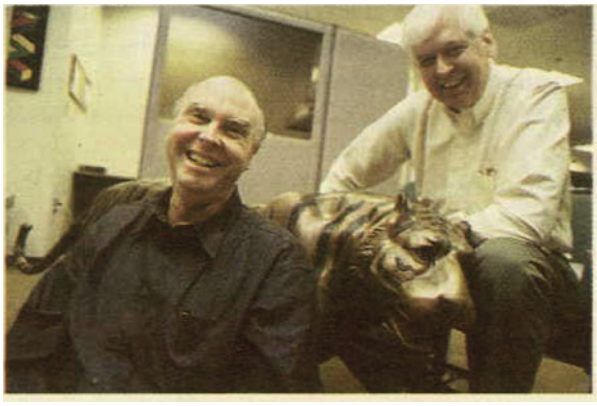

Fig. 4 Source: Anon., Craig Venter. "O 'perithoriakos' pou espire ton antagonismo" (The "marginal" who sowed the competition), Ta Nea, December 11, 1999, 19

scientist gets down from a pedestal and mixes with the rest of us. The photo suggests, not a mind devoted solely to science, but a person of flesh and blood.[42]" Moreover, this photography shapes an additional public image of scientist, which the reporters of the most popular newspapers reproduce—the image of "scientist-businessman" emerging in the "era of biotechnology."

[42]Jacobi 1989, 748.

These (bio)scientists' portraits contribute to establishing a particular public image about HGP. The scientists' portraits "have the function of giving science a face.[43]" These "both justify scientific discourse and make its reality felt, since science is defined by the people who produce it,[44]" but they have no connection to the public image of a "mad scientist," which has been shaped by public culture and the science fiction novels. Therefore, the media frame of (bio)scientists' portraits is that of "technoscientific 'progress' and the utility derived from it." This media framing has, as a result, the shaping a "positive" public image for the HGP, promoting the public euphoria and emphasizing the expected benefits from this "revolutionary" scientific advance.

3.2 HGP and (Bio)informatics

The second category of iconography highlights the special relationship and interconnection between the HGP and (bio)informatics. This interlocking of biosciences and (bio)informatics has a long history.[45] According to Olson, Professor of Medicine and Genome Sciences, as well as Director of the Genome Center at the University of Washington in Seattle, USA, "The Human Genome Project is the direct descendent of the wholly unexpected confluence of genetics and information theory. In a 1954 *Nature* paper, the cosmologist George Gamow pointed out, apparently for the first time, that 'the hereditary properties of any given organism could be characterized by a long number written in a four-digital system'. The term 'four-digital', soon to be replaced by 'base four', sounds quaint to the modern ear. This archaism is a colorful reminder that both molecular biology and information theory were then young. The confluence of genetics and computer science must rank as one of the great coincidences in the history of science and technology. In the same historical instant, humans discovered that biological information is digital—a mechanism of information storage and processing that evolved within cells over billions of years—and, quite independently, invented new technological means of storing, processing, and transmitting information based on digital codes. Thus, the two technological forces that are most profoundly reshaping the future of human culture—genetics and computing—are linked at their historical and conceptual roots.[46]"

In particular, the interconnection of HGP and (bio)informatics depicted in the public image of the (bio)scientific laboratory in the "era of biotechnology." In a photograph (Fig. 5) we see, as we are informed by the "linguistic message" of the

[43] Jacobi 1989, 739.

[44] Jacobi 1989, 749.

[45] See Rifkin 1998, 175–196.

[46] Olson 2002, 932.

Fig. 5 Source: Soufleri Ioanna, "I epanastasi tis Biologias" (The revolution of biology), To Vima, June 4, 2000, 58

caption, a technician (Zuneba Nuri) working in front of the consoles of "powerful" computers through which the human genome sequencing is being carried out. The (bio)scientific laboratory has been completely transformed as the vials, pipettes, test tubes, containers with reagents, optical microscopes, etc. are absent. In this laboratory, there is nothing beyond computers and complex computer programs. The rhetoric of the photograph is the message that everything is now left, not to the skilled manipulation and the ability of (bio)scientists, but to the "computational power" of computers. The (bio)scientists conduct their experiments using computers and interpret the results obtained. However, this produces a spurious public image of the workload and the process required for the human genome sequencing. The rhetoric of the image is such that it implies that, through the use of specialized computer programs, fewer scientists are required to work on the human genome sequencing. This is in stark contrast to the actual workload of the thousands of (bio) scientists who worked in the HGP.

Also, another digital depiction (Fig. 6) shows a part of the "decoded" human chromosome 10. In the "linguistic message" of the caption, which complements and enhances its rhetoric, we read: "The DNA microarrays give information about the genetic identity of each human. In each dot there is the sequence of a gene. Genes that are functional (expressed in the language of biology) are red in color, genes which are not expressed are green and genes with decreased expression are yellow. So, with a single experiment, the functionality of hundreds of genes is

Fig. 6 Source: Soufleri Ioanna, "Stin Kriti apokodikopiisame to chromosoma 10" ("In Crete chromosome 10 has been decoded") To Vima, December 12, 1999, 58–59

monitored simultaneously" (translated by the author).[47] In other words, the message is that the active genes (red) are those read from the "computer cell" and which provide the necessary information for the functioning of the human body/organism. This digital depiction with the black background and the bright dots reminds us, metaphorically, of a "digital punched card" by analogy with the punched cards of the classical computer (Fig. 7). In this way, the particular iconographic material, in combination with the dominant use of the linguistic metaphor of the genome as a "code" and the sequencing as "encoding process," contributes to shaping a public image of the HGP as another "computer program.[48]" Therefore, the significant role that (bio)informatics plays in the human genome sequencing and how interconnected these scientific areas are is immediately understood. The public image of the genome as a dataset and as a "computer program" is supported by the use of this type of iconographic material.

[47]Soufleri Ioanna, "Stin Kriti apokodikopiisame to chromosoma 10" ("In Crete chromosome 10 has been decoded", T*o Vima*, December 12, 1999, 58–59.

[48]For the linguistic metaphor of the "code", see Gogorosi 2005, 302–305; Calsamiglia 2004, 376–379; Nerlich 2004, 257–258.

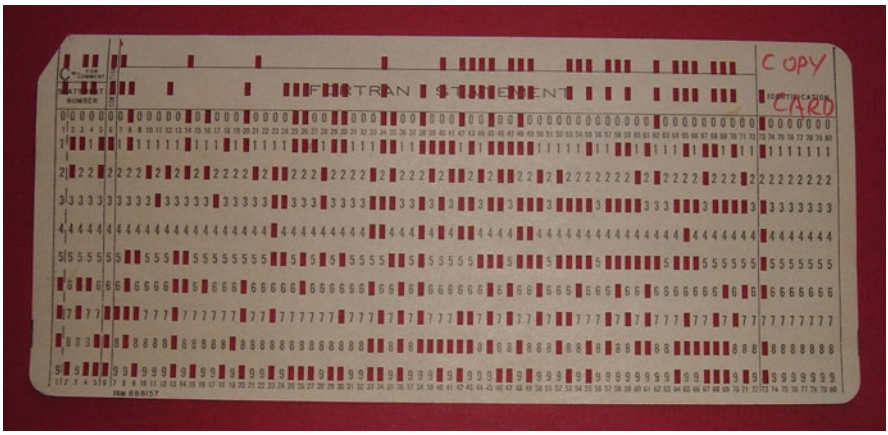

Fig. 7 Punch card in binary format. Source: Arnold Reinhold (free license)

The media frame of this category of iconography, which refers to the special relationship between the HGP and (bio)informatics, is also the frame of "technoscientific 'progress' and the utility derived from it." HGP and (bio)informatics are considered as two scientific revolutions taking place in conjunction and parallel, and this media framing emphasizes the multiple benefits which would emerge from them. Thus, the framing and rhetoric of this category of iconography shape a "positive" public image for the HGP, highlighting the projected benefits for healthcare and medicine.

3.3 HGP and Genetic Determinism

The third category of iconography includes a series of digital depictions, illustrations, and artistic pictures. Fears for the standardization of human beings, the reduction of genetic diversity, and genetic testing/screening, as conceived in the public imagination through public culture (science fiction literature, films, comics, etc.), are reinforced in the minds of readers with the reproduction and publication of several digital depictions and illustrations (Figs. 8 and 9) accompanying and framing the publications regarding HGP. The propensity of journalists to reproduce a genetic determinism today reflects "stereotypical" perceptions concerning the relationship between inheritance and behavior, or the nature versus nurture dispute, such as were taking place in public debates and controversies in the era of classical eugenics.[49]

[49]For the history of classical eugenics and the emergence of new eugenics and how this is linked to the utilization of genome sequencing, see Rifkin, 1998, 116–147 (Chap. 4, A Eugenic Civilization) and 148–174 (Chap. 5, The Sociology of the Gene). Also, about eugenics, see Lewontin 2001, 3–40; 315–340; Moranz 1998, 163–168; Jordan, 2002, 145–157; and Kitcher, 1996, 179–204.

Γενετικές διακρίσεις

Ένα βήμα μπροστά στην επιστήμη, δύο βήματα πίσω στα ατομικά δικαιώματα. Κι έπειτα παραξενευόμαστε για την υποχώρηση του ορθολογισμού.

Η Κιμ ήταν αφελής. Σε μια συνάντηση του προσωπικού της επιχείρησης στην οποία εργαζόταν, ανέφερε ότι η μητέρα της έχει πεθάνει από τη νόσο του Χάντιγκτον και ότι κατόπιν αυτού παρουσιάζει κι εκείνη 50% πιθανότητες να παρουσιάσει την επικίνδυνη αυτή γενετική ασθένεια. Μια εβδομάδα αργότερα πληροφορήθηκε την απόλυσή της.

Η Τέρρι Σάρτζεντ ενδιαφερόταν για την υγεία της περισσότερο απ' όσο έπρεπε. Πήγε στον γιατρό πιστεύοντας ότι για τη δύσπνοιά της έφταιγαν κάποιες αλλεργίες και διαπίστωσε ότι πάσχει από ανεπάρκεια Alpha 1, μια διαταραχή που θεωρείται κληρονομιά των... Βίκινγκς και συχνά αποβαίνει μοιραία. Το νέο μαθεύτηκε στη δουλειά της, οι προϊστάμενοί της τη χαρακτήρισαν «παθητικό» και δεν άργησαν να της δείξουν την πόρτα.

Μια άλλη 40χρονη γυναίκα που δεν έγινε γνωστό το όνομά της ήταν εξαιρετικά συνεπής στη δουλειά της, αλλά έκανε το λάθος να δεχθεί να λάβει μέρος σε μια γενετική έρευνα. Το αποτέλεσμα ήταν να αποδειχθεί θετική στο BRAC1, ένα γονίδιο που συνδέεται με ορισμένους καρκίνους του μαστού και των ωοθηκών, και να χάσει πρώτα την ασφάλισή της κι έπειτα τη δουλειά της.

Έρευνα που διεξήχθη πρόσφατα στη Μασαχουσέτη έδειξε ότι 582 αιτήσεις για δουλειά απορρίφθηκαν επειδή ανακαλύφθηκαν «ελαττώματα» στα γονίδια αυτών που τις υπέβαλαν. Το Συμβούλιο Υπεύθυνης Γενετικής έχει τεκμηριώσει περισσότερες από 200 περιπτώσεις γενετικών διακρίσεων από τους εργοδότες. Οι ερευνητές πιστεύουν ότι οι πραγματικοί αριθμοί είναι πολύ μεγαλύτεροι, αφού ούτε οι εταιρείες είναι πρόθυμες να δίνουν στη δημοσιότητα τέτοια περιστατικά ούτε τα θύματα των διακρίσεων τις καταγγέλλουν, φοβούμενοι ότι η θέση τους θα γίνει ακόμη χειρότερη. Την ώρα λοιπόν που η επιστήμη πανηγυρίζει για την αποκωδικοποίηση του γονιδιακού χάρτη του ανθρώπου, χιλιάδες άνθρωποι χάνουν τη δουλειά τους, την ασφάλισή τους και την οικογενειακή τους γαλήνη επειδή οι εργοδότες τους χρησιμοποιούν με παράνομο τρόπο τα γενετικά στοιχεία που τους αφορούν.

Και επειδή ούτε η επιστήμη μπορεί να κάνει πίσω ούτε η ελεύθερη αγορά επιδέχεται ελέγχους και περιορισμούς, το αποτέλεσμα αυτής της κατάστασης είναι να διστάζουν οι απλοί πολίτες να κάνουν γενετικές εξετάσεις που σε πολλές περιπτώσεις μπορεί να τους σώσουν τη ζωή. Έρευνα του Πανεπιστημίου Τζωρτζτάουν της Ουάσιγκτον έδειξε ότι ο φόβος των διακρίσεων αποτρέπει ένα στα 10 άτομα «υψηλού κινδύνου» από το να κάνουν εξετάσεις για κυστική ίνωση, νόσο του Χάντιγκτον, καρκίνο του εντέρου και άλλες κληρονομικές ανωμαλίες. Το αμερικανικό υπουργείο Εργασίας έχει διαπιστώσει ότι πολλές γυναίκες αποφεύγουν να κάνουν μαστογραφία φοβούμενες ότι τα αποτελέσματα θα γίνουν αντικείμενο αγοραπωλησίας από ιατρικά κέντρα και ασφαλιστικές εταιρείες.

«Σε τελευταία ανάλυση», λέει η Τέρρι Σάρτζεντ, «η ζωή μου είναι πιο σημαντική από μια δουλειά». Και γιατί δηλαδή να πρέπει να διαλέξει;

Του
Μιχάλη Μητσού

Fig. 8 Source: Mitsou Michalis, "Genetikes diakrisis" (Genetic discrimination), Ta Nea, September 21, 2000, 54

For example, an article on possible genetic discrimination is framed by a picture (Fig. 8) which shows a human face with a "genetic stamp/barcode." The message of the picture is evident and refers to the general idea that the human is the captive of his/her genes and is a "standardized product" based on some genetic prescription. Likewise, the digital depiction of the genome in the background refers to a perception according to which the human is configured via a "computer program," the program of his/her "genetic code." In this context, the "linguistic message" of the caption to this photo, "Human genome: its decoding is accompanied by massive

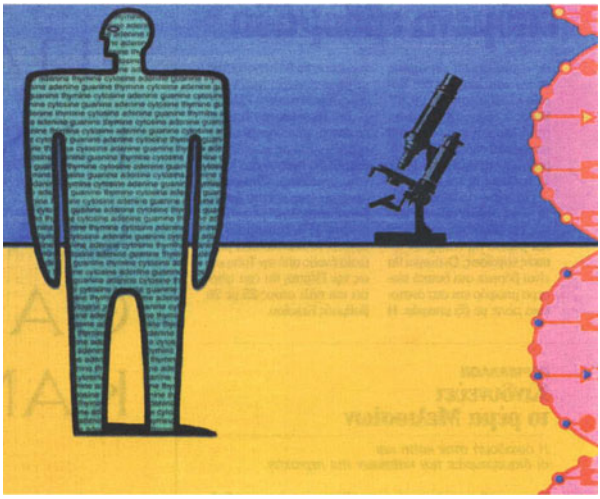

Fig. 9 Source: Coghlan Andy, "O pio thavmastos chartis metamorfoni tin iatriki" (The most wondrous map transforms medicine), To Vima, November 12, 2000, 54–55 (republished by New Scientist)

layoffs" (translated by the author), enhances the fears of readers about dystopian scenarios involving possible genetic discrimination that may afflict a person through the disclosure of information of its genome. This shapes and reproduces a "negative" public image for the human genome sequencing and the practical applications deriving from the HGP. This is an iconography which reminds us of many dystopian images from science fiction films like *Gattaca* (1997) and *In Time* (2011) and comics like *Megalex* (2012). As pointed out by Bates, "regardless of whether sci-fi tends to advocate for or against genetic technology, scholars have asserted that sci-fi guides the public's understanding of genetics and makes some uses of genetic technology acceptable or unacceptable to the public.[50]"

Moreover, another illustration (Fig. 9) reproduces a stereotypical perception according to which "the gene is clearly conceived as a command post installed at the very core of the individual and conditioning broad aspects of his or her behavior[51];" in other words, our fate is in our genes. In this illustration, we see a section of the double helix of DNA, a microscope, and the drawing of a human containing a sequence from the letters of the DNA bases (AATGCCGATGCAATTTAAT). In other words, the human is designed in such a way as it is regarded as nothing else but the total of the "genetic code" which it includes. The rhetoric of genetic determinism finds in this illustration a complete and perfect expression.

This iconography arouses in the minds of readers fears for the revival of eugenics while reproducing a "strong" genetic determinism. It creates a distorted public image of HGP, its scientific significance, and also the perspectives that this

[50]Bates 1995, 49.

[51]Mauron 2002, 958.

technoscientific fact offers to society as a whole. The implications of this event are described characteristically by Leontin: "The study of DNA is an industry with high visibility, a claim on the public purse, the legitimacy of a science, and the appeal that it will alleviate individual and social suffering. So its basic ontological claim, of dominance of the Master Molecule over the body physical and the body politic, becomes part of general consciousness. [. . .] Beyond the building of a determinist ideology, the concentration of knowledge about DNA has direct practical social and political consequences, what Dorothy Nelkin and Laurence Tancredi call 'The Social Power of Biological Information'. [. . .] [T]here is no instance where knowledge of one's genes does not further concentrate the existing relations of power between the individual and institutions.[52]"

Although recognizing the scientific importance of human genome sequencing, the use and reproduction of this iconography is a journalistic practice that aims at sensationalism and dramatization. Consequently, the reproduction of a widespread perception according to which the "human is determined and controlled by its genes" has the effect of generating an "incorrect or distorted" public image about the role of the genome in the configuration of a person. So the prevailing media frame of this iconographic material is the frame of "ethical concerns and risks." With this framing, the category of iconography that reveals a connection between human genome sequencing and genetic determinism contributes to the creation a "non-positive" public image for HGP.

3.4 HGP and Politics

According to Winner, one of the most recognizable and influential scholars in the scientific discipline of Science, Technology, and Society (STS), who in several of his works approaches the science and technology from a sociopolitical perspective, there are at least two levels where the artifacts embody political qualities/choices: the first level is a level at which technologies "could provide a convenient means of establishing patterns of power and authority in a given setting," although "technologies of this kind have a range of flexibility in the dimensions of their material form." The second level is a level at which the technologies "are strongly, perhaps unavoidably, linked to particular institutionalized patterns of power and authority."[53] Starting from the phrase of Winner, that "artifacts have politics," the fourth category of iconography includes a series of photographs which highlight that the human genome sequencing and HGP was a technoscientific fact that took place through the involvement of various political, economic, and scientific factors.

One of the most typical photos (Fig. 10) is from the celebration of the "triumphal" announcement of the completion of the human genome "working draft" (this

[52]Lewontin 2001, 164–165.

[53]Winner 1980, 134. For a critique to Winner, see Joerges, 1999, 411–431.

Fig. 10 Source: Anon., "To mellon ine edo. Ti ipe gia tin anakalipsi o proedros Clinton" (The future is here: what President Clinton said about the discovery), To Vima June 27, 2000, 1

was published on the front page of the newspaper *To Vima*). This portrays the main actors who took part in the achievement of the scientific program of human genome sequencing. Scientists are represented by Francis S. Collins, director of the US National Institutes of Health,[54] politicians by US President Bill Clinton, and the business world by Craig Venter. This photo was republished several times, not only in the period of the announcement of the completion of the human genome "working draft" in 2000 but in subsequent years. The central message and the rhetoric of this photograph is that the human genome sequencing is an accomplishment which is the result of a specific science and technology policy. The huge amount of money that was needed for HGP was covered by state funds in cooperation with research organizations and scientific programs. However, the human genome sequencing was also a matter of the markets. The business funds of Celera and the competition which these caused between public and private sector contributed to the faster completion of this scientific fact.

This category of iconography focuses on a group of (bio)scientists, politicians, and businessman which announced, to a fanfare of publicity, that the human genome was nearly sequenced (26 June 2000). As pointed out by the Nerlich and Hellsten: "The completion of the HGP is here conceptualized as the *summit* of an achievement from which we can *see* the future as *mapped out* on a new *landscape*" (italics in the original).[55] Thus, the media frame of these photographs is that of "technoscientific 'progress' and the utility derived from it." HGP becomes "a positive symbol of the genetic revolution.[56]" This media framing shapes a "positive" public image for HGP, and this "was portrayed as a watershed in history and depicted as promising great medical progress.[57]"

[54]For more information about Francis S. Collins, see https://www.nih.gov/about-nih/who-we-are/nih-director/biographical-sketch-francis-s-collins-md-phd. Accessed 28 May 2016.

[55]Nerlich 2004, 256.

[56]Nelkin 1996.

[57]Henderson 2007, 67.

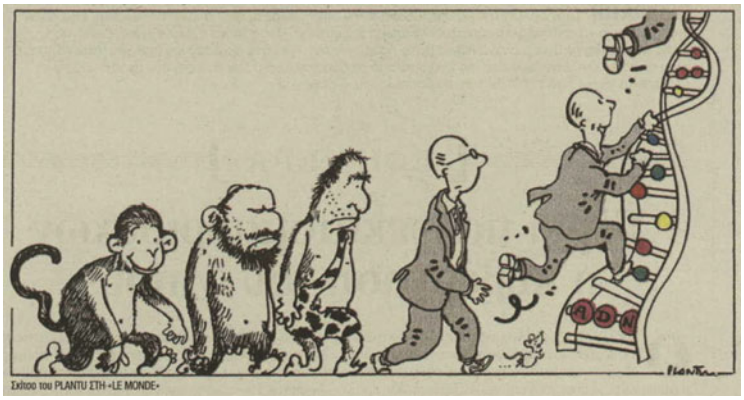

Fig. 11 Source: Soufleri Ioanna, "Farmaka… IX ferni to Vivlio tis Zois" (Private medicine provides the Book of Life), To Vima, February 14, 2001, 14

3.5 HGP and Newspaper Cartoons

The fifth category of iconography comprises a variety of newspaper cartoons on HGP. As highlighted by Giarelli, "Humor is a part of daily life that is considered to be a legitimate area of inquiry and cartoon humor is one channel for the communication of ideas about genetic science, technology, and their consequences.[58]"

The cartoon by Plantu (republished in the newspapers *To Vima* and *Le Monde*) (Fig. 11) reproduces a classic and ordinary public image for human evolution from ape to *Homo sapiens*, who subsequently, through a ("heavenly") scale helix of DNA, rises to the level of God. This iconography for the HGP highlights the "revolutionary" character of this technoscientific achievement and the rhetoric about the human genome sequencing, according to which in our time the human knows the "secret of life" or holds the "book of life" and so is raised to a higher "divine" level of self-awareness. The mouse at the bottom of the scale also symbolizes the importance of the small rodent in the development of biosciences.[59] Following the same rhetoric in the cartoon of Elias Makris (Fig. 12), we see the President of the USA, Bill Clinton, at the base of a double helix of DNA, which rises to the sky like a metaphorical "ladder of Jacob,[60]" to exclaim to the puzzled "dashboard" God "to ascend or descend." The cartoon's message is absolutely clear: Man, through the developments in the field of biosciences, succeeds to reaching or abolishing, in practice, even God.

The second cartoon by Plantu (republished in the newspapers *To Vima* and *Le Monde*) (Fig. 13) compares the scientific fact of the human genome sequencing to another great moment of scientific development: the exploration of space and the

[58]Giarelli 2006, 64.

[59]See Morange 2000; Jacob 1998, 47–64.

[60]*Genesis* 28: 12–13.

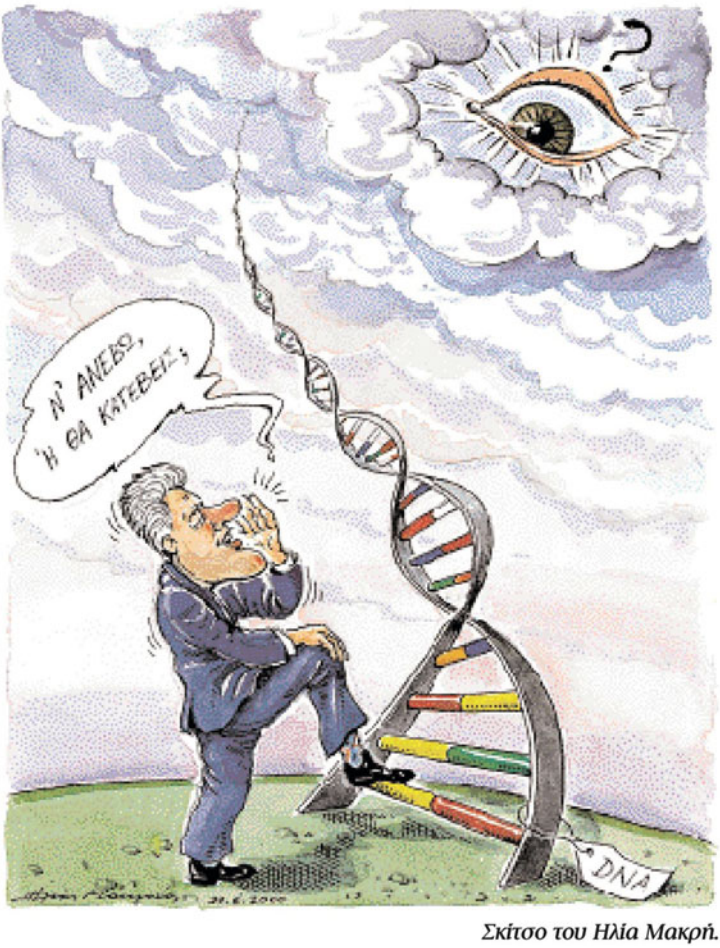

Fig. 12 Anon., DNA: oi elpides, oi anisichies kai oi Hellines (DNA: the hopes, the fears and the Greeks), I Kathimerini July 2, 2000: 1

Moon landing. It refers to the famous phrase by James D. Watson, who, with Francis Crick, won the Nobel Prize for solving the riddle of the structure of DNA: "We used to think our fate was in our stars. Now we know, in large measure, our fate is in our genes.[61]" Specifically, the cartoon depicts an astronaut holding a rope ladder in the shape of the double helix of DNA, which it starts from a spacecraft, while the figure himself is ready to step onto the surface of the Moon. The rhetoric of this cartoon aims at evoking in the mind and memory of readers the famous American astronaut Neil Armstrong, whom the (bio)scientists are likened

[61]Quoted in Jaroff 1989.

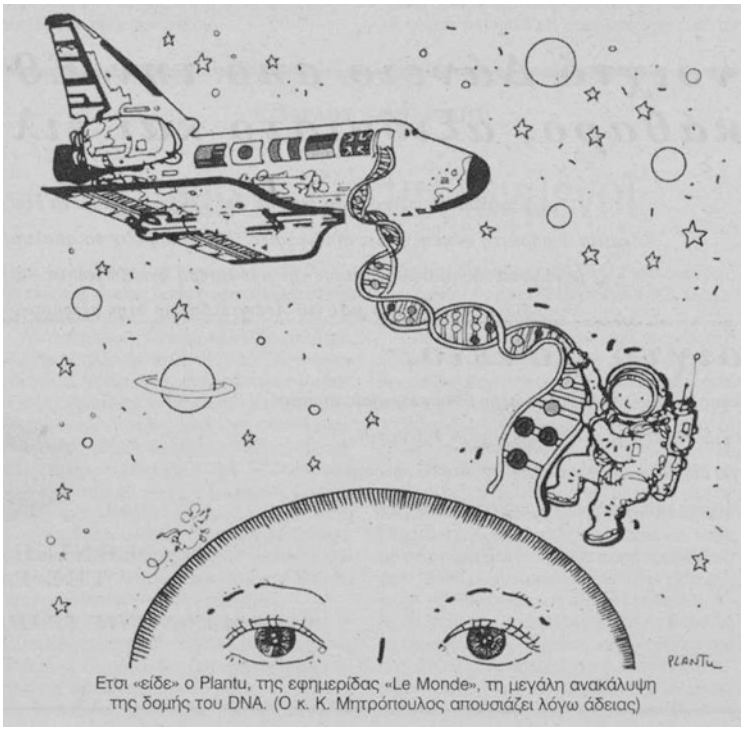

Fig. 13 Source: Editorial cartoon by Plantu (republished by Le Monde), To Vima, July 2, 2000: 70

to. Like him, the modern (bio)scientists participating in the HGP make "one small step for [a] man, one giant leap for mankind." Also, the flags of various countries (the USA, Japan, France, Germany, and the UK) on the spacecraft, which symbolize the international consortium of HGP, remind the readers that the human genome sequencing is an international scientific achievement. Again, the little mouse running on the lunar surface area is a particular comment by the cartoonist on the importance of this small rodent in the development of the biosciences.

As Giarelli notes: "Cartoons have claims to truth, as do other forms of art that attempt to represent and reflect reality and supplement the news presentations with statements of 'meaning'. Cartoons develop a subtle semiotic structure to generate a particular meaning that is humorous. The by-product is to gain support for an argument or point of view. [...] [Cartoons] reflect cultural attitudes and values, and record and perpetuate many commonly held beliefs.[62]" In this way, the cartoons of this category of iconography contribute to establishing a distinctive public image for HGP. The rhetoric and framing of these cartoons highlights the "revolutionary" character of HGP and the importance which it has for the

[62]Giarelli 2006, 64.

understanding of human nature. Thus, the media frame of these cartoons is that of "technoscientific 'progress' and the utility derived from it." Meanwhile, it reproduces the linguistic metaphors and stereotypes according to which human through science and technology plays or is "God."

3.6 HGP and Infographics

The sixth and final category of iconography includes a set of infographics (Figs. 14 and 15). They are graphic visual representations of information, data, or knowledge intended to present information quickly and clearly. The important role of infographics in science communication is highlighted by Mol: "Infographics use symbols, colors, graphics, and other design methods to present information in a way that is visual and easy for our brains to interpret. In addition to the information being easy to interpret, a specific piece of information can be found due to the shapes, symbols, and colors that facilitate the display of information. Compare that to reading an article where your brain has to stay focused on reading the information in a certain order, remembering all that information in order to continue through the rest of the article. Not to mention if you need to reread a specific piece of

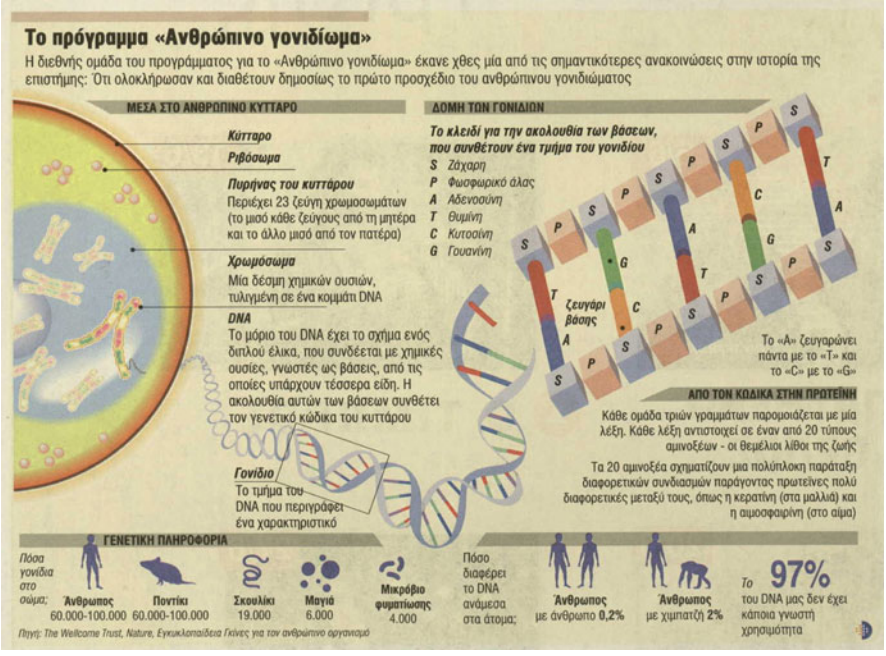

Fig. 14 Source: Vranas Roussos, Evi Eletheriadou, Christos Manolas, "Pos ftasame sto spasimo tou kodika" (How we got to breaking the code), Ta Nea, June 27, 2000: 20–21

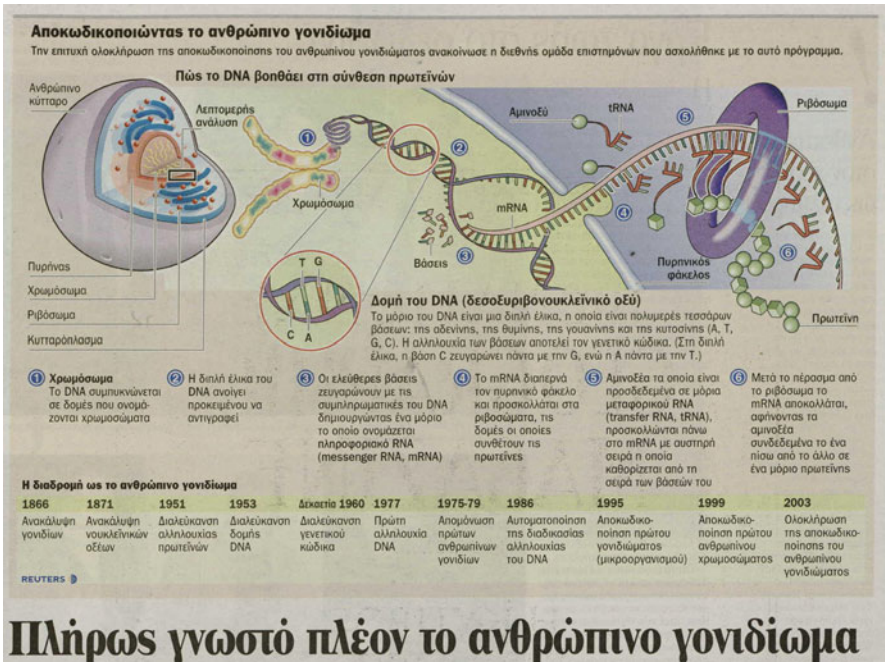

Fig. 15 Source: Soufleri Ioanna, Pliros gnosto pleon to anthropino gonidioma (The human genome is now known completely), To Vima, April 15, 2003: 37

information and you have to find a word or phrase in an elaborate text.[63]" Therefore, infographics are significant communication tools as they concisely provide information, making a set of complex technoscientific concepts more easily understood by the public.

The first infographic (Fig. 13), entitled "The 'Human genome' project" (translated by the author), provides information on the human cell structure, the structure of genes, the protein production, and a summary of information about the human and other organisms genome as well as the differences between them. This infographic reminds the readers the knowledges that they have learned at school on the organization structure and gives some additional information about the genome. This is not mentioned in HGP itself but in the subject of the program, the genome contained in each human cell. The second infographic (Fig. 14) entitled "Decoding the human genome" (translated by the author) provides information on the cell structure, the structure of DNA, and protein production. It also presents a timeline with the main milestones of the route by discovery of genes in 1866 to the completion of the human genome sequencing by the HGP in 2003. This infographic is a perfect example of storytelling. It is an "educative" graph that informs the

[63]Mol 2011, 47.

readers thoroughly about complex issue such as the human genome sequencing and HGP. Both have the Reuters news agency as their source and are republished in newspapers *Ta Nea* and *To Vima*, framing the news reports for HGP and providing more information on the subject of the program and the process of genome sequencing.

These infographics play an important role in how they shape a public image of HGP, visualizing in a simple way what the significance of the HGP is. They are attractive because of the illustrations and colors. Moreover, these infographics are capable of making the complex process of human genome sequencing more understandable by the readers of newspapers. According to Mol, "it has been proven that humans simply learn better and understand more, if the [infographics] are well designed and are aesthetically pleasing to the eye.[64]" This category of iconography with the infographics has the capacity to overcome certain educational and cognitive barriers and can highlight in the best way the promises of the HGP, communicating them to the public. Especially, the media frame of these infographics is that of "technoscientific 'progress' and the utility derived from it." So, this media framing has, as a result, the shaping a "positive" public image for the HGP, emphasizing the new horizons opened up for scientific research, gene therapy, and medicine.

4 Conclusion

Increasingly today, the science communication in mass media relies on iconography to clarify data, illustrate concepts, and engage with a public informed through an ever-increasing arsenal of photographs, pictures, digital depictions, infographics, illustrations, cartoons, and new media tools. Examples about the use of iconography in mass media are abundant, but relatively little attention has been directed to how it has been exploited by the science journalists in science communication and the role it has in the public engagement with science and technology. This article offers a context in which the significant role of mass media iconography in science communication might be examined.

As already presented above, the iconography for the HGP in most popular Greek newspapers helps shape a particular public image about it. The portraits of (bio) scientists who participate in human genome sequencing, and the explanatory infographics, produce a "positive" media framing for HGP. Moreover, the iconography that highlights the interaction and joint development of HGP and (bio) informatics, the close relationship of HGP with politics, and several cartoons emphasizes the "revolutionary" character of a technoscientific fact as the human genome sequencing achieved through HGP. On the other hand, there are also some digital depictions, illustrations, and artistic pictures which highlight the genetic

[64]Mol 2011, 46.

determinism that the newspapers often reproduce when referring to aspects of the human genome sequencing and the political, social, economic, and ethical impact of HGP in our modern world. In conclusion, the HGP and the human genome sequencing attracted intense coverage from most of the popular Greek newspapers. The newspaper titles, the rhetoric, and the framing of publications for HGP compared it to the greatest moments of social, artistic, and scientific developments. The analogies include the Moon landing; the discovery of "New World"; the revolutionary ideas of Copernicus, Newton, Darwin, and Einstein; the artistic creations of Leonardo da Vinci, Shakespeare, Mozart; etc. In this context, the iconography was chosen by the most popular Greek newspapers to strengthen the "positive" media framing for HGP and shape a general "positive" public image for this.

In conclusion, this paper suggests that the study of the mass media iconography for biosciences and biotechnology is a challenge for those interested in the effective communication of bioscientific developments, and this study should be a multidisciplinary enterprise. Researchers from disciplines like SciCom and STS contribute to the effectiveness of such efforts by turning a critical eye toward the functions, purposes, and effects of iconography in the science communication.

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Part VI
Narrated History

National Human Genome Research Institute History of Genomics Oral History Program: An Example of "Triangulation"

Christopher Donohue

Abstract The essay will describe the oral history effort at the *National Human Genome Research Institute* (NHGRI). Overseen since 2013 by the NHGRI Historian, the focus of the oral history effort is to capture the perceptive of scientific program staff, many of whom have worked at the National Institutes of Health since the origin of the Human Genome Project. Scientific program staffs have functioned as "managers" of the Human Genome Project and of subsequent genomics efforts. Program staff design granting programs and ensure that grantees and institutions meet their scientific targets and spend money in a cost-effective manner. They are an untapped source of knowledge behind the development of modern genomic science. Interviewing the program managers behind the Human Genome Project and subsequent genomics programs mitigates many of the issues surrounding traditional oral histories. Individual scientists often promote their own work at the expense of others. Individual scientists also are increasingly specialized; they can typically only discuss certain aspects of a large, international, and interdisciplinary science such as genomics. Program staffs are able more than individual scientists to discuss the whole of a scientific project. They are also, from their managerial perspective, far less likely to denigrate the work of one individual scientist for the promotion of another.

The last section of the essay will address the related difficulties with oral histories, as historians traditionally see them as social constructed and untrue, as the products of sheer careerism. However, because the NHGRI also possess an extensive institutional archive, oral history questions are checked and developed with ready access to archival sources. Program staff are interviewed multiple times

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in order to reduce inconsistencies and to make oral histories the sites of corroboration. The oral history effort puts multiple sources (the archive and the oral history) in a dialogue or in a "triangulation" as a recent historian of science has termed it. This reduces the issues inherent in oral histories. The essay ends with arguing that treating archival sources and oral history sources in a hierarchical manner is incorrect. Both are equally needed and both lead us closer to the truth.

Keywords Oral history • Triangulation • Archives • Scientific program staff

1 Introduction

The History of Genomics oral history effort at the National Human Genome Research Institute (NHGRI) begun in 2013 under the supervision of Eric Green, NHGRI Director, and under the direction of the NHGRI Historian, Christopher Donohue. The NHGRI, through the National Institutes of Health, remains the largest funder of genomic science in the world. The NHGRI has also been one of the principal intellectual forces guiding the programmatic development of the Human Genome Project and of subsequent genomics programs.

The NHGRI History of Genomics Program has had as its purpose, since its inception in 2012, to capture the history of the Human Genome Project (begun in 1990 and concluded in 2013) and to continue to catalogue, to describe, and to make available the historical materials generated by ongoing genomics programs until the present day. At the Institute, genomics typically refers to work derived from the completed sequence of the human genome or genomics efforts undertaken after the completion of the full sequence in 2003.[1, 2]

The files held in our institutional archive now number about 2.4 million. Such files have been essential to database development for scholarly access and most importantly for the corroboration of oral history accounts, as noted below.

Although much of the file content covers the NHGRI's role in coordinating and funding sequencing efforts (especially the efforts necessary for the draft sequencing), completed in 2001,[3] there are significant file holdings on the funding and planning of early (1991–1995) mapping and sequencing efforts,[4] the funding and planning of genome sequencing technology development, institutional collaboration efforts (with the United States Department of Energy as well as with various biotech companies, including Celera Genomics), the funding and planning of ethical and legal studies of the implications of genomics research, as well as the funding and planning of human variation and functional, comparative, and

[1] Collins et al. 1998.

[2] NHGRI 2016: A Brief Guide to Genomics.

[3] NHGRI 2016: 2001 Release: First Analysis of Human Genome.

[4] NHGRI 2016: Online Education Kit: 1995—Physical Map of Human Genome Completed.

regulatory studies[5,6] undertaken after the finishing of the human sequence.[7] The archives also include a significant amount of grantee correspondence after the funding of individual grants and projects. The archives themselves contain no grant-related material, and all materials relate to scientific programs after they have been funded.

Since 2014, the efforts of the NHGRI History of Genomics Program has broadened to include a visiting scholar lecture series, a database development cycle to enable outside researchers to access our files, as well as the promotion of scholarly work. At present, a special issue of the *Journal of the History of Biology* on the history of the Human Genome Project and of subsequent genomics programs is near completion.

The oral history effort uses an in-house production studio, which enables the oral histories to be both video and audio recorded. The oral histories are then transcribed by a professional contracting company. The transcripts are then corrected by History Program staff, including the Historian of the NHGRI. Both the edited video recordings and the edited transcripts will be posted on the NHGRI YouTube, GenomeTV. The oral history effort has to date (July 2016) completed over 30 oral histories. Plans are to continue to grow the oral history program, producing about one oral history a month (sometimes two) for a total of 20 per year.

2 The Purpose of the Oral History Effort at the NHGRI

The focus of the oral history effort at the NHGRI has remained capturing the stories of scientific program officers. These program officers are responsible for the bureaucratic management, guidance and development of the Human Genome Project and subsequent genomics efforts. Accordingly, many of the oral histories are not those of scientists, but rather of scientific program staff. Program staff at the NHGRI propose ways to support the growth and development of genomic science. They actively shape the future of genomic science with grantees as their managers and coaches. For example, the genomic variation programs at the NHGRI are designed to "support research aimed at discovering and typing single nucleotide polymorphisms (SNPs), indels, and other forms of genetic variation on a large scale across the genome.[8]" After grantees and research institutions are given funds for specific scientific projects, scientific program staff are then tasked with making sure scientists and their research institutions meet their scientific benchmarks and that the funds are spent in a responsible and efficient manner. Because of the emphasis on scientific program staff, the oral history effort at the NHGRI gives more of an

[5]ENCODE 2004.

[6]Thomas 2003.

[7]Gibbs 2003.

[8]NHGRI 2016. Genetic Variation Program.

"insider" view of the development, progression, and management of complex, big (or better, "collaborative") science efforts than those provided by grantees.

As importantly, focusing on the "managers," rather than the scientists themselves, works against the image of the heroic, male scientist. Men and women are about equally represented in scientific program management at the NHGRI.

As importantly [repeated], genomics, being a transdisciplinary and multinational effort, is better represented from the managerial viewpoint than from the viewpoint of an individual investigator. The International HapMap Project, begun in 2005, to develop a resource for studying the connections between human genetic variation and its connection to disease, for example, was guided and administered by the NHGRI, using samples from four populations and marshalling the efforts and resources in Japan, Canada, China, the United Kingdom, and the United States. If only individual investigators were interviewed, those individuals would tend to focus on their efforts to the exclusion of efforts of other groups and other investigators.

Furthermore, most contemporary biologists are highly specialized. Genomic science is by its very nature interdisciplinary. For example, the International HapMap Project involved not only sequencing but also genotyping, data coordination and deposition, data mining, and bioinformatics techniques, as well as the development of specific intellectual property guidelines. While individual scientists would have an intricate knowledge of one method, one approach (e.g., sequencing or genotyping, bioinformatics), or one phase of a scientific project such as the HapMap, program managers would have a sense of the program as a whole. Thus, it is perhaps the holistic view of the program which is among the most useful perspectives on contemporary science.

As the oral history effort advances into 2017 and 2018, more efforts will be made to integrate the perspectives of grantee scientists into the narrative. As importantly, many of these scientists will be asked to corroborate or challenge the picture of the science presented by interviewed program managers. This effort at corroboration (or better at "triangulation") will attempt to provide case studies of how oral histories can be a significant resource for contemporary history of biology.

This leads to the last section, which will argue for how the oral history effort at the NHGRI will attempt to address many of the issues surrounding the veracity and reliability of oral histories. It will do this in part by interviewing multiple subjects on the same topic. However, the greatest resource to resolve the difficulties with oral histories is the archive itself. Miguel Gabriel Sancho has argued, quite elegantly, that one of the solutions to problematic sources is one of "triangulation," which seeks to integrate the oral history accounts with the archival record.[9]

[9]García-Sancho 2016.

3 The Difficulties with Oral Histories

Oral histories, particularly of contemporary subjects, are intensely controversial. They are seen as constructions of their subjects and in a scientific context, the products of career promotion. Scientists when interviewed, tend to promote their work, and not the work of others. Scientists, like people generally, tend to also engage in slanderous gossip. Historians have proposed as Sancho points out that oral histories are sometimes used in the absence of archives or in order for the historian to gain access to archival sources. Oral histories are used as bribes: if you do an oral history of a scientist, he will lead you to his papers.[10, 11, 12, 13] Oral histories then do not have a good reputation in the historical community.

However, at the NHGRI, due to the availability of the archive, oral history questions are not only constructed with archival sources available, but oral histories of program officers are continually "fact-checked." This is done over successive interviews and in follow-up discussions. In this way, "triangulation" of archival sources and oral histories occurs. With the development of database resources at the NHGRI, the search of these archival resources can be ever more efficient. Thus, if a misstatement is identified, the archive is then checked (or in many instances rechecked). In a subsequent interview, the misstatement is discussed with the participant. It has been the experience of the Historian of the NHGRI that interview participants are deferential to the record (particularly when that record is a letter or note that they themselves wrote.)

Thus, it is also the supposition of the Historian that errors in the oral history made by participants are typically not malicious, having more to do with the uncertainty which comes with the passage of time. An account of the truth is possible through the triangulation of oral histories and archives. The best historical answer is not through one source, nor through an account which privileges one source over another, but that which treats sources in dialogue.

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[10]Thompson 2000, 16.

[11]Tonkin 1995, 13.

[12]Perks 2015, 19.

[13]Thomson 2007.

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Narrating Genes: How Patients with Chronic Inflammatory Bowel Diseases Interpret an Emerging Disease Aetiology and How We Can Make Sense Out of It by Developing a Historically and Sociologically Informed Framework

Dana Mahr

Abstract When it comes to genes, we consider them almost automatically as something that belongs exclusively to the spheres of science and biomedicine. We understand them as scientific concepts or treat them as epistemic objects—due to this, we describe them with an esoteric language using the vocabulary of "codes", "traits", "dispositions" or "susceptibility". This chapter seeks to broaden our view as well as our vocabulary by uncovering a "lived perspective" of genes and genomics. For this I propose to analyse the narratives and experiences of those who are actually confronted with their genomes, patients and families who lead their lives in the light of geneticized diseases. For this I use (on an exemplary basis) a material from semi-structured interviews I undertook in the course of a project titled "the lived genome and chronic inflammatory bowel diseases". The bioethicist Christoph Rehmann-Sutter and I conducted this project during the years 2013 and 2016 at the University of Luebeck (Germany).

Keywords Individualization of genetic knowledge • Social aspects of genetics • Sense-making • Chronic inflammatory bowel diseases • Oral history • History of genetics • Sociology of medical knowledge

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1 Introduction

Biomedicine is a dynamic actor in determining and defining diseases. Changes in the conceptualization of diseases have wide-ranging consequences, going far beyond the confines of clinical practice. In order to conceptualize and to investigate such dynamics, I suggest a multiperspectival approach, combining historical, phenomenological, sociological and ethical perspectives, for coming to terms with these multiple effects of recent and ongoing research in the life sciences. In this chapter, I suggest the analysis of chronic inflammatory bowel disease as an exemplary case.

In line with the well-known ascendancy of genetic molecular biology, inflammatory conditions such as ulcerative colitis and Crohn's disease have recently been characterized as clinical conditions with a genetically determined susceptibility, adding genetic factors to the explanation of a chronic autoimmune disease of yet not fully understood origin. This shift followed the contemporary predominance of molecular and genetic approaches in the life sciences and resulted in a reconfiguration of conditions that had changed their identity already several times during the twentieth century—and still does in the twenty-first century. Like the preceding transformations, also the genetic approach to chronic inflammatory bowel diseases highlights particular aspects while obscuring others. In particular, it changes the experts' understanding of the condition together with the lifeworld of those persons afflicted in relation to the alleged process of scientific progress. Analysing these changes offers unique opportunities for investigating the concepts, conflicts, negotiations, tensions and compromises underlying clinical research practices. With regard to chronic inflammatory bowel disease, at least the following trends and transformations can be distinguished and should be integrated: the search for a pathogenic agent, the psychosomatic interpretation of biographical patterns, interactions between organism and environment, risk factors, inherited disease traits and predispositions. With inflammatory bowel diseases being chronic conditions, these divergent approaches were always embedded not only in scientific discourses and clinical therapeutics but also in the lifeworlds of the patients and their families. Beyond reconstruction of scientific conceptualizations, the trajectories of explanations in the different knowledge spheres have to be analysed. Each of these conceptualizations encapsulates different meanings that link with specific, highly divergent approaches—like regulatory healthcare regimes, biopolitics or individual healthcare decisions and life choices. To get a more in-depth understanding of these, I suggest building a framework for the empirical investigation of the co-production of disease concepts and the socio-epistemic implications of medical explanations in conjunction with their embedment into larger biopolitical trends.

As a step towards this direction, I will outline in this chapter some cornerstones for an entangled account for studying the complex shifts within the disease aetiology of inflammatory bowel diseases by integrating historical considerations, biographical experiences and narrated disease concepts of patients and their

families. Through this I aim to show in which ways the dynamics of disease concepts are laden with socio-epistemic challenges.

After a short glimpse on the state of the medical research and science and technology studies' approaches towards inflammatory bowel diseases and their embodiment and governance (1), I will give a short overview over the history of the psychosomatization and geneticization of chronic inflammatory bowel diseases and the implicit potential of this historically consecutive labels to create different types of patient's lifeworlds and concepts of agency (2). Following this I exemplary analyse the lived experience of patients who experienced both historically entangled explanatory models. I will ask how patients and their families made sense out of these concepts and how these interpretations actually influenced their lifeworlds and life decisions (3). In the last part, I finally outline a theoretical framework that may help to explore and understand the complex knowledge production in this kind of socio-epistemic processes reflecting on biomedical knowledge in more detail: the concept of a "lived genome". This framework was first developed and tested by the bioethicist Christoph Rehmann-Sutter (Luebeck, Germany) and myself (Geneva, Switzerland) between the years 2013 and 2016 as part of a broader approach reflecting on patients' perspectives towards disease conceptualizations (4).[1]

2 Chronic Inflammatory Bowel Diseases in Discourse

Current opinion regarding the aetiology of inflammatory bowel diseases states that in a genetically susceptible host, an environmental trigger (e.g. infection, medication, smoking, alcohol) may be the inciting event, resulting in an exaggerated immune response to the resident gut bacteria.[2] A growing number of genetic risk loci have been identified recently.[3] Genome-wide association studies have led to a radical change in the predominant views on risk factors for the emergence of an inflammatory bowel disease in an individual person, shifting from an earlier psychosomatic paradigm[4] to a genetic susceptibility paradigm.[5] Up to circa 50 % of the risk is currently assigned to abnormal genes.[6] Current whole genome sequencing studies raise new issues about the ethics of fair patient and family-informed consent. Disease experiences and needs of patients have been studied, either quantitatively[7] or qualitatively,[8] but only little knowledge exists about

[1] See Rehmann-Sutter 2016.

[2] Szigethy 2011; Vinh 2013.

[3] Anderson 2011; Ellinghaus 2012.

[4] Enck 1996.

[5] Tsianos 2012.

[6] Franke 2010.

[7] Magro et al. 2009.

[8] Casati 2000; Anon. 2007; Fletcher 2006, 2008a and b; McCormick 2012; Norton 2012.

patients' and families' perspectives on the impact of genetic explanations of Crohn's disease and ulcerative colitis.[9] This also applies to the implications of genetic risks and the emerging testability of risk loci.

Furthermore, the history of inflammatory bowel disease especially from the perspective of the lived experience of patients and their families (and their transformation in the light of shifting disease paradigms) remains unclear. The state of the international discussion about the ethics of informed consent in exome and whole genome sequencing is mainly focused on privacy issues in data sharing[10] and the return of non-anticipated results to study participants,[11] while the patient's actual experience with genetic explanations is not yet considered as a crucial aspect of information and consent procedures.

There are some recent studies about participants' perspectives and expectations in clinical whole genomic sequencing,[12] but only a few studies on the concrete circumstances of disease phenomenology and the particular interpretations of the significance of "the whole genome" by patients affected by a distinct genetic or multifactorial condition such as Crohn's disease and ulcerative colitis.[13] But an integrative, sociologically and historically well-informed investigation of the sense-making of genomic knowledge of patients and their families is still a desideratum.

3 Psyche, Gene and Lived Experience

Patients contemporary living with chronic inflammatory bowel diseases have experienced (and lived within the framework) of at least two explanatory paradigms of their conditions—both with specific influences on their lifeworlds, coping strategies and ways for developing individual agency: psychosomatics as individual prerequisite and the genetic risk susceptibility paradigm.

Psychosomatics and genetics as explanatory models have been compared conceptually but yet not empirically, by taking lived experiences into account. The sociologist of health Monica Greco has, for example, distinguished two epistemologies of health.[14] For her a "psychosomatic" conception of disease differs from a "biomedical" (e.g. genetic) conception especially with respect to the social roles and the individual obligations they allocate.[15] In Greco's framework,

[9]Lippman 1991; Klitzman 2012.

[10]Tabor 2011.

[11]Caulfield 2008; Tabor 2012; see more critically Christenhusz 2012 and Rehmann-Sutter 2013.

[12]Oliver 2011; Townsend 2012; Tabor 2012.

[13]Mahr 2015; Rehmann-Sutter 2016; Wilhelm 2015.

[14]Greco 1993.

[15]Greco 1993, 357.

psychosomatic disease conceptions are linked to the moral duty of self-care, while a biomedical framing centres the state of being "a patient" in the sense of Parsons' as social patient role: By this being the state of illness is systemically justified and morally excused. By contrast, the psychosomatic model includes the seed of being seen as guilty accountable for being ill.

Greco's reflexive view is an exception since most scholars and practitioners who reflect on psychosomatic disease concepts did this more or less uncritically within the framework of Freudian psychoanalysis—especially when it comes to inflammatory bowel diseases (IBD).[16]

The study of the empirical impact of genetic and genomic knowledge on the level of individual lifeworlds is, however, relatively new. Sequencing techniques were back in the 1970s, 1980s and early 1990s at the cutting edge of science and not widely accessible. The most that has been published in this period was written from either a critical sociological or critical philosophical point of view but always speculative to a high extent.[17] The reflexive stance towards genetics and genomics and the view on individuals living a genome were joined with the possibility of real empirical findings not until the early 2000s. An important first work study that merged combined qualitative empirical narrative methodology with thinking about the impact of genomic knowledge in the contemporary lives of those concerned was conducted by Monica Konrad in her book *Narrating the New Predictive Genetics: Ethics, Ethnography and Science*.[18] It was followed up in the 2010s, by a new wave of publications dealing with questions of predictive and diagnostic genetic knowledge in social spheres, potential changes in self-image and body scheme.[19] This trend correlates with a rapid progress of the possibility of genetic testing and the rise of direct-to-consumer testing companies. Given this more and more people are taking part in "cultures" of genetic knowledge—either actively or passively.[20] On many levels, they are immersed and involved in communication about genetics. People in many countries get encouraged to make decisions about predictive or diagnostic genetic tests when starting a family: before,[21] during and after pregnancy and before, during and after illness. The omnipresence of available genetic knowledge and new sequencing techniques like whole genome or exome sequencing has changed the cultural frames for disease, health and responsibility. Furthermore, new private and public duties emerge: a possible duty of the individual to know his or her own genes and a possible duty of healthcare professionals to tell people about their genetic risks.[22] Hence, medicalization is followed by geneticization. Further questions concern explicitly the "meanings of genomic knowledge" for those who

[16]Latimer 1978; North 1994; Greene 1994; Gerson 2002.

[17]See, e.g. Nelkin 2004.

[18]Konrad 2005.

[19]See, e.g. Klitzman 2012; Zur Nieden 2013; Mahr 2015; Rehmann-Sutter 2016.

[20]Knorr-Cetina 1999, 2007 and 2013.

[21]Hens 2013.

[22]Lunshof 2014; Green 2013.

have to deal with it and integrate it, transform it and translate it into their everyday lives. We need a better understanding, as Barbara Prainsack and others have put it, of how "(...) whole-genome information is used by, and what it means to, a wide range of users... An understanding of what a broader range of users hope to learn from this type of whole-genome information, and whether it would lead to actual life and behaviour changes, would help in assessing whether personal genomics services are likely to be adopted in large numbers".[23]

This knowledge about the users' (and non-users') hopes, fears and subjective understandings with regard to genetic knowledge must be based on an adequate kind of evidence. Such evidence would be needed for planning the good governance of genomics.[24] Questions such as "What does my genetic make-up mean for myself and for my family? Or "In what sense 'am I my genes'?"[25] should therefore be occasions not only for theoretical speculation but also for empirical research, applying qualitative, hermeneutic, phenomenological and comparative methodologies. For this we need to study the ongoing "reflexive embodiment" of genomics[26] and relate it to other medico-scientific explanations and knowledge objects as well as concrete conditions.

4 The Lived Experience of an "Inflammatory" Genome

In the research project "the lived genome and chronic inflammatory bowel diseases" (funded from 2013 to 2016 by the German Research Foundation), the bioethicist Christoph Rehmann-Sutter and I explored the valuation, transformation and individual histories of genomic knowledge by individuals and families living with chronic inflammatory bowel diseases.[27] Hereby we discovered complex assemblages[28] of medico-scientific knowledge and lifeworld experiences.[29] The analysis of these assemblages could be used as a tool for doing two things: to explore a rich contemporary history of the co-production of biomedical knowledge in a case (or field), where we have directly concerned eyewitnesses of the ongoing shift between two completely different disease paradigms (psychosomatics/genetic risk susceptibility), and to operationalize this knowledge for the enhancement of participatory decision-making processes and informed consent procedures in genome studies between potential research participants, patients, their families, scientists and medical professionals.

[23] Prainsack 2008.
[24] Mahr 2015.
[25] Klitzman 2012.
[26] Crossley 2006.
[27] Rehmann-Sutter 2016.
[28] Rabinow 1989.
[29] See Charmaz 1990; Conrad 1990; Gebhardt 1990.

Many of the over 40 individuals and families we interviewed in the course of our research grew up with the explanation that their condition is not only triggered by but also founded in psychosomatics. The rising genetic explanation of their condition is relatively new to them. Yet most of them integrate it (despite its complexity) in highly individual ways into their *lives* and their *narratives or histories of illness*: Both are areas in which they are experts on their own. In this process something is created, which I call a "biographical genome". This biographical genome combines and integrates individual hopes, wishes, fears, experiences, and expectations, pictures about family life, about family history and about family future and discusses them against the background of genomics and other explanatory models. For example, the interview partners Mechthild (suffering from a severe case of Crohn's disease) and Heinz (a married couple in their late 1940s—both socialized within the psychosomatic paradigm) reconsidered their decision for a second biological child after they learned about the genomics of Crohn's disease and the herewith associated risks. Mechthild told us the following:

> Actually it was more [complicated] with the second child—because we said to ourselves, "we have already one, must we expect this again", yes, in terms of this disease [and the new knowledge we obtained—DM], yes. The first time it went well, maybe the second time it will be not so good. At this point, we have made our minds: That is why we choose to adopt our second child. (Interview with Mechthild; Timestamp: 00:08:27)

The everyday world is seen, interpreted and valuated through the eyes of the genome, and the genome is seen, interpreted and valuated through the eyes of the everyday world—as here, for example, the decision of a middle-aged couple to have a second child.

Through this dialectics, concerned persons give meaning and individual significance to the genetic explanation—something that science and medicine (as well as historians of science or bioethicists) cannot do for them. For Kerstin, a 43-year-old Crohn's patient, the genetic explanation of chronic inflammatory bowel diseases gives meaning to her history of being ill. Long she was told (and she also believed) that psychological factors trigger the disease. But her self-observation always seemed to contradict it: Despite of her sheltered life, her Crohn's grew worse and worse. And also other family members (from her cousin's side) had bowel diseases too. Kerstin told us, for example, the following:

> They always say, yes, uhm... the bowel [...] it's psychosomatic. But I had a very nice childhood experience. I could not say that something was [wrong] in my childhood, that something influenced me or gave me any trouble. I cannot remember anything [like that]... Nothing dramatic, a normal life...really nothing...that one could say ok...it's the mind that concerns you. But I have to say, that my father and my sister often had abdominal pain and diarrhea. But my father [...] had a colostomy...there's nothing. But there is really also the side [of the family] [to which] the second cousin and aunt belongs. So I think maybe it has anyway to do with my genes... that the genes bring these bowel-problems. (Interview with Kerstin; Timestamp: 00:40:30)

To have insights into these entangled narratives of patients and their families sharpens the eye for the permanent crossing between the boundaries of the epistemic and social spheres in biomedicine. The examples of Mechthild, Heinz and

Kerstin showed that changes in scientific explanations can have a direct impact on the lifeworlds of patients and their families. They affect whole concepts of life, self-images and social relationships—and this at various levels. For Mechthild and Heinz, the emergence of the genetic susceptibility paradigm leads to stress and reconsiderations with regard to their family planning. For Kerstin the impact affects the core of her identity—because she makes sense out of the genetics of inflammatory bowel diseases in terms of normalizing her individual experience and family history. At the same time, she questions the alternative explanatory model with which she has lived for 40 years.

What can we do with this besides a complex and thick historical or sociological description? For example, recognize the value of these sense-making strategies and integrate them into the elaboration of consent procedures. But it may also affect the core of genetic knowledge production—since exploring the "lived" side of the genome is just as important as the epistemic side. According to the so-called four-dimensional medicine (personalized, predictive, preventive and participatory), it is an imperative to bring both interpretations into dialogue. This could enhance the biomedical practice of research itself, for example, in the context of the recruitment of participants in genome studies. Having knowledge about the "lived side" of a genome may lead to a more participatory or deliberatively structured approach.

5 A Theoretical Framework for the Exploration of "the Lived Genome"

To achieve this, we need a theoretical framework that can integrate processes of the individual translation and management of genetic information and its integration into personal lives—as the example of Mechthild, Heinz and Kerstin has demonstrated. Explaining the genome as something that is both investigated and used in biomedical contexts but also "lived" individually, in families and in societies, could contribute to such a framework.

The reflexively embodied genome is thus charged with basically two sets of meanings that both differ and interact. One is the set of meanings that are attached to the genetic aspects in biomedical research and in clinical contexts. For the sake of simplicity, the bioethicist Christoph Rehmann-Sutter and I call this perspective, and what is seen in it, "genome 1". The genome, however, is translated and transformed into a related but dramatically different figuration that we call "genome 2", which is the genome seen within the lifeworlds of concerned individuals. Scientists who are socialized into the frames of genome 1 may think that their genome is the only true one, while the people's view on the genome is just a subjective translation. They may find many elements of genome 2 (in lay people's understandings) imprecise, off-topic or even incorrect. Their view on "reality" is the world of mathematical models, of physics and chemistry and of the complex charts of cellular systems with

which they work. However, some users of genomic knowledge (other than scientists and healthcare professionals, who may themselves be personal users of genomic information) may equally well find genome 1 too abstract and lacking clear sense for the practical decisions they need to make.

We do not think that either genome 1 or genome 2 is necessarily simpler but rather that they are related to different complexities. Similarly we do not think that either genome 1 or genome 2 is wrong or biased but that they have different truth criteria. Furthermore, genome 1 is not only the raw material for a simplification or application into genome 2. Both are valuable, and their interrelation is interesting to study. Both are concrete for people, and both are in some way necessary; however, they carry different phenomenological features of concreteness. The process of reflexive embodiment can hence be seen as an activity of mutual translations between different meaning contexts.

A linear deficit model of the popularization of scientific knowledge from medical experts to patients, which has been assumed for decades, has become largely obsolete within science and technology studies.[30] It is certainly not helpful for elucidating the process of reflexive embodiment of genetic knowledge. Both sides have advantages and deficits, and both sides need to tell each other what they know and how they know it, how their knowledge produces evidence and so forth. A deficit view does not allow the user perspective to be taken as seriously as the provider perspective, since users are considered to be at the receiving end of the communication cascades. A model, which assumes active contributions from both sides, seems to be more valid. The terminological symmetry between genome 1 and genome 2 should signify this. Sense-making in the field of genetics and genomics is a joint enterprise between producers and users of genetic knowledge and between science and society. The meanings on the two sides, however intertwined, differ considerably—and sometimes they clash.

While genome 1 is actually studied condition by condition—this is the aim of all big research programmes in current systems medicine—genome 2 knowledge is not yet gathered systematically. A similarly progressive condition-by-condition analysis of genome 2 would be needed.

I suggest using and developing genome 1, the biomedical genome, and genome 2, the lived genome, analytically, as perspectives of understanding. They represent two different but interrelated interpretative contexts of the genome and at the same time two different levels of interpretation. Genome 2 is seen in lifeworld contexts by those who know the biopsychosocial implications of genetic susceptibilities and diseases at first hand, that is, by people actually living a condition, having had or not having had a test, being directly involved as a patient or indirectly by being a member of an affected family. Also healthcare professionals are (at least in part) concerned with genome 2. In their professional work, which combines the biomedical and the patient-centred views, they are crossing the interface between the two perspectives. Genome 1 contains all the testable genetic variations, SNPs,

[30]Sinatra 2014; Nerlich 2009.

sequences and genomic data, together with the corresponding medical interpretation given by doctors, scientists and genetic counsellors. It includes explanations of genetics risks, of inheritable factors, etc., and explanations of the functioning of the genome and its variations in the cellular metabolism. Genome 2 is the genome in the understanding of those who embody the genome, who "live" it, who are affected by it, who narrate it and understand their relationships to others by using elements of genetic knowledge and who make life plans accordingly (choosing a partner, planning a family and so on—remember Mechthild and Heinz). The genome is imagined and continuously reconceptualized in the lifeworld of those individuals and families who live the genome. This lived genome interprets[31] the biomedical construct of a physico-chemical entity that is called genome, which—in contrast to other parts of the body like the beating heart, blood flow, breathing, etc.—is not accessible to direct experience. It thereby integrates culturally mediated symbols and metaphors of genetics (such as the genome as a language, a text, a programme, a mosaic and the like) and combines them with personal understandings into a partially comprehensible and partially mysterious text.

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Part VII
Genetic Counselling

The Establishment of Genetic Counselling in Sweden: 1940–1980

Maria Björkman and Anna Tunlid

Abstract Genetic counselling in Sweden may be traced to the eugenics movement in the early twentieth century. A rudimentary form of what we might call genetic counselling today was practised within the state governed Medical Board in the 1940s and 1950s by the scientific advisor Nils von Hofsten. In the 1950s, Jan Arvid Böök, professor of medical genetics at Uppsala University, realised the importance of studies in broadly distributed genetic diseases. At the same time as he established a modern laboratory for chromosome analysis, he also held genetic counselling sessions. In Böökss's ways of navigating between the older traditions of eugenics and the new movement towards individual choice, there are signs of both continuity and discontinuity in relation to the Swedish eugenic project and population policy of the 1930s and 1940s. When the correct chromosome number of man was demonstrated in 1956, medical genetics as well as genetic counselling changed in many ways. New types of diagnosis could be made and new at-risk groups were identified. The geneticists trained at Böök's department contributed significantly to transfer both laboratory research and counselling activities from the academic setting to the clinic. Development of medical techniques like amniocentesis and prenatal diagnosis further increased the need for more systematised genetic counselling within the healthcare system.

In this chapter we provide an overview of the beginning of genetic counselling in Sweden. More specifically, we analyse the ways in which the first three generations of genetic counsellors constructed their roles as medical and genetic experts and the norms and values that characterized their counselling activities. We argue that this

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period was characterised by the development of a professional ethos that, while emphasising the importance of individual autonomy, also underscored the psychological and socioeconomic benefits of new diagnostic technologies to decrease the number of genetically diseased children. During the period, there was a marked shift from state-controlled eugenics to individual autonomy. However, we want to emphasise that not only did the individual autonomy increase but also the individual responsibility. At-risk individuals and families were supposed to make informed choices about their reproduction. And even if the individuals were at the centre, societal interests were clearly present, both as norms and values about what constituted a good life and as economic calculations within the healthcare system.

Keywords Genetic counselling • Medical genetics • Clinical genetics • Professional ethos • Biological citizenship

1 Introduction

Genetic counselling has a long history. Its roots may be traced to the eugenics movement in the early twentieth century, whereas the social practices associated with such counselling emerged after the Second World War. Genetic counselling has developed through many phases and has been influenced by not only the increasing knowledge about human heredity but also the social, political and institutional contexts in which such counselling has taken place. In this chapter we provide an overview of the beginning of genetic counselling in Sweden. More specifically, we analyse the ways in which the first three generations of genetic counsellors constructed their roles as medical and genetic experts and the norms and values that characterised their counselling activities. We argue that this period was characterised by the development of a professional ethos that, while emphasising the importance of individual autonomy, also underscored the psychological and socioeconomic benefits of new diagnostic technologies to decrease the number of genetically diseased children.[1]

2 Biological Citizenship, Biopower and Individual Autonomy

During the past three decades, the fields of biotechnology, biomedicine and genetics have witnessed rapid advancements in terms of research, diagnostics and treatments. Scholars have argued that this progress has not only changed the individual's expectations regarding possible medical treatments but also the

[1]This chapter builds on research conducted in the project "Better Humans or Reduced Suffering? Historical Perspectives on Medical Genetics and Genetic Counselling 1950–1980", financed by the Swedish Research Council [Vetenskapsrådet].

individual's understanding of, response to and sense of responsibility towards these new possibilities. In this perspective, the biological life is no longer perceived as a destiny but instead as something that can be changed, corrected and even improved. Scholars argue that this has led to the development of what is called a biological citizenship.

Biological citizenship can be expressed in a variety of ways. It is expressed, for example, when a group of affected individuals put pressure on the authorities to gain better medical treatment after a disaster or when individual self-identity is affected by the new possibilities of finding out one's genetic disposition.[2] But biological citizenship does not entail possibilities only. It can also imply anxiety among individuals faced with difficult ethical and existential decisions—for example, on reproductive issues—or in relation to the dissemination of sensitive information to family members or relatives. Thus, it is generally agreed that biological citizenship implies both possibilities and responsibilities.

The concept has been developed from Michel Foucault's idea of biopower. Foucault used the notion of biopower to characterise the factual power that monarchs of the nation states had over their subjects; such power could be expressed, for instance, in imprisonments and executions. Later, since states took on a greater responsibility for its citizens, the notion of biopower has been used to characterise more subtle ways of controlling individuals, not only through the development of, for example, demographic and statistic technologies but also through public health programmes. In the more subtle forms of health control, expertise specialised in health and medicine has got an increasingly important role.[3]

The sociologist Nikolas Rose and the anthropologist Paul Rabinow have developed and modified Foucault's ideas to construct an interpretative framework. This framework makes it possible to analyse different societal discourses about human life and health, as expressed by different experts, as well as some of their consequences. According to Rose and Rabinow, the interventions by these experts contribute to the development of "modes of subjectification" in individuals. These modes of subjectification can be explained as individual ways of internalising the discourses of health, thereby acting responsibly in relation to questions about life and health. Often, this responsibility concerns not only the individual but also future generations. Internalising different discourses of life and health and acting accordingly are important steps in the development of biological citizenship.[4] According to Nikolas Rose and Carlos Novas, the development of biological citizenship can thus be observed when individuals use their accumulated knowledge and insights and act as if the new knowledge "creates an obligation to act in the present in relation to the potential futures that now come into view".[5]

[2]Petryna 2002; Rose and Novas 2005, 440–463.

[3]Foucault 2003; Macey 2009, 186–205.

[4]Rabinow and Rose 2006, 193–217.

[5]Novas and Rose 2000, 486.

Several scholars have provided additional insight into the discussion about biopower and biological citizenship.[6] One of those scholars is the sociologist Ilpo Helén, who has drawn attention to the development of advanced medical technologies and their effects on biopolitics. According to Helén, technologies like amniocentesis and serum blood tests, which are used to diagnose illnesses and genetic anomalies in foetuses, have imposed a new mode of ethical subjectivity. This subjectivity arises from an ethical split in foetal diagnosis during which the providers of the technologies assume responsibility for only the reliability of the technologies while leaving the ethical and existential choices about the selective termination of pregnancies to the pregnant women.[7] Naturally, this puts women in a vulnerable situation. In contexts such as these, it is important to bear in mind that technology is never neutral. Instead it is infused with norms and values of its makers and the organisational settings in which it is produced and used. In this respect, technology and diagnostic methods can provide an indirect discourse of life and health to users. Moreover, the experts who mediate technologies also bring norms and values about reproduction towards the individual. Thus, new technologies and the possibilities of ever increasing personal reproductive choices also contribute to the development of biological citizenship.

The increased individual responsibility that Helén discusses is not a new phenomenon. In fact, a certain degree of increased individual reproductive autonomy was discussed and tentatively implemented in Sweden already from the 1950s onwards. From a state-controlled and collectivist perspective on human reproduction, the attitude changed slowly towards increased possibilities of choice. The most rapid changes, which took place during the 1960s and 1970s, resulted from a growing critique of social policy, expert knowledge and ideals of social engineering as well as from feminism gaining ground. Among other things, this resulted in the introduction of a liberal abortion law in 1974.[8]

The relation between eugenics and biological citizenship is a contested issue among scholars. Rose and Novas stress the historical break, emphasising the different norms, values and practices that guided eugenics as compared with biological citizenship, stating that "[. . .] the links of biology and human worth and human defects today differ significantly from those of the eugenic age".[9] An important difference, according to this view, is that whereas the eugenics project was directed at the improvement of the population, biological citizenship concerns the individual management of genetic information. Accordingly these scholars and several others have applied the notion of biological citizenship primarily while discussing the past two or three decades. However, others have argued that the break between the past and the recent may not be so clear and that a focus on

[6]Rose and Novas are among the most influential, but see also Lemke 2011 for a critical review.

[7]Helén 2004, 28–54; Rose 2001, 1–30.

[8]Kerr and Shakespeare 2002, 65–69; Grunewald 2009.

[9]Rose and Novas 2005, 440.

changes may conceal the continuities that actually exist.[10] We share this latter view and identify several characteristics of biological citizenship in earlier times, for example, in the 1940s and 1950s.[11] In this chapter we use the concept of biological citizenship as a tool to analyse the discourse of health and disease that formed the basis of genetic counselling.

3 Genetic Counselling: A Beginning

The term genetic counselling was coined in 1947 by the American geneticist Sheldon Reed (1910–2003) while working at the Dight Institute for Human Genetics in Minnesota. At the time, this was a clinic that advised individuals on hereditary and eugenic issues. According to Reed's 1975 autobiography, he envisioned genetic counselling in the 1940s to be "a kind of social work, without eugenic connotations".[12] However, when studying the beginnings of the history of human genetics, it can be difficult to determine where eugenics ended and human genetics started: human genetics, at its inception, had intimate connections to eugenics. During the 1950s, in the aftermath of WWII and the Nazi applications of race hygiene, it became a more pressing task in various countries to disconnect the emerging discipline of medical genetics from eugenics. But the older eugenic practices and the medical genetics and genetic counselling activities were often interwoven with each other in complex networks of institutions, practitioners and social relations. This is stated and exemplified by historian Diane B. Paul in her account of the history of the American Society of Human Genetics. Five of the six first presidents of this society were also board members of the American Eugenics Society. Several of these presidents considered genetic counselling important, not only for the purpose of providing individuals and families with genetic information but also to prevent unwanted traits from spreading in the population.[13] Similar connections between eugenic practices and medical genetics can also be seen in the Swedish example from the 1950s.

In fact, a rudimentary form of what we might call genetic counselling today was practised in Sweden within the state-governed Medical Board in the 1940s and 1950s. The counselling was given by the scientific advisor and zoology professor Nils von Hofsten (1881–1967). von Hofsten exerted a major influence on Swedish eugenics and its practical applications from its introduction in the beginning of the 1900s to well into the 1960s. The events leading to von Hofsten's influence were complex but could be summarised as follows. von Hofsten was one among a

[10]Kerr 2003, 44–50; Koch 2004, 315–331; Raman and Tutton 2010, 711–734; Comfort 2012.

[11]Björkman 2015, 489–513.

[12]Cited from Resta 1997, 376. Stern 2012, 20, suggests that Reed had a strong eugenic agenda in many cases.

[13]Paul 1995, 121, 125–126; Stern 2012, 18.

number of eugenically minded and genetically interested scientists who contributed to the introduction of genetics to the Swedish audience in the first decade of the twentieth century. He engaged in a successful eugenics lobby campaign aimed at opening a race biological research institute in Sweden, which eventually opened in 1922. Following the institute's opening, he became one of its board members. He wrote several influential textbooks on genetics, published mainly in the 1920s and 1930s.[14]

Apart from the race biological research institute, another aim of the eugenic network was to launch a sterilisation act, which was passed in 1934. Following this, von Hofsten became a scientific advisor to the group within the Medical Board that decided on sterilisation applications, a position he held between 1935 and 1953. The law was revised shortly after it was launched; this process was led by von Hofsten as a member of a large population commission that was active between 1935 and 1938. The revision resulted in the Sterilisation Act of 1941, which considerably widened the target group for eugenic sterilisation. This widening led to what has been called "a sterilisation offensive" in Sweden.[15] The offensive was directed against individuals that were considered to be of lower biological quality and thus not appropriate for engaging in reproduction. During his entire career, von Hofsten believed that if a sufficient amount of "feebleminded" individuals (one of the main target groups of the 1941 Sterilisation Act) were sterilised, this would have a positive effect on the population. Thus, von Hofsten encouraged methods of increasing the number of voluntary sterilisations, methods that are viewed today as coercive.[16]

Eugenicists like von Hofsten also stressed on the importance of the procreation of biologically fit individuals for Sweden to secure its position as a prominent civilisation which he and the lobby group persistently claimed. The dissemination of eugenic ideas in the population from around 1910, together with the sterilisation practiced since the second half of the 1930s, contributed to an awareness of the importance of taking responsibility for one's reproductive activities especially in relation to heredity.

This responsibility also caused an unintended consequence, a phenomenon von Hofsten coined "heredophobia".[17] This condition could affect women, men or couples and was expressed as unfounded fear and anxiety about passing on hereditary diseases to one's children. This fear was at times so strong that it impelled the affected to apply for abortion, sterilisation or both in order to avoid passing on hereditary defects to their offspring. von Hofsten found this troubling as he believed that individuals affected by heredophobia often belonged to the part of the population that should be encouraged to reproduce in view of their fine biological qualities.

[14]von Hofsten 1919, 1923, 1927; 1931.

[15]Björkman 2011, 161–205.

[16]Ibid.

[17]von Hofsten 1963.

von Hofsten started to provide genetic counselling to calm and comfort these individuals, as a strategy to cure and prevent the condition.[18] His activities can be traced from a series of reports he wrote to investigate especially complicated cases of abortion and/or sterilisation applications, to which the heredophobia cases belonged. von Hofsten reported the results of his investigation of hereditary information presented in each case. He calculated genetic risk based on this information and genetic literature. In his report, he either endorsed the application or objected to it. Although it was the board that made the formal decisions, von Hofsten's recommendations were almost always followed.[19]

The number of heredophobia cases represents merely a fraction of the total number of sterilisations and/or abortions performed. Nevertheless, these cases are interesting because they provide information about how individuals could react to and internalise information about eugenic communication and intervention from hereditary experts. They are also interesting because the fear expressed through the abortion and sterilisation applications exemplifies an increased demand for genetic counselling services.

When individuals applied for abortions or sterilisations out of fear of passing on severe inherited diseases and the individual clearly had a predisposition to such diseases, von Hofsten would recommend the intervention applied for. When it came to the heredophobia cases, though, he found that the fear was unrelated to the factual hereditary patterns of the individual. Even so, he found that in some cases the fear itself posed a threat to the health of the individual. In von Hofsten's opinion, this could justify an intervention to restore the psychological health of the affected individual. This was the case in 1948, for example, when a woman diagnosed with psoriasis applied for sterilisation out of fear that she would pass on the condition to her offspring.[20] This woman was examining her children ceaselessly to see if they had developed any signs of psoriasis. According to her doctor, she was in a distressed state due to the worries. While calculating the risk, von Hofsten wrote that the woman's children had a 12 per cent risk of developing psoriasis, which he considered to be minor. In spite of this, he felt the need of recommending the sterilisation because the woman's worries seriously affected the quality of her life and that of her children. He wrote:

> It is understandable that Mrs. X, in her belief that her children are endangered, has started to observe them, looking for rashes, but after the information now provided she should of course not do so; by doing that she is damaging both her own and her children's nerves. Since she, through the sterilisation, has eliminated her fear of pregnancy, she should, as the capable and decent person she evidently is, attain a better mental balance and start to hope the best for her children.[21]

[18]Ibid.

[19]Tydén 2002, 301–303; Runcis 1998, 219–232; Björkman 2011, 163–164.

[20]Protocol 19 May, 1948. Sinnessjukvårdsbyrån, avd. rättspsykiatri. Korr. i abort- och steriliseringsärenden. 1935, EIX vol. 1, MB, SNA.

[21]Ibid.

In this case, von Hofsten assessed that this woman was both "capable" and "decent", qualities he did not usually ascribe to individuals he considered to be suitable for sterilisation. It is worth noting that this woman belonged to a group von Hofsten considered suitable for reproduction.

Even though heredophobia could sometimes lead to sterilisations, abortions or both, in most cases von Hofsten did not consider the condition serious enough to motivate a surgical intervention. In these cases, his strategy was to instead "calm and comfort" the affected individuals by writing personal comments to them in the report to the Medical Board. In such cases, the report could work as a therapeutic document, which aimed at helping the individual to overcome her or his fears. In two cases, von Hofsten later recalled, he even went further in his efforts to calm individuals by either telephoning them or making a personal visit.[22]

One heredophobia case, from 1950, illustrates how von Hofsten's calming and comforting in the report was supposed to serve as a therapeutic tool. In this case, a woman applied for an abortion due to her fear that the foetus she carried would be affected by the same severe and multiple physical deformities as a previous child of hers.[23] When von Hofsten performed his genetic risk calculations, he found that the risk of inheritance was not sufficient to motivate an abortion. The woman's fear was not of a kind that motivated the intervention either. Instead, von Hofsten tried to calm the woman by writing in his report:

> One can also ask Mrs. X to try to realise that worrying about the defects of her last child does not help anything get better, quite the opposite. It might be a poor consolation to know it could have been worse, but she should anyway consider the many mothers who have children with more severe defects, especially those of a psychological nature.

That this consolation from von Hofsten was intended as a therapeutic document becomes even clearer when we consider the fact that the woman's physician helped her with the application because he believed that a report from the Medical Board would help calm her. Thus, this case exemplifies that the contact with the board at times could serve primarily to calm worried individuals. This also indicates a sort of alliance between the representative of the Medical Board and the physician.

The actions taken by the women in these two cases exemplify what can be defined as an early emergence of biological or genetic citizenship. The two women, with the aid of their physicians, applied for sterilisation or abortion owing to their fear of passing certain conditions onto their offspring. These surgical interventions would have had a major effect on their present and future reproductive capacities, so both their worries and their decisions show that they acted with clear responsibility in relation to the biological knowledge they had access to. Not only were they able to access expert knowledge via the Medical Board, they also had their responsibilities and decisions examined by this body.

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[22] von Hofsten 1963, 50.

[23] Protocol 8 August 1950. Sinnessjukvårdsbyrån, avd. rättspsykiatri, Korr. i abort- och steriliseringsärenden, 1935, EIX, vol. 1, MB, SNA.

4 Genetic Counselling Within Medical Genetics

In the 1950s, genetic counselling was also carried out outside the context of the Medical Board within the emerging discipline of medical genetics in Uppsala. This was done by the geneticist Jan Arvid Böök (1915–1995). Böök started his career as a classical geneticist under the guidance of the geneticist Arne Müntzing (1903–1984) at Lund University. In addition to this education, he was also trained as a physician with a specialisation in psychiatry. Between 1951 and 1956, he was employed at the Institute of Race Biology as deputy director under Gunnar Dahlberg (1893–1956) and in 1957, he succeeded Dahlberg as Head of the Institute of Race Biology. A few years later, the institute was transferred to Uppsala University and incorporated in the medical faculty as the Department of Medical Genetics (see below).

In the 1940s, Böök held traditional eugenic views that supported the idea that the Swedish population could be improved by a sufficient number of sterilisations of biologically "defective" individuals. In 1941, he and von Hofsten gave a lecture—held under the auspices of the National Association for Public Health—on the population benefits of sterilisations. This organisation was started in 1941 by mainstream eugenicists as a protest against the emerging left-wing eugenics. By lecturing on this subject, Böök clearly positioned himself in the group of eugenicists that believed in state-controlled sterilisation to prevent the spreading of physical and mental disease in the population.

By the 1950s, however, Böök no longer believed that sterilisations would have any positive health effects at the population level, and he also emphasised individual autonomy in reproduction issues to a greater extent than von Hofsten did.[24] The difference in the views of the two men is exemplified by a conflict between the two while working as heredity experts in a state committee on medical impediments to marriage, which was set up to revise the Swedish Marriage Act of 1915. The work of the committee, which took place between 1956 and 1960, was marked by a split between two groups of experts. On one side was von Hofsten who defended the power invested in the previous Act to prevent marriages he considered eugenically unwanted. On the other side were Böök and the psychiatrist Carl-Henry Alström (1907–1993), who clearly distanced themselves from the eugenic approach of the previous Act. Böök and Alström stated that there was no intrinsic value in limiting marriages due to medical reasons. This was because it was impossible to limit the spreading of genetic disease in the population by preventing marriages (or using eugenic sterilisations). This was because most genetic diseases were (and are) spread recessively, and therefore it was impossible to identify all at risk of passing disease onto their offspring. Instead, it was the social effects of medical conditions on marriage that were important. von Hofsten agreed that most genetic diseases were impossible to prevent by marriage legislation or eugenic sterilisation, except

[24] Björkman 2011, 149-150, 191-192.

for the feebleminded. Alström and Böök did not agree with von Hofsten.[25] However, they found it important to consider heredity when it came to abnormal conditions or diseases that could cause lifelong suffering for children born within marriage.[26]

Böök's engagement in this committee was not the only activity during the 1950s in which he drew a line between medical genetics and older eugenic activities. In 1955, he was asked by the American Eugenics Society to write an article about medical genetics and genetic counselling for their journal *Eugenics Quarterly*. The article was to be placed in the section of "hereditary counselling" and was motivated by the fact that the journal noticed an increased interest in the journal from Europeans, according to the managing editor Mrs. Helen G. Hammons (1905–1996). The American Eugenics Society supported "research and educational activity directed toward increasing of the proportion of children born with better than average potential for intelligence and character and toward diminishing the burden of hereditary disabilities".[27]

Böök wrote the article, but in doing so he emphasised:

> Genetic counselling, in a wide sense, consequently has its individual as well as medico-social aspects. The latter are concerned with the genetic risks for larger groups of people and how these risks can be met with public information, legislature, changes of the environment and other procedures. This field of activity, which could be called the epidemiologic control of genetic disorders, has hardly been touched upon yet insofar as public health organizations and planning are concerned. I should like to make it perfectly clear that such undertakings have practically nothing in common with previous so-called eugenic movements, the most outstanding effect of which has been to discredit medical genetics.[28]

Even though Böök repudiated older eugenic traditions in the text above, it is interesting to note that he defined the task of medical genetics to take "epidemiologic control of genetic disorder". This matches, for example, what von Hofsten considered to be the objective of eugenics.

In the 1950s, Böök also provided genetic counselling at the Department of Medical Genetics. Patients were referred to him from hospitals around Sweden, and he also counselled "walk-ins". The patients that were referred to him could for example be affected by skeletal deformities, Basedow's disease, haemophilia or Huntington's disease. Those who contacted Böök on their own initiative often asked for correct information on heredity. This was why a Russian-Jewish couple wrote to Böök from the USA in 1955. They had lost their 18-month-old daughter to Tay-Sachs disease and wanted to know the exact heredity patterns for the disease. Others had questions about marriages between cousins, possible genetic effects of radiation or treatment of children with mental disabilities.

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[25]SOU 1960:21, 31–32; von Hofsten to Romanus April 30, 1960 (with appendix). Komm. för med. äktenskapshinder 1956, YK 2178, MB, SNA.

[26]SOU 1960:21, appendix 3, 134–140.

[27]Hammons to Böök, 19 January 1955, IMG F6:25, UUA.

[28]Böök 1955a, see also article manuscript "Medical Genetics and Counselling Practices", p. 6, IMG F6:25, UUA.

Even though Böök never used the term heredophobia in documenting his counselling sessions, he did hear from individuals that held deep concerns about hereditary issues. For example, in 1958 Böök corresponded with a young individual that was deeply worried about radioactivity and X-rays in relation to future marriage. Böök wrote to the individual to offer support, stating that it was not possible to hold "any eugenic obstacles to marriage." Furthermore, "That is something that society should not interfere with. The question of whether or not to have children should be resolved in a voluntary way through education as well as access to specialist consultations".[29]

Here, Böök emphasised the role of the genetic expert when it came to deciding on whether to have children or not. While stating in this passage that reproductive decisions should be voluntary, Böök also suggested indirectly that genetic expertise, through education and counselling, should provide the information that individuals needed to exercise their free will. In this way, the experts could transfer their values to the individuals while still leaving the decisions to them. Böök thus encouraged individual free choice among his counselees while also claiming that they needed expert help to be able to make reproductive decisions. Individuals also seemed to accept the need for expertise, as we have seen in the examples from von Hofsten's and Böök's counselling activities.

Another example from Böök's counselling illustrates these educational ambitions. In this case, Böök educated a counselee on how to assess genetic risk when the counselee wrote to him expressing worries about marrying a cousin.[30] In his reply, Böök confirmed that there was a higher risk of a child born of a marriage between cousins to inherit certain diseases as compared with a child born of a marriage between unrelated spouses. However, several of the diseases that could be passed on were medically treatable, Böök stated. Such treatable diseases did not need to be considered as "disastrous". He continued:

> In principle, I do not believe that one should give any definite advice. Attitudes to risks are very diverse. It is, however, important to have a rough understanding of the state of things. Then you should not worry in advance. It is, after all, most likely that everything will go well. If you do have a defective child after all, then it is a new situation. Then a proper investigation should be conducted. It will then be possible to assess the risks for any following children with greater accuracy. Of course, I believe that you should marry the one you hold dear, and not enter into marriage on the basis of a scientific investigation. One should not focus blindly on just a specific kind of risk. One constantly takes various risks in life without constantly worrying about them.

In Böök's view, the proper time for worrying was not in advance but first when a "defective" child was born. By comparing genetic worries to other things one worries about, Böök encouraged the counselee to take on his (Böök's) own view of the matter.[31]

[29]Böök to counselee, 20 October 1958, IMG F6:11, UUA.

[30]Böök to counselee, 1 June 1956, IMG F6:11, UUA.

[31]Ibid.

5 A Modern Institute for Medical Genetics

When Böök succeeded Dahlberg as professor in 1957, the position was still located at the Institute of Race Biology. In 1958, the institute requested that its name be changed. According to its board, the term "race biology" was in many respects inappropriate for the research at the institute. Moreover, "race biology" (Rassenbiologie) was totally discredited due to its use in Germany under the Hitler regime.[32] When the institute was incorporated within Uppsala University in July 1959, it re-emerged as the Department of Medical Genetics. At the same time, Böök's professorial subject turned from race biology to medical genetics. The suggestion to change the name was probably initiated by Böök, who as newly appointed professor worked hard to transform the institute into a modern research institute devoted to medical genetics.

To meet this ambition, the institute needed not only a reorientation but more resources.[33] The main argument that Böök used to obtain more funding was that genetic diseases had become a major public health problem in modern society. According to Böök, 5–10% of the population were at risk of getting "a serious genetic disease" during their lives. The increasing emphasis on genetic diseases had several causes. The use of antibiotics and vaccines, improved sanitation, and the control of nutritional deficiencies had reduced mortality from infections. It had caused a shift in the major threat to public health, from external causes to "disease-producing agents and weaknesses that are inherent constituents of the individual"—that is, genetic disorders.[34] Another important change was the increasing number of people that got older and hence would suffer from genetic diseases later in life. Finally, a new threat was looming: ionising radiation that would increase the mutational load of the population and thereby the risk of genetic morbidity and mortality.[35] Hence, there was a strong focus on epidemiology and public health when Böök tried to legitimise his new research programme.

However, studies at the population level were not enough. Böök emphasised strongly that the focus on genetics as well as statistical and mathematical methods, which had been the focus under Dahlberg's leadership, had to be complemented by experimental research to uncover the mechanisms of genetic diseases and ultimately to develop treatments for them. The hope to be eventually able to develop cures for genetic diseases, once they had been identified, was a strong driving force for medical genetics and at the same time a way to legitimise the new research field. With funding from private foundations such as the Rockefeller Foundation and the Swedish Knut and Alice Wallenberg Foundation, Böök managed to set up a new laboratory for biochemical studies as well as cell culturing.[36] On the

[32]Petita 1959/60, RBI B1:3, UUA.

[33]Proposal to the Government, 1 August, 1957 and Petita, 1958/59, RBI B1:3, UUA.

[34]Böök 1955a, 174.

[35]See also Böök 1955b.

[36]RF Collection, diary "EC 2/24/56", R.G.1.1., Series 800, Box 5:32.

recommendation of the medical geneticist Lionel Penrose (1898–1972), he recruited Marco Fraccaro (1926–2008), who had been working at Penrose Laboratory for a couple of years, to serve as deputy director of the institute and be responsible for the experimental work at the laboratory.[37] A few years later, the medical student Jan Lindsten (b. 1935), who had a background in genetics and statistics, joined the group and initiated a productive collaboration with Fraccaro.

6 Cytogenetics and the Search for Chromosome Aberrations

At the same time, as the new laboratory was developing at Uppsala University, a discovery was made at the Department of Genetics at Lund University; this discovery would change the fundamentals of medical genetics. In early 1956, Joe Hin Tjio (1919–2001) and Albert Levan (1905–1998) from the department published a paper suggesting that the correct chromosome number for humans was 46, not 48, thereby challenging the consensus at the time.[38] The paper attracted the attention of human geneticists around the world, and the suggestion by Tjio and Levan was soon confirmed by several other research groups.[39] Having established the correct chromosome number, researchers began searching for deviant numbers. In 1959, Jérome Lejeune (1926–1994) and his co-workers in Paris demonstrated that Down's syndrome was associated with an extra chromosome (chromosome number 47 rather than the normal 46). In the years that followed, the search for aberrant chromosome numbers and their links with various clinical syndromes became a hot topic of research in the field of cytogenetics.

The institute at Uppsala jumped on the bandwagon. One reason might be that the study of biochemical genetics in blood cultures, which was one of the main projects at the new laboratory, turned out to be more difficult than expected and did not produce many results. Instead, tissue culturing of the skin and bone marrow from human foetuses and adults was undertaken for chromosome analyses (karyotypes) and to look for chromosome aberrations. The change in research focus turned out to be successful; in 1959, the Uppsala cytogeneticists managed to publish several papers on chromosomal aberrations related to Down's and Turner syndromes.[40] These were not the first reports of the two chromosomal disorders: Charles Ford (1912–1999) and his co-workers at the Medical Research Council's (MRC) Radiobiology Unit at Harwell had, for example, already reported a karyotype with a missing X-chromosome in a female with Turner syndrome. But Böök and his colleagues were on the same track as the pioneers. Indeed, when the paper by

[37]Harper, interview with Fraccaro. In: www.genmedhist.org/interviews.

[38]Tjio and Levan 1959. See also Martin 2004; Harper 2006; Arnason 2006; de Chadarevian 2015.

[39]de Chadarevian 2015, 133.

[40]Böök 1959a; Böök 1959b; Fraccaro 1959a and b.

Ford and co-workers was published, Böök wrote to Lejeune: "As you might have seen the English have beaten us with their publication in Lancet about three weeks ago".[41]

During the years that followed, research on new chromosomal syndromes or variations of syndromes that were already identified became a main focus of the laboratory in Uppsala. According to Böök, karyotyping was of great importance for both diagnosis and prediction of genetic diseases and defects. This was reflected in his application in 1961 for a large grant from the Swedish Medical Research Council for a programme called "Clinical and cytogenetic investigations of some diseases and defects".[42] In the application he assumed that several hospitals would soon have cytogenetic laboratories and indicated that the programme would be of great clinical value. It should range from karyotyping of congenital diseases and defects with uncertain aetiology to more detailed studies of variations in karyotypes of specific clinical diagnoses such as Down's syndrome. The research council funded the programme for 10 years, during which new research questions were added successively.

The laboratory seems to have been most productive in the late 1950s and early 1960s.[43] In 1962 there were 35 persons working at the department, according to Böök, who never missed an opportunity to point out that it was the only department in Sweden that was devoted entirely to medical genetics and that his own professorship was the only one designated for this subject. As a consequence, the department became almost an obligatory point of passage for PhD students with an interest in medical genetics.[44] During this period the institute also had numerous international contacts. For example, a training programme in the early 1960s, funded by NIH, made it possible for physicians from the USA to work at the institute and learn medical genetics.

The research was conducted in cooperation with physicians working at different hospitals and treating patients with genetic diseases or chromosomal disorders. Sometimes, these physicians contacted Böök to discuss complicated cases and unclear diagnoses; at other times Böök took the initiative. In 1959 he wrote to a physician:

> Presently we are conducting detailed studies [of mongolism] and of course we also want to examine parents, brothers and sisters. Families of the kind you are describing in Acta. Genet. 7:533–549, 1958 is of special interest to us. I wonder if it would be possible to get biopsies (preferably bone marrow and skin, or possibly only skin) from the mother, the

[41]Böök to Lejeune, 8 May 1959, IMG B2:2, UUA. Lejeune had some exchange with Böök and the other researchers at the institute in Uppsala. He visited the institute several times in the late 1950s.

[42]Böök, Kliniskt cytogenetiska undersökningar vid valda sjukdomar och defekter. Application to SMR, 21 January 1961, SMR F1:15.2, SNA.

[43]Both Fraccaro and Lindsten have testified to the increasing problems and conflicts at the laboratory. They both left in the late 1950s/early 1960s. See interviews with Peter Harper in www.genmedhist.org/interviews.

[44]The other place to do research on human genetics was Albert Levan's cytogenetic laboratory, Lund University, but this laboratory was almost entirely devoted to cancer chromosome research.

> child and the grandfather of the family you have investigated. If only skin biopsies are taken, they are so small that they are of almost no discomfort to the patients.[45]

These contacts made possible for Böök to get research material in the form of tissues—skin biopsies, bone marrow and blood—for culturing. Besides this, tissues could also be obtained from aborted foetuses. The easy access to research material for cytogenetic studies was, according to Böök, a specific advantage for Swedish cytogeneticists.[46] One reason was that abortions on specific indications were legal in Sweden since 1938 although getting permission was a complicated process, as is evident from the previous section about von Hofsten.

The contacts with physicians created links between scientific research and clinics. This also aroused a nascent interest in genetics among some physicians (mainly paediatricians, gynaecologists and psychiatrists). Böök received many requests for chromosomal analyses, but he was quite selective with this kind of work because such analysis was both expensive and time-consuming.[47] At the same time, he strongly emphasised the need for professional medical geneticists in clinical practice. As he stated in a letter to the geneticist Theodore Puck (1916–2005): "I do not, of course, object to the fact that genetics is disseminating into all sorts of clinical specialities [sic] but I feel it is essential that a trained geneticist is given the responsibility of evaluating and integrating the results. If every clinician or clinical biochemist is going to be his own genetical [sic] expert without sufficient training in genetics, it will be impossible to maintain respect for medical genetics as a speciality".[48] Together with cytogenetic colleagues from the Nordic countries, Böök argued for more resources to laboratories that could do this service to the clinics. In a joint statement, they emphasised that cytogenetic service could only be carried out by persons with training and experience in cytogenetics, thereby stressing the link between scientific research and the clinic. In the long run, there was, according to the group, a need for specialisation in medical genetics in the medical education.[49]

7 Medical Genetics Moves to the Clinic

The early 1960s also saw a growing interest in medical genetics at some hospitals in Sweden. At Uppsala Academic Hospital, a small chromosome laboratory, funded by a private foundation, was set up at the paediatric clinic under the leadership of

[45]Böök to Schlaug, 25 maj 1959, IMG B2:2, UUA.

[46]Böök, Kliniskt cytogenetiska undersökningar vid valda sjukdomar och effekter. Application to SMR, 20 March 1962, SMR F1:151, SNA.

[47]Böök to the vice chancellor's office, Uppsala University, 16 August 1962, IMG B2:3, UUA.

[48]Böök to Puck, 18 August 1961, IMG F6:6, UUA.

[49]Protocol from Nordic clinic cytogenetic conference, 16–17 December 1961, IMG F6:3, UUA. See also Böök 1962, 1037–1038.

the paediatrician Karl-Henrik Gustavson (b. 1930).[50] He was one of Böök's co-workers and was working on a thesis about the relation between karyotypes and clinical symptoms of children diagnosed with Down's syndrome.[51] The laboratory diagnosed genetic diseases, estimated the risk of transferring the diseases to future children and provided genetic counselling.

A similar development took place in Stockholm. Jan Lindsten (b. 1935), who began doing chromosome analysis at the Department of Medical Genetics at Uppsala University in the late 1950s, moved to the Karolinska Institutet (KI) in 1960 to pursue his PhD studies. In 1963, he defended his thesis about chromosome aberrations in Turner syndrome.[52] In addition to carrying out cytogenetic studies, he also analysed in detail the clinical picture of Turner patients to provide material for further studies in clinical genetics. The research was conducted at the Department of Endocrinology at KI, where a small division of medical genetics was established with Lindsten as its head. Besides his own research, he provided cytogenetic analyses to other clinics, taught genetics to medical students and provided genetic counselling to outpatients. An explicit aim of his research was to develop the knowledge in support of genetic counselling, for example, chromosomal aberrations related to repeated spontaneous abortions or when parents that had conceived a child with congenital disorders wondered about the risk for a future child.[53]

During the 1960s, cytogenetic laboratories were also set up at some other hospitals, but none was run on a regular basis. Such laboratories reflected the initiative taken by individuals with a special interest in medical genetics and cytogenetics. Besides doing cytogenetic analyses, the geneticists working at these laboratories also provided genetic counselling. They often emphasised that genetic counsellors needed both genetic knowledge and clinical experience and that it was an advantage to perform counselling at the hospital where it could be integrated in the clinical practice. This led to the emergence of a profile of the genetic counsellor as a person trained in both genetics and medicine, and genetic counselling began to appear increasingly as a task for experts. This development also meant that genetic counselling moved gradually from academic institutions to the medical healthcare system. However, it was not until the late 1970s that clinical genetics, including genetic counselling, became established as a speciality within the Swedish healthcare system.

[50]Jerring to Vahlquist, 31 December 1962, IMG E1:5, UUA.

[51]Gustavson 1964.

[52]Lindsten 1963.

[53]Lindsten, Strukturella autosomala aberrationers betydelse för uppkomsten av. missbildningar och utvecklingsstörningar hos människa, 17 March 1965; Lindsten, Betydelsen av. strukturella kromosomaberrationer för uppkomsten av. spontana aborter, perinatal dödlighet, missbildningar, och utvecklingsstörningar hos människan, 30 March, 1966. Applications to SMR FI:302, SNA.

8 Genetic Counselling at the Clinic

During the 1960s, genetic counselling was still very much based on risk calculations and risk assessments.[54] A precondition for such counselling was a diagnosis of the disease as well as knowledge of the inheritance pattern of the disease. In some cases, the disorder followed a Mendelian inheritance and the risk could be calculated based on the Mendelian laws. However, several clinical diagnoses were genetically heterogeneous — the same clinical symptoms could have different genetic backgrounds — and the genetic causes (the aetiology) for many genetic disorders were still unknown. In such cases, one had to estimate the empirical risk. Such calculations, which were based on epidemiological studies, were similar to the ones that Böök had performed in the 1950s.

Another tool for genetic counselling was chromosomal analysis. In the 1960s, these could be performed only postnatally as the technology of analysing foetal cells in the amniotic fluid was not yet developed. The analyses were used for diagnosing suspected chromosomal disorders such as Down's syndrome in children and adults. Usually, these kinds of syndromes were not inherited—that is, they were not caused by deviation of the parents' chromosome. But there were some exceptions. In Down's syndrome, for example, a family pattern was recognised in a few cases. In such cases, there was a hereditary risk, and a chromosomal analysis of the parents could be used to calculate the risk of having another baby with the syndrome.[55]

Most people that sought genetic counselling had a child with a disorder or condition that they (or the physician) suspected was hereditary: they wanted to know about the risk of having another child with the same disorder. The task of the genetic counsellor, according to its practitioners, was therefore to inform the parents about the heritability of the disease and to provide information about the estimated risk of having another child with the same disease. The counsellor was not to influence the parents' decision regarding whether to have a child or not. The ethos of individual autonomy was thus stressed. This was sometimes further underscored by stating that genetic counselling had no eugenic intention but was in the interest of the individual or the family.[56]

Despite the emphasis on objective and neutral information, some of the genetic counsellors came to recognise the psychological aspects of the situation and the difficulties the counselees faced in making decisions on these issues. One aspect was related to the level of risk that people were willing to accept. According to the paediatrician and genetic counsellor Karl-Henrik Gustavson, most people accepted a risk that was less than 10% but were less willing to accept a risk of 25%.[57]

[54] See Stern 2012, 28–52, for an extensive discussion about risk assessment in genetic counselling.

[55] This kind of familial occurrence of Down's syndrome was discovered by several cytogeneticists in 1960, see, for example, Fraccaro 1960.

[56] See, for example, Gustavson 1967.

[57] Ibid.

Another aspect was the guilt that many parents experienced at conceiving a child with a genetic disorder and their anxiety about having another child with the same problem. As Gustavson pointed out, discovering that a child was mentally disabled was a big trauma for many parents, and they often wanted to know the risk for another child. Like Böök, Gustavson also stressed that the risk was often less than what parents expected, and in these cases the counselling could serve to calm them. A common way of reducing the feeling of guilt was to point to the accidental nature of the situation such as, for example, in the case of Down's syndrome. In other cases, the counsellor could explain that all people were carriers of several recessive genes, and it was only if the parents had the same recessive genes that they were at risk of having an afflicted child. In this way, the psychological part of the counselling was mixed with educational elements, similar to both von Hofsten's and Böök's approaches to genetic counselling. However, the fact that parents felt guilty indicates that they faced not only a rational decision but were also confronted by larger questions to do with health, disease and the value of life.

9 The Advent of Prenatal Diagnosis and Prevention

In the 1970s, the context for genetic counselling changed dramatically due to the development of amniocentesis—the possibility of withdrawing a small amount of the amniotic fluid that contains foetal cells and to culture these cells in the laboratory.[58] This procedure opened up the possibility of identifying chromosomal disorders of the foetus as well as some genetic diseases that could be detected by biochemical analysis. The most common diagnosis based on this technology was some kind of chromosomal deviation. The prenatal diagnosis increased the demand for genetic counselling markedly, thereby also raising new ethical questions. This placed prospective parents who worried about giving birth to a genetically diseased child in an entirely new situation. Instead of having to make a decision based on risk figures, they could now base their decision on more direct knowledge of whether the foetus was affected or not.

Prenatal diagnosis was first introduced in Sweden in 1970. In 1972, it was already being practiced at several large hospitals in the country, "a routine diagnosis" as it was called in a short paper introducing the technology in the Swedish journal for physicians (Läkartidningen).[59] The paper noted that the technology allowed the diagnosis of several genetic diseases early in pregnancy: this made it possible to abort foetuses that were affected and avoid abortion of healthy foetuses. When amniocentesis was offered to "risk families", as the paper put it, it was possible to "guarantee that a future child will not have the genetic disorder of which they have an increased risk". Prenatal diagnosis was, according to the paper's

[58] See Stern 2012, 147–167, for a brief history of the development of amniocentesis.

[59] Kjessler 1972.

authors, important from both family and socioeconomic perspectives, even though the latter aspect was not elaborated further. Some of the themes introduced in this paper would go on to spawn discussion on amniocentesis during the following years.

One of the themes concerned the criteria used to determine the eligibility of women to take the test—that is, the kind of indications that would be considered valid. According to the medical geneticist and psychiatrist Hans Olof Åkesson (1933–2005), it was not possible to offer amniocentesis to all pregnant women who desired to have the test even though that would give "maximal effects". Economic factors precluded the test being made available to all pregnant women.[60] Instead, the focus was on particular groups of individuals that were considered to be at risk.

One group consisted of the so-called risk families. These were families that already had an affected child or parents with a known hereditary disease. This was the same group that had in the past turned to genetic counsellors for information and discussions about the risk of having another child with a disease or genetic anomaly. Another group that was often considered to be at risk was pregnant women older than 35 years, as it was well known that the risk of having a child with Down's syndrome increased with age.[61] A third group included pregnant women that worried about having a child with a chromosomal aberration although they did not belong to any known risk group. According to one study, some of these women would have applied for a legal abortion if they had not been offered the prenatal diagnosis.[62] Such cases suggest that the "heredophobia" that von Hofsten identified in the 1940s persisted in the 1970s.[63] Medical and psychological indications were thus considered to be valid reasons for requesting amniocentesis, but the requests of women that simply wanted to know the sex of the expected child were denied. Thus, the genetic expertise served to set up the framework for when and how this new technology was to be used.

The possibility of prenatal diagnosis increased the demand for genetic counselling. A woman that wanted to undergo amniocentesis had to be informed about the test as well as the advantages and the risks associated with the procedure (although most physicians considered the risks very small). In addition, the woman had to be informed that the only "treatment" when a chromosome aberration was identified was an abortion. Whether amniocentesis should be offered to a woman that, for moral or some other reason would not accept an abortion, was an issue of discussion among physicians. Some thought that these women should not be offered the examination.[64] Others argued from a more psychological perspective and thought that these women should also be offered prenatal diagnosis. They felt that in case

[60] Åkesson 1973.

[61] The risk for older women to get a child with Down's syndrome had been demonstrated by Penrose. See Kevles [1985] 1995, 161–162.

[62] Bartsch et al. 1973.

[63] Munthe 1996, 22, 26, 29–30.

[64] Bartsch et al. 1973.

the foetus was found to be healthy, the examination would have a calming effect, whereas if the foetus was found to have a congenital disorder, it would be better to know this.[65]

In general, though, there was agreement among the genetic counsellors that they should not interfere in women's decisions regarding abortion. According to the counsellors, prenatal diagnosis as well as genetic counselling more generally served the interests of women or their families and had no eugenic intentions. The need to distantiate genetic counselling from the former eugenic praxis was apparently still strong. However, the preventive aspects of genetic counselling, and especially amniocentesis, were increasingly emphasised.[66] Prenatal diagnosis aimed at diagnosing as many genetic diseases as possible so that the parents could be guaranteed "not to have a child with the disease that there was an increased risk for", as two physicians wrote in a letter in 1973 to the Social Board, arguing that clinical genetics should be established within the healthcare system (see also the quote from Läkartidningen above).[67]

The preventive argument presented by the genetic counsellors had two aspects. One was that the prenatal diagnosis would prevent the birth of children with genetic diseases and thus relive suffering in families. The other was that healthy foetuses would not be aborted of fear of having an affected child. Prenatal diagnosis thus sought to abort diseased foetuses but to prevent the abortion of healthy foetuses.[68] In light of this, one could say that the decision to prevent births of individuals with undesirable traits was transferred to women and families, as it had been in the counselling activities of Böök, for example. The new technology, on an aggregate level, did however make it possible to prevent a large number of births of children with undesirable traits. As we shall see, geneticists acknowledged this possibility and discussed it in various ways.

10 Societal Dimensions of Prenatal Diagnosis

As mentioned earlier, abortion on specific indications was legal in Sweden. In 1963, in the wake of the thalidomide catastrophe, where children were born with malformation of limbs due to intake of drugs containing thalidomide by pregnant women, the law was extended to also include foetuses with a suspected deformity.[69] Around this time, calls were being made to confer the right to have an abortion without any special indication (free abortion): in view of this, a governmental investigation in

[65]Lindsten et al. 1975.

[66]See Munthe 1996, 37–50, for an ethical analysis of the preventive argument.

[67]Zetterström and Lindsten to the Social Board, 31 January 1973, SB 5E1:191, SNA.

[68]Lindsten 1973, 40.

[69]Lennerhed 2015.

1971 recommended a more liberal legalisation.[70] In 1974, a new law was adopted that conferred on women the right to terminate a pregnancy without any specific indication during its first 18 weeks. When prenatal diagnosis was introduced in the early 1970s, the debate about legalising abortion was in full swing and influenced the discussions about selective abortions of foetuses with genetic diseases or chromosomal aberrations. A common argument from many physicians and genetic counsellors was that if abortion was to be legal for all women and needed no special indication, there was no reason to hesitate to terminate a pregnancy if the foetuses had deformities or defects. As one physician said: "In a world and in a nation where one out of four pregnancies ends with abortion, I can't see why we should not aim to give young people the possibility to have a healthy child". Whenever a metabolic disease was suspected and the amniocentesis didn't show a clear result, the prevalent practice was to terminate the pregnancy: "rather one abortion too many than a sick child".[71]

The preceding discussion suggests that a key argument for prenatal diagnosis was that it could relieve families from having to bear the burden of a sick or disabled child. But the argument regarding the preventive capabilities of prenatal diagnosis was also applied at the societal level. It was argued that the ability to diagnose and terminate certain pregnancies could reduce healthcare costs. At times, a cost-benefit analysis was performed to compare the costs for prenatal diagnosis, including amniocentesis, with the costs for medical care and other kind of societal support. Even though this kind of analysis was complex and subject to possible criticisms, a group of medical geneticists in the mid-1970s concluded that clinical genetics was economically viable.[72] This opinion was stated in an expert report about the possibility of establishing clinical genetics within the Swedish healthcare system. It could thus be argued that such analyses were part of more overarching efforts to limit the costs of the healthcare system and that the same kind of rationale also applied to other health services.

However, this is not the only interpretation possible. The argument that disabled people were a burden to society and that their numbers should be reduced was like an echo from the past. It was brought up repeatedly in the eugenic discourse of the 1930s and 1940s and was heard in the post-war period too. For example, in one of the most frequently used academic textbooks in genetics, professor of genetics Arne Müntzing in the mid-1960s emphasised that disabled people and those with serious genetic illness imposed large costs on society. He argued that even if we were to take good care of such individuals, they were too many, and it was therefore in the interest of society to reduce their numbers, if possible.[73] Even though the situation for people with disabilities improved gradually during the post-war era (e.g., a new legislation was adopted in Sweden 1967 to secure their rights), the attitudes towards

[70]SOU 1971:58, *Rätten till abort*.

[71]Svennerholm 1973, 37–38.

[72]Lindsten (Ed.) 1976.

[73]Müntzing 1964, 359–360.

these individuals varied a lot, and their position was still weak. It could therefore be inferred that the argument about prevention represented persistent norms and attitudes about the value of the lives of the disabled and the possibilities they had in society. The widespread use of amniocentesis opened up new preventive possibilities, and at times the discussion became infused with the eugenic understanding that it would be best if children with severe genetic anomalies were never born.

Böök was one of those that argued that it was important to reduce the numbers of people with Down's syndrome. In a draft text sent to a World Health Organization (WHO) colleague, he suggested that testing for Down's syndrome could be justified with respect to a larger group of women—expectant mothers from 40 years of age:

> As 30–35 per cent of all cases of Down's syndrome originates from mothers of this age group [40 years of age] there is no doubt that whole scale screening of such pregnant mothers and selective abortions would significantly reduce the prevalence of such defective individuals in the population. If one was to worry over the costs of such operations it can be added that there is no doubt that these costs would be very much less than the costs for treatment and care of the defective children who otherwise would be born.[74]

Böök, in his discussion, embraced the possibility that amniocentesis provided for realising eugenic ideas that were widespread at an earlier stage. Thus, in mediating the use of the new technology, he contributed to framing it in terms of an indirect discourse of life and health—that it was desirable for both individuals and society to prevent Down's syndrome.

Although the argument about prevention was often used while discussing the possibilities of prenatal diagnosis, genetic counsellors emphasised that the decisions regarding having an amniocentesis and possibly terminating the pregnancy would be taken by the woman. There was increased understanding among the counsellors that these choices as well as other decisions related to the genetic counselling could be difficult and that they depended on emotional and psychological factors as well as the situation of the family.[75] Moreover, cultural and societal norms and values about what constituted a good life probably also influenced the decisions.

11 Ethical Dimensions of Prenatal Diagnosis

Whereas the debate about prenatal diagnosis in the early 1970s was characterised by a rather optimistic tone of geneticists about this possibility, the discussions during the latter part of the decade had a different orientation. The emphasis on preventing the birth of disabled children to thereby reduce the societal costs of health care for people with disabilities stirred up strong emotions and triggered

[74] Böök, "Prevention and Treatment of cytogenetic Disorders", IMG F6:29, p. 3, UUA.

[75] Lindsten et al. 1975. Cf. Lindsten (ed.) 1976, which argues that psychologists should be tied to the genetic counselling.

ethical debates about the consequences of prenatal diagnosis.[76] One fear was that selective abortions could reduce the intrinsic value of humans and change the attitudes towards the disabled. Another fear was that a more systematic screening could promote "an unacceptable eugenic view of humans".[77]

The ethics committee of the Swedish Society of Medicine recognised the problem but argued that prenatal diagnosis was here to stay. At the core of their arguments were information and individual autonomy. A general screening was not possible according to the law, for every kind of diagnosis had to be approved by the woman. Instead, the committee discussed the principle of prenatal diagnosis as it applied to individual cases. It emphasised that the woman should get all the information that was needed to take a decision. Further investigations were recommended if there was reason to suspect a serious disease or deformity of the foetus, but this too required the woman's consent. It was the physician's responsibility, according to the committee, to determine, based on his/her medical expertise, the type of medical examination that was warranted. The responsibility was thus divided in a manner Helén has suggested. That is, the woman was responsible for obtaining and handling information and taking a decision while considering the ethical and existential aspects that arose in this kind of situation. The genetic counsellor was responsible for providing the right information and for suggesting the relevant examinations.[78] Hence, the kind of responsibility that is at the core of the biological citizenship was clearly stated. The woman was expected to act in a manner that was best for herself, her family and even the expected child. The ethics committee assumed that no one would want to give birth to a child with severe birth defects that could cause great suffering. Decision-making was far more difficult when it came to cases in which the foetus was affected but would probably be able to lead "a reasonably decent life". However, it was the woman that had to make this determination. In this way the ethics committee confirmed a discourse based on an assumption about the quality of life that the woman had to respond to.

12 Genetic Expertise and the Ethos of Genetic Counselling

In summary, the period from the 1940s to the 1970s exhibits some distinct changes in how genetic counselling was formulated and undertaken by its practitioners. During this period genetic counselling was gradually transferred from the state authorities to the clinical context, situating the counselling activities within the realm of medical and genetic expertise. Constructing the role of the expert thus became an integrated aspect of the professionalisation of genetic counselling.

[76]Munthe 1996, 59–62, suggests that the economic motives were primarily used in order to establish clinical genetics.

[77]Svenska läkaresällskapets delegation for medicinsk etik, 1979.

[78]Helén 2004.

The importance of expertise is evident already in von Hofsten's activities. In all aspects of the eugenic efforts, von Hofsten stressed the importance of genetic expertise by a central authority to help individuals get the heredity information they needed. Böök brought medical aspects to bear on this issue. As a geneticist and psychiatrist, Böök strongly emphasised the need for professional medical geneticists, and he was active in the boundary work that defined the realms of medical genetics. Böök, like von Hofsten, never worked as a physician, and the genetic counselling Böök provided was done within the academic setting. It became the task of the next generation of medical geneticists, trained in the late 1950s like Lindsten and Gustavson, to establish medical genetics in the clinic and to develop genetic counselling in this setting. For this generation, genetic counselling needed to be integrated in the healthcare system and a trained physician needed to perform the counselling activities.

The professional ethos that evolved during this process emphasised increasingly the importance of individual autonomy. However, in von Hofsten's case, individual autonomy was clearly conditioned. Only individuals that he considered to be of good biological quality were granted a degree of autonomy when it came to decisions that involved abortion and sterilisation. Böök appears to have taken an ambivalent stand in relation to individual autonomy. On one hand, he protested against state decisions on the right to marry and argued for the individual right to assess risk figures in relation to hereditary decisions. On the other hand, as soon as the new diagnostic method of amniocenteses emerged, he embraced its future preventive possibilities and the prospects of eliminating Down's syndrome from the population. This indicates that the eugenic components of the discourse had not disappeared.

The generation of medical geneticists following Böök exhibited a strong ethos of individual autonomy in their counselling activities. They viewed their responsibility in terms of providing information and correct risk calculations and educating the counselee about the genetic risks. Information was often regarded as objective and neutral. However, the strong ethos of individual autonomy did not exclude a more prescriptive approach outside the counselling room. Here, the advantages of preventive genetic diagnostics and selective abortion could be emphasised over public spending for care of the disabled.

The prevailing discourse about life and health that contributed to the development of biological citizenship was influenced by various norms and values. In order to grasp what genetic counselling meant, we thus need to complement the professional ethos of individual autonomy and neutral information with an understanding of such norms and values.

The development of reproductive technologies created a novel situation for women and families. Amniocentesis and karyotyping provided a different type of information from the empirical risk figures. This enhanced both the opportunity and the responsibility of the individual and added new aspects to the development of biological citizenship. The responsibility involved ethical and existential decisions that could now be made with more reliable information to internalise and act responsibly towards.

As discussed earlier, the medical geneticists as experts argued that genetic counselling should relieve women and families of the burden of a disabled child. With the advent of the technology of amniocentesis, they could provide highly reliable information that formed the foundation for the women's decisions. They could also mediate the technology in a way that sought to relieve the women's guilt and anxiety. This led to the prospective goal, as evident from the discussion among geneticists, of "guaranteeing" a healthy child to families at risk.

At the same time, the availability of the amniocentesis technology along with changing attitudes towards abortion (with free abortion legislated in 1974) seems to have revived eugenic ideas in internal discussions among geneticists. These were manifested in cost-benefit arguments of the prospective socioeconomic outcomes of genetic counselling—that is, lower societal costs for the care of the diseased and disabled. Thus, when the technology and legislation became available, this seems to have brought on a eugenic rationality. These were two important aspects of the genetic counselling of the time: to both advocate a high degree of individual autonomy and simultaneously establish the socioeconomic cost savings that could be accomplished through genetic counselling. At the basis of the genetic counselling was a discourse of life and health that individuals reacted to and acted upon when they made their reproductive choices. Our analysis indicates that this form of biological citizenship has a long historical continuity.

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Counselling, Risk and Prevention in Human Genetic Early Diagnosis in the Federal Republic of Germany

Birgit Nemec and Gabriele Moser

Abstract In this paper we examine the relationship among human genetic counselling, early detection and the concepts of risk and prevention in the Federal Republic of Germany. For the period 1949–1989, concepts and practices in human genetic early diagnosis are examined, with a focus on the negotiation processes between science and larger societal fields. We are specifically interested in the early decades of this development, when a comprehensive establishment of human genetic counselling centres in medical clinics, at university departments and in private practices took place. How did nuclear research, the Contergan catastrophe and the rise of genetics to a leading science change anxieties and wishes addressed to the health of the unborn? What was the relation between counselling, research politics and society? How was a professionalization in the field of human genetic counselling shaped by specific sponsorships, for instance, through grants from welfare institutions and from an industry?

Important steps in these developments have already been researched in terms of their basic structures from the perspective of the history of medicine, with a special focus on actors and institutions. However, we still know very little about the role of actors *on the margins of* the narrower medical field, about media coverage and about the processes of political decision-making. In this context, the role of social organizations as a part of an emancipatory social movement, patient organizations of affected people and foundations with a variety of agendas regarding the justification and implementation of state prevention programmes is highly relevant. Our contribution focuses on the early phase of this development in order to reveal how the expansion of human genetic counselling was linked to a change in the concepts of risk and prevention. In the first step, we will look at the immediate post-war years and ask how eugenic and racial hygienic approaches transformed in discourses

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about health politics in the 1950s and 1960s. Special focus will be given to sterilization after 1945. In the second step, we will focus on the 1970s, the period in which human genetic counselling was established in the FRG. We will analyse this institutionalization process as a parallel development to the introduction of cytogenetic screening procedures in the FRG but also against the wider background of changing perceptions of the normal and the pathological and reproduction in the second half of the twentieth century. The aim of this paper is to reveal the complex interrelationship between notions of risk and prevention that shaped the history of human genetic counselling and early diagnosis in the FRG.

Keywords Marriage counselling • Genetic counselling • Human genetic early diagnosis • Federal Republic of Germany (FRG) • Risk • Prevention

1 Medical Counselling, Eugenics and Public Health Politics in the Late 1940s and the 1950s

In order to better understand counselling practice in matters of reproduction in the early post-war period, we will first summarize developments in the first half of the twentieth century up to 1945.[1] Since the medical reform movement of the mid-nineteenth century had turned its attention towards living and working conditions as connected to people's health, improvement of social and medical support for the poor had been demanded. When the Weimar welfare state introduced a diversified and wide-ranging system of group-related health-centred welfare during the 1920s, the political focus shifted from charity to a politics of the body and "welfare eugenics".[2] In this context medical counselling became an important means to strengthen the population and uplift the human race. In 1926 municipal marriage counselling centres (*Eheberatungsstellen*) were founded and offered medical examinations for physical and eugenic fitness for marriage and consultations regarding contraception and sexual education to engaged couples.

Because marriage was conceived as the only possible form of reproductive union until long after the end of the Second World War, marriage counselling and assessment of the "marriage suitability" of the fiancées were of central importance. An older definition concisely summarizes the administrative perspective on the functions of marriage: "Marriage is a form of sexual intercourse authorized by state and church for the production of healthy offspring".[3] The "physical fitness" demanded by the marriage counselling centre referred to venereal diseases and tuberculosis in the first place, two widespread infectious diseases at the time but also to inherited mental illness. The risk of having a mentally or physically ill

[1] Stöckel and Walter 2002; Ekberg 2007.

[2] Weindling 1997.

[3] Gastpar 1928, 1.

offspring was calculated on the basis of the fiancées' physical appearance and their family history. The assessment of the fiancées' "eugenic fitness" aimed at the detection of (hereditary) diseases that would make the offspring permanently dependent on medical assistance and social benefits.[4] If illnesses were detected in one of the fiancées or in his or her pedigree, the counsellor dissuaded the couple from getting married in order to prevent an "undesirable" offspring. It is important to note that the preventive perspective seeking to maintain health had shifted from a focus on the adult human being in the nineteenth century (occupational health and safety) to a focus on the pregnant woman as responsible for the health and well-being of the unborn at the beginning of the twentieth century (maternity services and welfare benefits). Finally, during the years of the Weimar Republic, the moment of conception was regarded as the earliest possible point in time to influence the quality of the offspring. The concept of marriage counselling originated from this hygienic and eugenic thinking, as social hygienist Felix Tietze (1883–1960) summarized in his assessment of the historical development of medical counselling in 1930.[5]

During the first half of the century, this sector was strongly shaped by publicly funded institutions. While municipal welfare institutions in larger cities had adopted marriage counselling by the 1920s and could also offer help in situations of social dilemma, people seeking advice in the countryside depended on the district medical officer as the state representative in health matters. Politically conservative medical professionals understood marriage counselling solely as a means to advise families to have as many children as possible, but they also warned about the uninhibited reproduction of "inferior life" ("Minderwertige") and appealed for "resolute action" ("entschlossenes Handeln") in a eugenic or racial hygienic spirit.[6]

The National Socialist state then subordinated the individual interests of the citizen entirely to a fostering of the "Volkskörper". The public health departments (*Gesundheitsämter*) newly created by the NS state, as well as their directors, were not only obliged to oversee the smooth organization of the forced sterilizations prescribed by the "Law for the Prevention of Hereditarily Diseased Offspring" from 14 July 1933[7] but also had to take care of marriage counselling according to the "Law for the Protection of the German Peoples' Hereditary Health (Law on marriage health)" (*Gesetz zum Schutz der Erbgesundheit des deutschen Volkes (Ehegesundheitsgesetz)*) from 18 October 1935. The latter prohibited marriage in cases "(a) of infectious disease, (b) of incapacitation, (c) of mental disorder, (d) of hereditary disease in one of the engaged". In contrast to the voluntary nature of marriage counselling in the Weimar Republic, the Nazi law made sure that the prohibition on marriage was observed by requiring the fiancées to present their

[4]Cf. Matz 1980 for marriage prohibition in case of poverty.

[5]Tietze 1930, 34.

[6]Scheumann 1932.

[7]Ley 2003; Westermann 2009.

"official marriage suitability certificate" (*Ehetauglichkeitszeugnis*); this sanctioned the entering of marriage by false pretences and declared marriages entered in this way as null and void.[8]

Public health departments were one central actor in the regulation of reproduction that outlasted the NS era.[9] The eugenic and racial hygienic hereditary health policy of the NS state was based on this unequal evaluation of its citizens' "dignity for reproduction": the phenotypically healthy, with a genealogical tree free from hereditary diseases, were encouraged to engage more in the upbringing of children, whereas chronically diseased people or people on welfare for a longer period of time were to be prevented from starting families, because their inability to perform was perceived as hereditary property. Independently of these categorizations, the prohibition of marriage also referred to the union of citizens of Jewish "descent" and those of "German blood". "The laws on the protection of blood and on marriage health" ("Blutschutz- und Ehegesundheitsgesetz") formed a unit, as the combined annotation in the 1936 edition proves;[10] they formed another component in the stigmatization and segregation of Jewish people in German society, an important precursor of expulsion, flight and mass murder.[11]

After the end of the Third Reich, which, among other atrocities, had forced 350,000 victims to undergo compulsory sterilization, physicians, medical specialists, medicinal officers and medical researchers found themselves in a judicial vacuum in terms of the legitimacy of eugenically motivated interventions in human reproduction. The allied forces now representing state power instead of the German Reich had to varying degrees be abolished, partly suspended or left in existence the laws and prescriptions enacted between 1933 and 1945. After the process of denazification, well-known names from the NS era found themselves back in important positions within the public health system, whose administration was now once again to be organized federally at the level of the Länder.

In this period of uncertainty and negotiation, public health departments were able to assert their role as experts in matters of reproductive medicine. As an alternative to ecclesiastical institutions, these institutions offered counselling and information centres that were driven by health insurance authorities. These can be seen as precursors to the genetic counselling that emerged slightly later.

The above-cited notion of "marriage" as *Fortpflanzungsgemeinschaft* (reproduction community) was still valid 30 years later, as evidenced by the statement of a Berlin doctor working in modern "anticonceptional counselling" at an information centre of the public health service. One should begin early, she emphasized, to point out to the young "that the substance of a healthy family life must always be the procreation and upbringing of the children. Our girls (*Mädel*) must learn again that their first and proper vocation is that of the mother, our young married couples must

[8]Flügge 1940, 108.

[9]Vossen 2001.

[10]Gütt 1936.

[11]Gruner 2008.

learn again that children warrant more happiness and a healthier and luckier family life than the possession of a motor-cycle or a television set".[12]

In northern Württemberg and Baden, the new Landtag had already decided on the "Law Nr. 347 on the Submission of a Health Certificate previous to Marriage"[13]—on 14 March 1949. The certificate, to be issued or at least confirmed by a public health officer, had to say whether one of the two fiancées was suffering from a communicable disease—just like the Weimar and the Nazi regulations, this referred primarily to tuberculosis and venereal diseases—or whether a mental disease or weakness was present. This health certificate had to be presented to the registrar before marriage; he could, however, marry the couple against the physician's reservations.

This regulation differed from that in the NS "Law for the Protection of the German People's Hereditary Health (Law on marriage health)" from 18 October 1935, according to which a lack of "marriage suitability" represented an impediment and even a bar to marriage. In the 1950s, such a "sharp intrusion into the personal freedom of the individual" did not seem to reflect contemporary legal opinion, but issuing marriage health certificates as well as acquitting public health officers from their medical confidentiality towards the engaged was still justified as thoroughly appropriate "in view of the happiness of the engaged and the children to be expected".[14]

With regard to their content, the same hygienic criteria that had been used by the NS state to classify marriage and reproduction between two people as undesirable for eugenic reasons were defined as an "impediment to marriage" in the post-war Germany of 1949. The well-known adolescent psychiatrist Werner Villinger (1887–1961) was one of the most important people in the formation of networks between prior race hygienists like Lothar Loeffler (1901–1983),[15] geneticists like Hans Nachtsheim (1890–1979)[16] and eugenicists and psychiatric colleagues such as Hermann Stutte (1909–1982).[17] In 1961, Nachtsheim, Villinger and his former Breslau University assistant doctor Helmut Ehrhardt (1914–1997) were consultants on the question of compensation for the victims of NS compulsory sterilization at the German Bundestag and unanimously recommended rejecting the compensation claims, since no injustice had been inflicted upon the victims—no wonder, as the consultants of 1961 had been among the scientific backers, if not among the actors of the NS policy[18]: Villinger had been an active assessor in a High Hereditary Health Court as well as consultant for the Aktion T4 for the murder of psychiatry

[12]Brandt 1958, 139.

[13]"Gesetz Nr. 347 über die Vorlegung eines Gesundheitszeugnisses vor der Eheschliessung" Wollenweber 1950, 482.

[14]Wollenweber 1950, 483.

[15]Loeffler 1955.

[16]Nachtsheim 1950, 1952.

[17]Rexroth 2003.

[18]Weingart 1992, 631–668; Klee 2001, 254–279; Klee 2003.

inmates; Nachtsheim had expanded his studies on the pathology of the inheritance of epilepsy from rabbits to children in asylums.[19]

Besides continuing his academic career, Nachtsheim, who, like Villinger, had supported the continuation of the eugenic health policy after the end of the war, hoped that a widespread popular education in genetics could help awaken "the will to eugenics" among the people.[20] For the practical implementation of his eugenic claims, Nachtsheim repeatedly advocated eugenically indicated sterilization as a means of birth control[21]—a field in which Villinger had also been active as a political consultant in the early post-war years from 1947 on.[22]

A commission of the health committee within the Länderrat in the US Zone chaired by Villinger had prepared a "Draft for a Law on Sterilization and Refertilization", which was also discussed at the different regional health advisory boards, for example, in Schleswig-Holstein in March 1948. Invoking its scientific basis, the justification for the necessity of this new legal regulation was introduced as follows: "By the current state of our knowledge on the heredity of diseases a law on sterilization is necessary"—only to continue in a surprising way: "The individual must be granted the right to sterilization in order to avoid serious hereditary disease".[23] Even if one suspects rhetorical tactics here, the formulation may anticipate an emerging shift in perspective in the debate on hereditary health in the Federal Republic: it was no longer society or the "Volkskörper" that were named as beneficiaries of the intervention; the individual himself/herself is introduced as a potential subject—even if identical interests are imputed to both the individual and the legislative body.

During the 1950s a vast new field of activity developed in the Federal Republic of Germany that was to shape the coming decades. The establishment of numerous new associations and foundations in the field of marriage and youth counselling, as well as the emergence of medical and professional support for parents of sick and disabled children like the *Heidelberg Rehabilitation Trust* (Stiftung Rehabilitation); the *Foundation for the Disabled Child* (Stiftung für das behinderte Kind); the *German Society for Marriage and Family* (Deutsche Arbeitsgemeinschaft für Jugend- und Eheberatung), later *Pro Familia*; and the *Campaign Sorrow Child* (Aktion Sorgenkind), is evidenced by a large range of brochures, book and journal publications. The *German Society for Marriage and Family*, later renamed *Pro Familia*, founded by Hans Harmsen in 1952, stands out as the most important among these. Beyond this, as we will show, general public acceptance of the medical management of pregnancies until the late 1980s was sustained by organizations that mediated between politics, science and industry. The fact that leading human geneticists supported the constitution of the most important organizations

[19]Nachtsheim 1941; Schwerin 2004; Weindling 2003; Holtkamp 2002.

[20]Nachtsheim 1959, 99.

[21]Nachtsheim 1950, 1952.

[22]Bundesjustizministerium 1958, 54.

[23]Landesgesundheitsbeirat 1948.

for marriage and youth counselling suggests the two following assumptions: first, new "apolitical" fields of activity had to be found in the silence surrounding the genetics of the old generation, active during the NS era, and second, the need for regulation in matters of reproduction in general, but also regarding the quality of offspring, was nevertheless clearly seen.

2 The Implementation and Extension of Human Genetic Early Diagnostics and Counselling

In the last section we looked at the immediate post-war years to show how eugenic and racial hygienic approaches transformed in discourses about health politics in the 1950s and 1960s. In this section we focus on the 1970s, a period known for the establishment of human genetic counselling in the FRG. While until the mid-1960s counselling was offered only upon request by anthropologist or medics with different specializations (mostly gynaecologists and paediatricians) in clinics, private practices and the newly founded departments for human genetics, the 1970s saw an exponential growth in institutionalized counselling centres: from two model centres in 1972—one in Marburg as a test centre for rural areas and one in Frankfurt as a model centre for urban areas[24]—to 37 in 1977 and 41 in 1982,[25] with a rapidly expanding capacity (e.g. in Erlangen, counselling cases quadrupled in the mid-1970s, from 70 in 1972 to 337 in 1977).[26] In 1968 Friedrich Vogel and Walter Fuhrmann, with *Genetic Family Counselling: A Guideline for Students and Doctors*, published the first counselling book in the German language; the book was translated into English in the following year and re-edited twice before the 1980s.[27] In 1973 human genetics was successfully implemented in the medical licensure act.

The expansion of counselling went hand in hand with the expansion of genetic diagnostic procedures. Amniocentesis, the most important diagnostic technique in this period, was introduced in 1970 to supplement non-invasive procedures such as ultrasound and examination of the mother's blood.[28] In its annual reports, the newly organized expert community celebrated the ever-growing number of procedures as a manifestation of a growing demand that they were able to satisfy: in 1970 six amniocentesis were conducted, in 1971 16, in 1972 49, and in 1977 as many as 2,648.[29] Both developments were linked to a growing volume of research in the field of prenatal early diagnosis, which is evidenced by an increase in funding, in

[24] Bundesministerium 1979.

[25] Stengel-Rutkowski 1973–1982.

[26] Koch 1977; Tünte 1979, 76–77.

[27] Vogel 1968, 1975 and 1982.

[28] Nippert 1991.

[29] Murken 1973–1982; Thomaschke 2014.

cytogenetic laboratories, in the exchange of research materials and in the registration and standardization of techniques and approaches.

From an international perspective—comparisons were mostly drawn with the Anglo-American world—the FRG was late in institutionalizing genetic counselling[30] and introducing prenatal diagnostics.[31] As is mentioned in almost every foreword of programmatic publications, geneticists had a difficult societal standing in the post-war decades because of personal, rhetorical and contentual/argumentative continuities and hybridizations with Nazi eugenicists and racial hygienicists, as discussed in the previous section. However, after a successful publicity campaign in parallel with a generational shift in the chairs for human genetics, the 1970s marked a successful period for geneticists, characterized by a constantly evolving area covering an array of techniques and approaches—a dispositif[32] of diagnostics and counselling.

Important moments for the local debate were the CIBA symposium "Man and his Future" in 1962, which fuelled public debates in the FRG with speculation about the future relevance of genetics,[33] and the WHO report "Genetic Counselling" of 1969, which helped geneticists to convince politicians of the necessity of the establishing of counselling centres. But more important for a public swing in opinion and a related readiness to release funding were local initiatives. The 1960 advice of the German Council of Science (whose members came from science, industry and politics) to establish a human genetic chair at every medical school[34] was an initial spark. Two symposia around 1970 ("Genetics and Society"/"Genetik und Gesellschaft", 1969 and "Genetics in the biological revolution"/"Genetik in der biologischen Revolution" 1970) followed as important steps.[35] The organizers of the 1969 symposium, Georg Gerhard Wendt (1921–1987, Marburg), Peter Emil Becker (1908–2000, Göttingen) and Friedrich Vogel (1925–2006, Heidelberg) invited other students of Nachtsheim, Lenz and von Verschuer to talk with actors from the media, public health, hygiene, legal medicine, paediatrics and several medical actors about the potential societal applications of genetics. Despite debates on the indistinct rejection of "old eugenics",[36] the symposium proved successful in suggesting a demand for genetic counselling and early diagnostics, and it finally led to negotiations with the ministry for health and families and the release of considerable state funding.[37]

As is clear in the published round-table discussions of the Marburg Symposium of 1969, the young generation of geneticists in the FRG developed an ambivalent

[30]Cottebrune 2015, 199.

[31]Schloot 1984, 11.

[32]Cf. Löwy 2014.

[33]Wendt 1970, Schloot 1984, 9.

[34]Empfehlungen des Wissenschaftsrates 1960.

[35]Schloot 1984, 9.

[36]Wendt 1970.

[37]Cottebrune 2008, 177.

and contested approach in the phase of community building that remained powerful throughout the 1970s.[38] As we will later show in more detail, economic reasoning was mixed with an individual medical, population genetic and preventive approach to heredity, regulation and the future of society. *Prevention* served as a powerful label that differentiated the new stance from that of older generations.[39] Leading actors in human genetics in the FRG were known for their inconsistencies, since they proclaimed a turn towards individual, non-directive counselling and the support of autonomous decisions but applied a radical and discriminating rhetoric that, according to the dominant discourse on disability,[40] defined anormality as a harm and as a cost factor. In the 1980s, in the context of the turn from a state-controlled, paternalistic, hierarchically directed stance to a more autonomous self in human genetics,[41] a reflection process and a more pluralistic and individualistic approach to counselling gained acceptance in spite of the idea that preventive measures were primary.[42]

As we shall argue in the following sections, the introduction of genetic counselling and diagnosis, which took the form of a powerful dispositif in the historical context of the FRG, can only be properly understood if we look at changing perceptions of risk and prevention. First we will show that the Contergan catastrophe led to a radical change in public and political awareness of risks to the unborn. This conceptual shift went hand in hand with the release of substantial amounts of money for research and preventive measures such as the implementation of codes of conduct for pregnant women, monitoring programmes, cytogenetic screening procedures, human genetic research, the implementation of an array of technical means of early detection and the institutionalization of genetic counselling. Second, we will examine the legal and economic changes that supported this development and the conceptual shift from concerns about the *Contergan children* to the definition of so-called risk children, by providing a new perspective on the liberalization of abortion law and the preventive turn in the welfare system, among others. Third, and last, we will briefly look at the growing critique of the early 1980s that challenged dominant notions of the normal, the anormal or defected, risk, and prevention.

We will argue that a form of economic reasoning served as a common thread through the decades under examination and that this was increasingly advertised using the label of "prevention". Further, we will suggest that the professionalization of human genetic counselling was clearly shaped by the activities and agenda of lobbying associations, mediating between science, politics, patient organizations and powerful industrial sectors.

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[38]Cf. the round-table discussions in Wendt 1970 and Schloot 1984.

[39]Klee 2001, 272 and 274.

[40]Bösl 2009; Schenk 2013.

[41]Cf. Waldschmitt 1996.

[42]Maasen 2011.

3 Contergan and the New Risks for the Unborn

When in 1961 it became public knowledge that taking the popular soporific Contergan during pregnancy caused severe foetal malformations, the human genetic landscape fundamentally changed. Although there was no direct link between the teratogenic effect of the active pharmaceutical ingredient thalidomide, which caused an exogene disorder in the embryonal development when taken between the 27th and 40th day of conception,[43] and questions of heredity, the Contergan catastrophe changed public awareness of risks to the unborn in the FRG and triggered the development of a close monitoring regime for pregnant women in order to prevent the birth of children with defects. This paradigmatic change fostered a readiness to expand screening methods and the development of codes of conduct for pregnant women, as well as the release of public funding for long-term research projects that were successful in their goal of establishing diagnosis and counselling centres at universities and in private practices, as well as testing and improving diagnostic procedures. What is more, the medial presence of the *Contergan children* (or *Conterganis*, as they later called themselves) and the fact that the pharmaceutical company Grünenthal was asked to make substantial (though not sufficient) compensation payments to the victims drew public attention to the burden and costs of disabled children. Human genetics profited from the climate of political remorse and public concerns.

The *Foundation for the Disabled Child* was founded by geneticists in 1966 in Marburg under the direct influence of the "thalidomide shock", supported by high-ranking politicians and the pharmaceutical industry as an "[i]nsitute to advance prevention and early diagnosis of child defects".[44] More research is yet to be done to examine this power structure in full detail, but archival research supports the assumption that with the Contergan catastrophe non-profit organizations, the Foundation of the Disabled Child being the most important, became highly important actors in the FRG. They competed in a newly established market for research funding, communication of knowledge, political lobbying, public influence and counselling, as I will describe further below.

Before the Contergan case became the epitome of risk and a lack of effective preventive measures (the West German drug law was changed in 1964), the risks of radiation and fallout were the most powerful keywords in releasing money for human genetics.[45] Similar to developments in the USA, where the experiences of Hiroshima and Nagasaki had by the mid-1950s led to the release of funding for research on mutagenicity,[46] the first international conference on human genetics in the FRG that took place in Barsinghausen in 1959 was organized and funded in the context of mutagenicity and radiation research. The German atomic commission's

[43]Friedrich 2005; Kirk 1999.

[44]Foundation for the Disabled Child, 1966–1988.

[45]Schwerin 2012; Schwerin 2015; Thomann 2005.

[46]Cottebrune 2008, 223–235.

working group IV/4 "radiation biology", e.g., was created in 1956 to guarantee the future use of atomic energy without sanitary and hereditary risks[47] yet provided an important impulse for the re-establishment of genetics in the FRG as it provided funding in the area of cytogenetics and became an important platform for networking for Vogel, Wendt and Becker—in cooperation with and supported by their mentors Nachtsheim and von Verschuer, to name the main profiteers.[48]

When the German Research Foundation (DFG), as a reaction to the Contergan catastrophe, sponsored two priority programmes—"Course of pregnancy and development of the child" (1964–1977) and "Prenatal diagnosis of genetic defects" (1973–1979)—between 1964 and 1980 with 25 m DM, a dispositive of surveillance and control was developed that built on earlier networks developed in the context of research on mutagenicity. This consisted of laboratories for clinical genetic and mutagenicity tests and centres for prenatal diagnosis and centres for genetic counselling. The first priority programme linked statistical data from gynaecological and paediatric clinics to calculate exogene risk factors for the "[c]ourse of pregnancy and development of the child". Maternal living conditions, eating and drinking habits, socio-economic factors, work load and medication, as well as environmental factors, vitamin supply and illnesses in 14,800 pregnancies were all statistically related to health data for newborns and toddlers. Recapitulatorily, the DFG defined the consumption of alcohol, cigarettes and medication during pregnancy as the main risk factors and recommended a "steady monitoring of pregnant women from the beginning to the end of pregnancy".[49] The second priority programme is built on the first. Exogene risks, such as long-term exposure to certain chemicals in the environment, were still an important argument for gaining funding.[50] In practice, however, the programme was explicitly determined to regulate reproduction and support the birth of a healthy offspring by advancing early genetic detection: "By the funding and development of diagnostic possibilities it will be possible in families with known hereditary diseases to get healthy offspring", stated the DFG.[51] The statistics presented in the annual reports confirm that exogene factors played a marginal role; in most cases hereditary risk factors led to prenatal diagnostics, with "advanced maternal age" as far and away from the main indication, followed by "previous child with Down's syndrome" and "chromosomal deviations".[52] In other words, Contergan, along with other potential dangers such as radiation, had changed the thinking about risks to the unborn and enhanced, though not directly linked, the expansion of human genetic counselling and early detection to prevent the birth of children with hereditary diseases.

[47]Klee 2001, 269; Koch 1979, 295.

[48]Vogel as quoted in Weisemann 1997, 118; Nippert 1991, 52.

[49]DFG 1977, 7–9; see also Koller 1983 for the publication of the study.

[50]DFG 1973, 78.

[51]DFG 1973, 78.

[52]Murken 1973–1982.

For a deeper understanding of the contemporary historical panorama, it is important to note that children in general and children and adults with disabilities in particular had a weak position in the early FRG.[53] Only in 1960 did compensation processes for disabled victims of the NS euthanasia programmes, a largely marginalized group, slowly begin. In 1963 a physician in Frankfurt administered a deathly syringe to a two-and-a-half-year-old boy with malformations caused by thalidomide at the behest of his mother. The father, who brought a charge against the pair for the killing of his son, was represented by a counsel from the *Interest Group Contergan Children* (Interessensgemeinschaft Contergan geschädigter Kinder), while the mother and the physician were represented by a legal counsel of the director of the research department of *Grünenthal* and co-inventor of Contergan.[54] If we consider the aloofness of political leaders with regard to recognition and support of the victims of Contergan, with the ministry Schwarzhaupt as the most prominent case, it is no surprise that transgressions such as the killing of a Contergan child did not cause public outrage—despite the fact that the renowned weekly journal *Die Zeit* reported on the case. Remember that the compromise agreement with Grünenthal had to be fought by the *Association of Parents of Physically Handicapped Children* (*Contergankinder-Hilfswerk e.V.*) and only in 1971, after tedious negotiation processes with a powerful pharmaceutical industry and medical professional association, was the *Foundation Aid Organisation for Disabled Children* (Stiftung Hilfswerk für behinderte Kinder) founded.[55] In the next section, we will show how this power constellation intertwined with the shift from monitoring to early detection and the foundation of an expert community.

4 From Monitoring to Early Detection: Dissemination Strategies of an Expert Community

The introduction of amniocentesis in the FRG in 1970 allowed a genetic diagnosis of the unborn and fundamentally changed human genetic counselling. Karl Knörr, head of the women's clinic in Ulm, and his wife Henriette Knörr-Gärtner were experts in cytogenetics with a focus on teratogenesis and mutagenicity when they learned about the new technique at the *6th World Congress on Gynaecology and Obstetrics* in New York in April 1970. In the same year, they were able to introduce amniocentesis in Ulm; the DFG priority programme "Prenatal diagnosis of genetic defects" allowed the nationwide introduction of the technology.[56] "I expect it an act of mercy towards the patients and their families, if these children (BN: children with severe congenital metabolic diseases) were not born but the pregnancy

[53]Rudloff 2002, 402–409.
[54]Die Zeit, 6.12.1963, 49.
[55]FA Koblenz 1971–1975.
[56]Nippert 1991, 52.

terminated in case the diagnosis on the fetus suggest it", advertised Horst Bickel with regard to the benefits of the priority programme in the popular weekly *Die Zeit* in 1977.[57]

The shift from monitoring to early detection induced by the introduction of amniocentesis had great impact on the counselling sector, as can be traced in guideline publications that appeared on the Western German book market and that served as professional sources of information and practical guidance in counselling situations, whether in counselling centres or in the medical practice of a gynaecologist or general practitioner.[58] Following international trends, ancestral charts, statistics, experimental genetics, geminology and anthropological elements from 1970 were complemented by prenatal screenings and diagnostics and medical genetics and from 1980 by Gen-Diagnostics, risk calculations by conditional probability and psychological and social aspects of genetic counselling. However, as the different foci of the guidelines suggest, they were at the same time important means to differentiate positions in a growing expert community.

Friedrich Vogel, by then chairman for anthropology and human genetics in Heidelberg, and paediatrician Walter Fuhrmann suggest a moderate, family-centred approach to counselling (as opposed to a population genetic approach); they wanted to "eliminate fears and insecurities" and "provide the basis for a responsible decision" through careful diagnosis and risk calculation that were to be communicated in two consecutive counselling meetings, ideally with both spouses. Despite the fact that they warn of value thinking in genetic counselling, they promise that the counselling will have a "eugenic" effect since it will be potent in reducing the rate of the "hereditary ill".[59] Vogel and Fuhrmann certainly set the standard for counselling practices in the FRG—we see them referenced in several other guidelines—but, as the case of Jan Murken, who in contrast to Vogel and Fuhrmann's critical perspective accepted selective abortion,[60] shows reception was selective. Geneticist Wilhelm Tünte, in contrast to Murken, problematized selective abortion and cost-benefit calculations,[61] rejected the aims of improvement as "eugenic"[62] and, like Vogel and Fuhrmann, proposed a family-centred programme that he called "social genetics".[63] Georg Gerhard Wendt's position, to name the most radical and controversial, was shaped by his theoretical background in anatomy and his practical work in Bethel, a clinic for the mentally disabled, where he worked under the aforementioned Werner Villinger. Similar to the neurologist and psychiatrist Peter Emil Becker, again much younger, Wendt promoted an economic, risk calculatory reasoning, a radical discrimination of people at risk of "genetic disabilities" and a

[57] Die Zeit 27th May 1977.

[58] Vogel 1968, 1975, 1982; Murken 1972; Wendt 1974; Tünte 1979.

[59] Vogel 1975, 6 and 117–118.

[60] Murken 1972, 9–12.

[61] Tünte 1979, 60.

[62] Tünte 1979, 2.

[63] Tünte 1978, Part 1, 1.

practice of genetic counselling designed to prevent "genetic degeneration" and "risk children" for the benefit of the "hereditary health" of "future generations".[64]

Despite these major contentual differences between the chairs of human genetics in the FRG, the process of community building led to the fast expansion of counselling and early detection and proved successful. The main reason for this was that the DFG priority programme "prenatal diagnosis of genetic defects", besides the introduction of amniocentesis as a routine procedure and the exchange of knowledge and research material, aimed at community building, exchange and collective knowledge production.[65] A strong and united expert community with targeted public relations was built up in order to guarantee public acceptance and political support beyond the DFG's initial funding. The success story of the dissemination of amniocentesis and of the expansion of counselling centres in the FRG was stressed in annual reports, visualized on the title pages and publicly advertised with a radical new rhetoric. While in 1969 Horst Bickel had objected to the "prevention" of disability by offensive genetic counselling,[66] in 1977, as speaker for the priority programme, together with other members, he advertised the costs saved by early detection through amniocentesis in relation to the cost of the care of patients with Down's syndrome in a popular scientific journal.[67] Wendt was invited to give an expert opinion on "genetic prevention" in the German Bundestag and disseminated his arguments for preventing the birth of disabled children for the benefit of future generations—an argument that is without doubt deeply rooted in eugenic thinking—into political discourse.[68]

The aggressive rhetorics proved successful—the funding of a constantly evolving dispositif of diagnostics and counselling was in many cases taken over by the federal states. But to set up and maintain the small-scale practical dimensions of this array of techniques and approaches, lobbying institutions were needed. Heiko Stoff has pointed to the important role of lobbying institutions in the FRG in the context of industrial food production, where they acted as mediators between a generally reluctant state, scientific experts, industries, patient and self-help organizations and the public. They were involved in political decision-making by a modulation of interests in expert groups, in the commissions of the DFG, in the assembly halls of ministries and at round tables.[69] Similarly, lobbying, non-profit and self-help institutions in the field of "prevention" of and "support" for children with disabilities, such as *The Association for the Disabled Child*, *The Heidelberg Rehabilitation Trust*, *The Campaign Sorrow Child* and the *Federal Association Lebenshilfe*, to name the most important, with different foci, acted as mediators between state, industry, science and the public in transforming abstract research

[64]Wendt 1974, 120–133; Cf. Becker 1973; Wendt 1974.

[65]Cf. Murken 1973–1982.

[66]Wendt 1970.

[67]Bickel 1977, 283.

[68]Bundestagsdrucksache 1975, 787–796.

[69]Stoff 2015.

data into everyday practices. In the case of *The Association for the Disabled Child*, laboratories were equipped; staff were employed; genetic counselling was provided; leaflets and mailshots were sent out to bring those people to human genetic counselling centres that were hard to reach yet thought to be in need of counselling, namely, lower social classes; guidelines for genetic counselling institutions and codes of conduct for pregnancy were published; further training for medics, teachers and social workers was provided; counselling and diagnostic centres were founded, maintained and sponsored; and television advertisements were produced to raise public and media awareness; special-needs schools were funded, along with the implementation, continuation and development of the means of prevention and early detection in clinics, institutes and organizations all over the FRG.[70] To sum up, the associations had an enormous influence on the cultural climate in the FRG in general as well as on the routinization of notions of norm, prevention and risk in everyday life.

As we will show in the next section, the strong support of the DFG for advancing the diagnostics of "prenatal damages"[71] cannot be abstracted from an expected change in abortion law. "The examination (BN: amniocentesis) only makes sense if a reform of abortion law considers genetic charges of the embryo", stated Karl Knörr in 1972.[72] Traute Schröder-Kurth, a cytogeneticist at Vogel's department in Heidelberg, said it was "unethical" to provide prenatal diagnostics without being able to offer abortion to women.[73] Under the influence of ongoing societal debates, the DFG invited geneticists from the USA, Poland, the UK and Denmark to evaluate possible problematics linked to prenatal diagnostics in round-table discussions before approving the programme in 1972[74]—a decision that had an important influence on debates on the legalization of abortion by "medical indication".

5 A Changed Approach to Abortion and Prevention

The reformation of the "§218 StGB" in the 1970s—in the sense of a liberalization of abortion law in the FRG—is primarily thought to be a sociopolitical development: as a call for self-determination over the female body and one's own life in the moment, Schering brought the first contraceptive pill "Anovlar" to the market, and the societal influence of the church was diminishing. As a tedious political and legal negotiation process, it was shaped by differences between the SPD and the FDP on the one hand and CDU/CSU fractions on the other, with the result being the 15th amendment to criminal law ("Strafrechtsänderungsgesetz") on 18 May 1976, which

[70] BA Koblenz, 1973, B129/28123; Hartung 1986.

[71] DFG 1973, 78.

[72] Knörr in Murken 1972, 57.

[73] As cited in Osten 2012, 164.

[74] DFG 1973, 78; Nippert 1991, 56–7; Schloot 1984, 11.

contained a widely construable "indication regulation". We know that the main actors were political parties, the church and most of all the dedicated public, especially supporters of the women's movement and advocates of the so-called sexual revolution, the protagonists of the newspaper Stern's reportage "We have aborted" of the 6 June 1971. However, on the basis of the sources we examined, the prehistory of the so-called liberalization of abortion was a parallel development or—even a consequence—of developments in the medical field, especially in human genetics.

The decision to introduce "indication regulation" was without doubt a triumph for the medical profession. The church refused abortion but under certain conditions silently tolerated "medical" indications;[75] liberal circles around the women's movement demanded an unconditional legalization of abortion;[76] the SPD and FDP fractions contributed a (almost successful) proposal for the legalization of abortion within a defined time limit, as an abortion on demand ("Fristenlösung").[77] The CDU/CSU fraction, counselled and in close connection with leading medical circles,[78] de-legalized the German Bundestag's decision for abortion on demand in 1975 and succeeded with an indication regulation[79] that was—with minor variations—de facto a legal confirmation of medical practice. Only a "medical" indication, validated by an expert report, could justify an abortion. The interpretational sovereignty over the value of the unborn remained in the medical realm.

From abortion statistics of the years prior to the German Bundestag's decision, we learn that abortion practice, controlled by medics, provided the reform debates with an important brisance. Approvals of abortions by "medical indication" in the years 1968–1975 rose from 2,826 to 17,814.[80] The imbalance between practice and law was evident to the public, here fostering a literal abortion boom;[81] doctors sought legal coverage and control by pushing back the high number of illegal abortions. After 1945, abortions for "medical reasons" went unpunished[82] but differed by occupation zones, and then by county, and were in general subject to the belief of the approving doctor. Although medics felt the need and issued guidelines to define "medical" indications, the grey area continued into the 1960s and stimulated the new reform debates.[83]

By around 1970, the majority of medics supported the abortion of a fetus with a "severe genetic disease" or "malformation"[84] and, apart from some exceptions,

[75]Commission of the protestant church 1971, as quoted by Krauss 1972, 73; Gante 1991, 84–5.
[76]Achtelik 2016, 15–25.
[77]Gante 1991; Behren 2004.
[78]Bundestagsdrucksache 1974a.
[79]Behren 2004.
[80]Bundestagsdrucksache 1974b, 15.
[81]Behren 2004, 414; Gante 1991, 105–109.
[82]Behren 2004, chapter 1.2–2.3.
[83]Naujoks 1954; Müller 1964; Ahrens 1972.
[84]Kraus 1972, 74.

regarded a "medical" indication ("therapeutic", "in children", "eugenic", "embryopathic" and "genetic" were in general used synonymously) as the only legitimate kind. Arguments for an indication regulation were reinforced by collaborations with legal experts,[85] yet it is remarkable how abortion on demand was seen as a threat to the established, and often sexist, doctor–patient power relationship. Emotional debates in the leading periodical *Deutsches Ärzteblatt* reveal that medics feared the "subjugation" of the doctor under the "pure will" of the pregnant woman,[86] as well as "impending anarchy" and the "vociferous propaganda that, under the motto of a women's right over her body, practically supports the expectant mother in aborting, just as she wants".[87] The most important point of reference for local medical debates was the Oslo "Declaration of the World medics congress for therapeutical abortion" of 1970. In 1973, at the peak of the reform debates, the *German Doctors Day* (Deutscher Ärztetag), the biggest gathering of medics in the FRG, referred to the Oslo declaration in its resolution "Reform of the §218: Rejection of Abortion on Demand". Abortion by "medical indication" was defined here as a "therapeutical" act that considered "dangers to the life of the pregnant or the danger of a serious negative impact on her wellbeing". The important change was that this included dangers to "mental well-being", either caused by a "forced pregnancy" (rape) or by "the founded fear of a severely impaired child".[88]

Earlier in 1973 the German Medical Association went as far as to contest "social emergencies" or rape as valid indications but named "the severe psychic burden that grows" from "the fear of a pregnant woman to deliver a severely unhealthy child, unfit for the challenges of social life" as a prime example of a legitimate indication for abortion. As it was added, in the same moment, the DFG announced the approval of its priority programme, "new diagnostic procures promise an early diagnose of an impairment of the fruit of the womb".[89] To sum up, while at first sight the broad definition of "medical indication", with an emphasis on a possibly disabled unborn child, as the favoured solution for medics is striking, a closer look at debates within the medical expert community reveals that the reintroduction of a "eugenic" indication—introduced in 1935 in NS Germany, abolished by the allies and discussed since the foundation of the FRG in the late 1940s—[90]was apparently at the basis of an abortion practice that in the end reinforced the decision to settle on an "indication solution" in 1976.

At the turn of the 1960s, a profound change in the medical landscape of the FRG in the form of a preventive turn provided the link between abortion, healthcare and genetic counselling with a new dynamic. "Health prevention through early detection of diseases", including human genetic early detection, became a new maxim. It

[85]Murken 1972.

[86]DÄBL 1973a, 2797.

[87]DÄBL, 1970, 2690.

[88]DÄBL, 1973b, 2971.

[89]DÄBL 1973a, 2797.

[90]Gante 1991, 59.

was legally manifested in 1970 in the second health insurance amendment law, pronounced at the German Doctors Day in 1971 and published in "Programmed Early Detection of Diseases" by the executive secretary of the German Medical Association Josef Stockhausen, an important advocate of the indication regulation who represented Germany in Oslo and was one actor who coined the term "risk children".[91] In 1975 the reformulation of maternity guidelines explicitly defined genetic counselling as a preventive procedure that was to be covered by medical health insurance services.[92] The label "prevention" then provided even more justification for counselling as a means to prevent the birth of children with genetic "defects", as this statement by Wendt, at this time head of the *Foundation for the Disabled Child*, in a monthly protestant magazine shows:

> All estimations show that genetic counselling as the corner stone of preventive medicine cannot only take human burden from some family but set free considerably sums that are, until now, spent for our disabled. From this consideration arises the suggestion that from now on we should use 5–10% of the money that is at the moment in the federal republic used for the care of the disabled, for the prevention of disability. The necessary care for the disabled will worsen only temporarily. Because if we reduce the increase of the disabled the care for the existing disabled can become more comprehensive by constant input.
>
> Genetic counselling does not seek eugenics, it does not want to improve the hereditary factors of future generations. Genetic counselling looks after the health of the children that are conceived and born today. It is at the moment the main medical duty of preventive medicine.[93]

Wendt's position gives a good impression of the sociopolitical reasoning of this period. Genetic counselling, as one means to reduce the number of people with disabilities, was added to the insurance catalogue just before, in 1977, a cost-dampening law was created to further reduce ever-rising welfare costs and as a "flanking measure" of the §218 reform, where counselling was in turn added as a compulsory part of a legal abortion.[94]

To sum up, in the 1970s an economic reasoning that implied discrimination against unborns that were presumably "ill", "disabled", "genetically charged" or in general "abnormal" with regard to the cost of expected medical and social care was supported by important legal and economic developments. These developments promoted the expansion of genetic counselling and early detection and—in the unwelcome case of emergency—the possibility of late abortion by medical indication. The link between regulation of reproduction and cost calculations that understood society as an economic community was everything but a new topic. What was new was the fact that preventive measures that had belonged to the realm of the state-ruled public health sector were now the province of licenced physicians—opening up genetic counselling and early detection to the free medical market and its players.

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[91] Stockhausen 1971, 68.

[92] Bundesanzeiger 1975.

[93] Wendt 1976, 252.

[94] Bundesgesetzblatt 1975 and 1976.

6 Challenges to Notions of Risk and Prevention

In the 1980s, the dominant view on the unborn of actors in human genetics was increasingly challenged. It became a controversial question whether the *Foundation for the Disabled Child's* principle "prevent disability" had a problematic impact on developments regarding pregnancy, practices of genetic counselling, and diagnostics, and the acceptance of otherness in FRG society. Geneticist Irmgard Nippert noted in a critical evaluation of her experiences in genetic counselling that about 9–10 % of amniocentesis were conducted "without a detectable increased risk" and she criticized "the fear of having a mongoloid child" and "the wish for a predictable and healthy child" of many parents, especially the well educated.[95] Especially in the context of the disability rights movement that was shaped by, among others, the work of Udo Sierck and Nati Radtke, a reflexivity was induced that changed public opinion as well as scientific discourse.[96] In 1990 the ministry counted 40 centres for genetic counselling and 34 disability associations, of which five had a general focus and 29 a specific focus[97]—just to give an idea of the changed power structure on an institutional level.

Other critical voices came from the protestant church, which among others challenged the funding practice of *Campaign Sorrow Child*—which decided to fund genetic counselling from 1979 with 1.5 m DM annually and founded an institution to fund genetic counselling in the FRG[98]—and also the women's movement. After a silent phase that followed the lost reform debates of the §218[99] in 1985, two prominent congresses united as "Women against Gene and Reproduction Technologies". The protagonists criticized the politics of reproduction in the FRG and challenged, in particular, medical-genetic and pharmaceutical practice, notions of "illness", "non-directiveness", "voluntariness" and "prevention".[100] While in the early 1970s feminists did not much care about the problematic aspects of "eugenic indications" but demanded the possibility of abortion for whatever reason, in 1985 they demanded an elimination of §218 "also because of the eugenic indication".[101] The majority of participants in a special symposium asked for a termination of genetic counselling, diagnostics and public research funding in this area.[102] This was not realized, but the claim definitely gives an impression of the direction of public debates.

Within medical discourse, the first signs of a move towards more inclusive and participatory approaches to the individual in genetic counselling were seen at a

[95]Nippert 1984, 114–115.
[96]Wunder 1982; Sierck 1984.
[97]Nippert 1990.
[98]Cottebrune 2012, 193.
[99]Achtelik 2016.
[100]Kaupen-Haas 1985b, 68.
[101]Groth 1985, 88.
[102]Bradish 1989, 9.

forum in Bremen in 1981 on the "Possibilities and Limits of Human Genetics", held in cooperation with actors from the disability rights movement.[103] *Schroeder-Kurth*, in the climate of several critical evaluations of genetic counselling practices,[104] notes in a *leading gynaecological periodical* that "we kill ill foetuses to prevent their 'illness'" and problematizes the "automatisms of prenatal diagnostics and selective abortion" and the established "duty of the non-disabled child".[105] Among others she targets the question of a right to "genetic autonomy" and the right not to know. At the same time, the German Bundestags created a commission that developed guidelines for genetic counselling and diagnostics based on current critical evaluations.[106] The final report of the priority programme, "prenatal diagnosis of genetic defects", warned of the "misuse of prenatal diagnosis as an unreflected routine method" and advocated the acceptance of "variability" as a factor of social stabilization (DFG 1982). However, since in information material of the *Foundation for the Disabled Child*, slogans like "Our child shall be healthy" or "The main point is, that it [BN: the baby] is healthy" were distributed well into the 1990s, it is questionable when and how these principles were implemented in practice.

7 Conclusion

In this paper we have examined counselling, risk and prevention in human genetic early diagnosis in the Federal Republic of Germany, with a focus on the 1950s to the 1970s. In the first part we looked at the post-war period to show how eugenic and racial hygienic approaches transformed in discourses about public health politics. In the second part, we looked at the establishment of human genetic counselling and diagnosis in the 1970s against the background of changing perceptions of reproduction, the normal and the pathological, risk and prevention. In both parts we were especially interested in the role of legal, technical and institutional settings as a context for processes of exchange between actors in- and outside the narrower medical field, i.e. between human geneticists, self-help associations, patient organizations, social organizations, larger social reform movements, the media and political decision-making.

In the FRG PKU screening was the benchmark for success in the treatment of human genetic diseases and the implementation of genetic counselling and screening measures in the FRG. However, only close cooperation among human geneticists, paediatricians, counselling institutions and self-help associations on a local and international level made this quick nationwide introduction possible.[107]

[103] Baitsch 1997.

[104] Reif 1986; Reif 1989, Fäßler-Trost 1989.

[105] Schroeder-Kurth 1989.

[106] Catenhusen 1987, 151.

[107] Osten 2012.

Similarly, in the more controversial field of cytogenetics, a vast new field of activity for experts and people concerned with parent and non-profit organizations, as powerful players between politics, scientists and industry, developed in the 1950s. The involvement of human geneticists in the constitution of the most important associations and foundations in the field of marriage and youth counselling, as well as the emergence of medical and professional support for parents of sick and disabled children, led to the assumptions that new, "apolitical", fields of activity had to be found to delineate from the genetics of the old generation that had been active during the NS era. This also meant that the need for regulation in matters of reproduction was clearly visible. The routinization of early detection and preventive acts in prenatal care was supported by historic caesuras such as the Contergan catastrophe and changes in abortion and health insurance law, which were put into practice through negotiations between non-profit organizations and charitable and self-help associations, thus challenging the interpretational sovereignty of science, politics and industry on notions of norm, illness, risk and prevention.

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"The Happiness of the Individual Is of Primary Importance": Genetic Counselling in the GDR

Susanne Doetz

Abstract Against the background of the National Socialist past and the Lysenko doctrine, human genetic research in the German Democratic Republic (GDR) was practiced on a limited scale up until the late 1960s. The doctrine of Lysenko, which had been transferred from the Soviet Union, proclaimed the inheritance of acquired characteristics as a fact and denounced genetics as pseudoscience. Nevertheless, East German scientists practiced human genetic research in some niches, and the concept of hereditary diseases existed in spite of Lysenko.

The paper presents the different political and scientific developments that led to the establishment of a genetic counselling family service in the GDR in the 1970s against the backdrop of the Cold War. It shows how genetic counselling was embedded in the socialist health-care system and integrated into family planning. In addition, I analyze how it was put into practice. Limiting factors of genetic counselling in the GDR were not ideological reasons but the lack of resources. Overall genetic counselling was characterized by an ambiguous situation: on the one hand, genetic counselling should be strictly voluntary, and on the other hand, its goal was to prevent the birth of disabled children and to foster the birth of "healthy" ones.

Keywords Genetic counselling • GDR • Prenatal diagnosis • Disability • Lysenko • Human genetics

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1 Introduction

In regard to human genetics in the countries of the Soviet sphere of influence, the Soviet agronomist Trofim Denisovich Lysenko (1898–1976) is more likely to come to mind than the establishment of genetic counselling. Lysenko proclaimed the inheritance of acquired characteristics as a fact and denounced so-called formal genetics, like the germ plasm theory of August Weismann (1834–1914), as pseudoscience.[1] Moreover, the entanglement of German human genetics with the National Socialist hereditary health and race policies discredited human genetics.[2] Further, the German Democratic Republic (GDR) had a decidedly anti-fascist self-image.[3] Considering these difficult conditions, how could human genetics become established in the GDR? Furthermore, how could the concept of genetic hereditability be reconciled with a Marxist perspective that holds man as an "ensemble of social conditions?"[4]

The establishment of human genetics as a science was the product of a transnational network of researchers, who collaborated across national and continental borders. Hence, other important questions arise that highlight the impact of the Cold War on the development of human genetics in the GDR: Given the travel restrictions GDR scientists had to face, how could they participate in this process? Moreover, how did human genetic knowledge and techniques cross the Iron Curtain? To answer these questions, I first want to show how—in spite of Lysenko—East German scientists performed human genetics in some niches. Moreover, I will demonstrate that the concept of hereditary diseases existed despite of Lysenko's ideas. After outlining the different political and scientific developments that led to the establishment of a genetic counselling family service in the GDR in the 1970s, I will explore how counselling was performed in a socialist state, as the SED (Socialist Unity Party of Germany) proclaimed that the interests of a socialist society were identical with the interests of single individuals living in that society.[5] What were the ramifications of those paradigms for the people who sought genetic advice and for the possibility to establish patient advocacy groups? To what extent was criticism of the official position possible? However, genetic counselling cannot be described as a simple top-down process. Embedded in GDR family politics and preventive health care, it was shaped by different human actors like human geneticists, physicians, social scientists, health policymakers, laboratory staff, and people who sought genetic advice. Considering the role that scientific objects played in the process of knowledge-making in general and in practicing

[1] For Lysenko's ideas, see, inter alia, Roll-Hansen 2005, Krementsov 1997, and Medwedjew 1969.

[2] For examples of the reciprocal relationship between human geneticists and the Nazi state, see Weiss 2010 and Schmuhl 2005.

[3] Preamble of the GDR constitution, Gesetzblatt, 1968.

[4] Marx 1969, 6.

[5] Dietl 1984, 87.

genetic counselling in particular, genetic counselling was also shaped by human resources such as amniotic cells or lymphocytes and by culture media, microscopes, and adequate space for laboratories.[6]

Unlike in the USA and other countries, the profession "genetic counsellor" did not exist in the GDR. Instead, counselling was performed by physicians or biologists with a special training in human genetics, even though they were mostly self-taught at first. This means that human genetics and genetic counselling were closely connected in the GDR. I will therefore occasionally skip between the two fields in this paper.

2 Human Genetics in the Aftermath of World War II

After the end of the World War II, the institutional basis for human genetics was missing: None of the former chairs or university institutes for racial hygiene or heredity and race biology, which were installed during National Socialism, continued their work. While in West Germany (FRG) human genetic research, in a broader sense, found shelter in several university institutes for anthropology, in East Germany, only the Institute for Anthropology and Ethnology in Jena continued to exist.[7] Moreover, the complete revocation of the "Law for the Prevention of Hereditarily Diseased Offspring" (*Gesetz zur Verhütung erbkranken Nachwuchses*) in the Soviet occupation zone marked a clear break with Nazi sterilization politics.[8] Even though this did not mean that eugenic ideas completely disappeared, as I will show later.

Besides these influences of the Nazi past, another important reason for the muted development of human genetics in the GDR was the doctrine of Lysenko—or rather Michurin biology, as the term was called in the GDR—which questioned the basic concepts of genetics in general. Starting as a local conflict between two scientific groups in the Soviet Union, the historian Nikolai Krementsov demonstrated that in the context of an intensifying Cold War, the debate on Lysenko's ideas was transformed into a huge ideological campaign. Soviet science was now considered incompatible with Western science. Subsequently, in 1948, the Soviet government

[6]Examples for the role of objects in knowledge-making are Rheinberger 1997 and Knorr Cetina 1999.

[7]Cottebrune 2012, 30–33; Hoßfeld 2005, 213–214. The Institute of Anthropology was reestablished at the Humboldt-University in East Berlin in 1955. See UAHU, Rektorat, Nr. 326a, foil 149.

[8]The situation in the other zones of occupation was confusing. The law—in most of its parts—was not applied anymore, but it was, with the exception of Bavaria, not completely revoked. The victims of this law, in both East and West Germany, were not considered "victims of Fascism" ("Opfer des Faschismus") and "victims of National Socialist persecution" ("Opfer der nationalsozialistischen Verfolgung"), respectively. See Doetz 2011, 234–235, 246–249.

officially condemned genetics as unscientific, and, therefore, Lysenko's doctrine was spread in the Soviet zone of influence.[9]

In the GDR, the influence of Lysenkoism on human genetics was significant, yet it was never totalizing. The authorities disseminated Lysenko's ideas, and several scientists and propagandists accepted and promoted them in schoolbooks, lectures, newspaper articles, and scientific publications. In 1950 alone, the SED party organ *Neues Deutschland* published more than 60 articles referring positively to Lysenko or the Russian botanist Ivan Vladimirovich Michurin (1855–1935).[10] However, these articles remained quite superficial. Their authors mostly did not convey a deeper insight into the ideas of Lysenko or the so-called teachings of Michurin. Rather, they used the names of Lysenko and Michurin affirmatively—connecting them to change, progress, and even peace while the names of the so-called formal geneticist like Gregor Johann Mendel (1822–1884), August Weismann, and Thomas Hunt Morgan (1866–1945) stood for idealism and a reactionary stance.[11] Thus, far from any debate on the terms of heredity, referring to Lysenko and Michurin was a way to prove that one had the correct political orientation.

Nevertheless, some East German universities continued to teach classical genetics as well. Notably, the SED was reluctant to insist on the adherence to Lysenkoism in order to not loose highly qualified specialists, at a time when it was still possible for them to leave the country via the open border between East and West Berlin—the Berlin Wall was built in 1961. One scientist, who did not leave, but used the possibility to defend the interests of genetics throughout the GDR, was the geneticist Hans Stubbe (1902–1989), director of the Institute for Cultivated Plants Research (*Institut für Kulturpflanzenforschung*) in Gatersleben which belonged to the Academy of Science (*Akademie der Wissenschaften*). Stubbe and his colleagues worked to disprove Lysenko's theories through their own experiments.[12] Indeed, as early as 1951, Stubbe had cautiously but clearly expressed criticism of Lysenko's ideas at a meeting with agronomists at the Central Committee (*Zentralkomitee*) of the SED.[13] When confronted with those persistent newspaper articles against classical genetics in the early 1950s, Stubbe countered by threatening to leave the GDR to accept an appointment in West Germany. Hereupon, Walter Ulbricht (1893–1973), first secretary of the SED Central Committee and the one who

[9]Krementsov 1997, 105–131 and 143–183; Krementsov 2002, 179–202. For the situation in Poland and the ČSSR, see DeJong-Lambert 2012, 499–525 and Simunek, Hossfeld 2013, 84–88.

[10]The newspaper *Neues Deutschland* (ND) is completely digitalized from 1946 to 1990: http://zefys.staatsbibliothek-berlin.de/ddr-presse/volltextsuche/, last access: 17 July 2016. The chosen keywords were "Lyssenko" in different spellings and "Mitschurin." Literature on Lysenkoism in the GDR is Höxtermann 2000, 273–300; Fäßler 2001, 177–194; Hagemann 2002, 320–324.

[11]Examples are ND, 23 June 1949, 3; ND, 4 January 1950,3; ND, 7 June 1950, 3; ND, 15 September 1950, 3; ND, 16 September 1950,5; ND, 4 October 1950,2; ND, 29 December 1950,4.

[12]Höxtermann 2000, 273–300; Fäßler 2001, 177–194; Hagemann, 2002, 320–324. A biography of Stubbe is written by Käding 1999.

[13]Stubbe 1952, 96–112.

determined East German governmental policies, aligned himself with Stubbe so that he was able to continue his work untroubled.[14] As this move suggests, human capital in the form of specialists was more important to the political leaders than dogmatizing science.[15]

But, what was the situation of human genetics in the GDR during the postwar years? GDR genetic counselling pioneer Herbert Bach (1926–1996) described Lysenko's influence as follows:

> The fear that the party will interfere of its own volition again and again in scientific disciplines, which it considers ideological relevant, almost certainly had the effect that some people preferred a less risky discipline than genetics and made it difficult for politicians to support such a burdened field. This, in my opinion, was one of the reasons, why the establishment of human genetics in the GDR proceeded only slowly, and why, besides a few physicians, mainly biologists got involved. While physicians did not have problems finding work in their traditional disciplines, professional opportunities were clearly worse for biologists.[16]

The negative connotations of human genetics in the GDR also meant that research in the field received little political and financial support, unlike in West Germany or the USA, where it benefited from concerns about the effects of radiation on human genetic endowment.[17]

Nevertheless, some scientists practiced human genetics in some niches like the Biological Institute in Halle. The institute belonged to the medical faculty and its director was the biologist Paula Hertwig (1889–1983). Her research interests concerned the influence of radiation on the genotype. During the 1930 and 1940s, she performed large-scale experiments with radiation. She used mice as experimental animals in order to proof the validity of the mutagenicity experiments, originally performed on drosophila, for mammals, and thus—so the assumed consequence—for human beings. She continued her work with her mutant mice strains after the end of World War II, when she was appointed director of the Biological Institute in Halle in 1946. There, she taught genetics and also human genetics to medical students and supervised dissertation projects that used her

[14]Käding 1999, 112–113.

[15]Ash, 1997, 13–14.

[16]Bach 1997, 87. In the German original: "Die nicht grundlose Befürchtung, daß sich die Partei in ihrer absoluten Machtvollkommenheit in Wissenschaftsdisziplinen, die sie für ideologisch relevant hält, immer wieder einmischt, hat mit Sicherheit dazu geführt, daß manch einer ein risikoärmeres Fach als die Genetik bevorzugt hat, und sich Politiker schwer taten, sich für ein derartig vorbelastetes Gebiet einzusetzen. Dies war m.E. einer der Gründe, warum in der DDR die Etablierung der Humangenetik nur langsam in Gang kam und sich neben wenigen Ärzten vor allem Biologen um das Fach bemüht haben. Ärzte hatten auf ihren ursprünglichen Fachgebieten keine Beschäftigungsprobleme, während die beruflichen Chancen der Biologen deutlich schlechter waren." For Herbert Bach, see also Pittelkow 2015 and the chapter on Herbert Bach by Jörg Pittelkow in this volume.

[17]Kevles 1986, 229–230; Kröner 1997, 69–82; Schwerin, von 2012, 88–93. Schwerin, von 2015.

mutant mice strains in order to explore genetic diseases of the eyes.[18] Hertwig's research on the danger of radiation also drew the interest of the Office for Nuclear Research and Technology (*Amt für Kernforschung und Kerntechnik*) when, in 1955, the development of nuclear energy started in the GDR, and radiobiology became an important topic. Although there is no concrete evidence that the concerns about the genetic consequences of radiation exposure enabled GDR geneticists to mobilize substantial financial resources for their research, one may say that it at least stimulated the acceptance of human genetics.[19]

One example for the application of human genetics was the field of determination of paternity which, among other things, was based on blood tests. While during the National Socialist regime physicians and scientists used these tests, inter alia, to determinate Jewish or "Arian" ancestry, in the aftermath of World War II, the tests gained importance in the cases of children who were conceived by rape or war veterans who were not confident about their fatherhood.[20] As most of the blood groups that were used for this purpose are passed down according Mendel's principles, it was ironical that while Mendel's principles were officially rejected, they were used in practice.

Moreover, the proclamation of Lysenko's doctrine did not mean that the concept of heredity diseases vanished in the GDR: In 1950, the Temporary East German Parliament (*Provisorische Volkskammer*) adopted the "Law concerning the protection of mother and child and women's rights" (*Gesetz über den Mutter- und Kinderschutz und die Rechte der Frau*). This law included a paragraph regulating abortions. Henceforth, abortions were only allowed for medical or eugenic indications. The previously acceptable social and ethical indications ceased to exist. Eugenic indication meant that an abortion was permitted if one parent was "burdened with a severe hereditary disease" (*mit schwerer Erbkrankheit belastet*).[21] Even though the expression "eugenic indication" was not used in the law, it was used by physicians referring to the paragraph in question.[22] Over time, it was partly substituted by the term "hereditary medical indication" (*erbmedizinische Indikation*) in order to stress that the focus was on the individual and not on the society.[23] However, the commissions who decided on an abortion interpreted the

[18]Schwerin, von 2004, 122–126; Cottebrune 2008, 165–166; Gerstengabe 2012.

[19]BArch, DF 1/1503. For the history of nuclear research in the GDR, see, for example, Müller 2011.

[20]Jeske 2008, 194–203, 269, 272.

[21]Gesetz über den Mutter- und Kinderschutz und die Rechte der Frau, 1953, 37; Harsch 1997, 56–57.

[22]Examples can be found in the files of the Ministry of Health Care of the GDR, for instance, in Barch, DQ 1/2036; DQ 1/21170; DQ 1/1843.

[23]Stoltenhoff 1955, 265–266; Ministerium für Gesundheitswesen an Rat des Bezirks Rostock, 11 June 1956, Barch, DQ 1/2040.

law quite strictly, with the result that by the midst 1950s, the GDR had one of the lowest rates of legal abortions in the industrialized world.[24]

The regulation of sterilization was not a part of the law. Sterilization was only allowed in case of a severe danger to the life or health of a woman.[25] Nevertheless, affected persons, or parents of children with mental disabilities, or physicians applied for their or their children's or patient's sterilization, respectively. The reason given most frequently was feeling overloaded with the care for a mentally disabled child. "Feeble-mindedness"—as the term was still called—was mostly considered as a hereditary disease. The symptoms demonstrating the diagnosis were thereby vague and quite similar to those used during the time of the Nazi sterilization law: numeracy problems, a low level of basic education, a somehow "feeble-minded" relative or in any way abnormal kinship, illegitimate children, and uninhibited sexuality.[26]

In spite of those requests for sterilization, the GDR government explicitly regulated sterilization only in 1969.[27] Thus, the government was more reluctant than the physicians. Instead of sterilization, the Ministry of Health Care (*Ministerium für Gesundheitswesen*) promoted better information strategies about contraceptives and the institutionalization of affected persons.[28] During those years, the primary concern of the GDR's population policy was to stimulate the birth of children—as they really needed to grow the workforce.

Consequently, the situation in the aftermath of World War II was characterized by breaks with the National Socialist heredity health and race politics on the one hand and by the continuity of the—in my opinion—vague concept of heredity burden with regard to some diseases on the other. The idea of heredity diseases did not disappear from medicine. For a lot of physicians, who had received their medical education before or during World War II, the ideas of Lysenko were irrelevant.[29] Besides that, scientists like Hans Stubbe or Paula Hertwig found it possible to conduct at least some genetic research. These individuals and their colleagues formed an important foundation for later research in human genetics and the emerging field of genetic counselling.

[24]Stellungnahme und Empfehlungen der Kommission zu Problemen der Schwangerschaftsunterbrechung in der DDR, BArch, DY 30/IV A 2/19/22; Harsch 1997, 60. In the GDR, the common term was "interruption of pregnancy" rather than "termination of pregnancy."

[25]Verfügungen und Mitteilungen des Ministeriums für Gesundheitswesen, 16 March 1954, BArch, DQ 1/2040.

[26]For examples, see the applications in BArch, DQ 1/2040 and DQ 1/2036. For the diagnostic framework of the Nazi sterilization law. See Doetz 2011, 87–93.

[27]Hahn 2000, 215–217.

[28]BArch, DQ 1/2040 and DQ 1/2036.

[29]One of the rather rare physicians who linked the ideas of Lysenko to heredity diseases is the psychiatrist Dietfried Müller-Hegemann. See Müller-Hegemann 1955, 242–251.

3 The Turning Point for Human Genetics in the GDR

In 1956, after Nikita Khrushchev's (1894-1971) secret speech on the 20th Party Congress of the Soviet Communist party, in which he condemned Stalinist crimes and personality cult, and after the successful petition of 100 Soviet scientists who advocated for the resignation of Lysenko from president of the Academy of Agriculture in the USSR, a critical discussion of Lysenko's ideas emerged in the *Neues Deutschland*.[30] At the same time, the GDR followed worldwide trends and devoted increasing attention toward human genetics research, despite the fact that it had no institutions specifically dedicated to the field and that studies were not always defined as "human genetics" research. Instead, they were often based on clinical questions.[31] One important factor for the international development of human genetics in those years was the introduction of the lab into human genetics. This not only led to spectacular discoveries like the extra chromosome in the case of Down syndrome in 1958 but also enabled clinical applications. Moreover, these new and more reliable techniques for counting and analyzing chromosomes as well as biochemical analysis for enzymes and metabolites shifted the focus away from diagnosis like "congenital feeble-mindedness" or schizophrenia that had been the most common cause for a coercive sterilization during the time of National Socialism to chromosome aberrations and inherited metabolic diseases.[32]

In the GDR, the first chromosome laboratories were settled at the beginning of the 1960s, for example, in the dermatology clinic of the Charité in East Berlin or the children's clinic of the university in Rostock.[33] Chromosome analysis is one example for a transnational scientific transfer. Up until the construction of the Berlin Wall (1961), it was possible to cross from the Eastern to the Western part of the town and vice versa. So did the biologist Regine Witkowski (b. 1934) who worked at the Charité. She studied the method of chromosome analysis with the help of the West Berlin geneticist Thea Lüers (1907–1990) who in turn had learned it from the geneticist Jan Böök (1915–1995) in Uppsala in Sweden.[34]

The historian Jörg Schulz characterized the development of human genetics in the GDR as a development from the medical disciplines.[35] One crucial example for this was the research on phenylketonuria (PKU) by the dermatologist and nutrition scientist Alwin Knapp (1918–1995). He started to explore this inherited metabolic disease in the midst of the 1950s at the Institute for Nutrition (*Institut für Ernährung*) in Potsdam-Rehbrücke. To help a child who suffered from PKU, he developed a less-phenylalanine protein hydrolysate in 1959. This became the

[30]Khrushchev 1956; Medwedjew 1969, 151–152; ADN 1956, 5; Arnold 1956, 11; Havemann 1956, 9; Klaus 1956, 4; Stern 1956, 4.

[31]Wittwer 1966, 775–787; Witkowski 1992, 66–72; Schulz 2007, 1281–1305.

[32]Harper 2008, 145–163, 344–347, 352–353; de Charadevian 2013, 141–152; Doetz 2011, 64.

[33]Witkowski 1992, 67–68; UAR, Personalakte Heinrich Kirchmair, foil 98.

[34]Witkowski 1992, 67; personal communication by Karl Sperling (Berlin, 22 December 2015).

[35]Schulz 2007, 1281–1305.

starting point for the industrial production of *Berlophen* that made a dietary therapy possible and—that was crucial—the GDR independent from imports from so-called capitalist foreign countries. In 1969, the GDR started a newborn screening on PKU in some districts. One year later, that measure was enlarged to the whole GDR.[36] The screening and treatment of PKU became a success story that demonstrated the benefit of human genetics.[37]

During the 1960s, East German science and the politics surrounding human genetics underwent a profound transformation. Besides the abovementioned technical innovations and the input from medical disciplines, the Lysenko era finally came to an end with the overthrow of Khrushchev in 1964.[38] Moreover, cautious reforms following the construction of the Berlin Wall resulted in a revaluation of science including human genetics. One reform in particular, the installation of the New Economic System of Planning and Management (*Neues Ökonomisches System der Planung und Leitung*), had a positive impact on human genetics.

In the face of growing economic problems in the GDR, the competition with West Germany proved to be a driving force for scientific investment. In the early 1960s, the state's economic problems had become blatantly obvious. Walter Ulbricht, head of the Central Committee of the SED, had failed to fulfill his promise to surpass West Germany's per capita consumption of food products and consumer goods by 1961.[39] As a result, the GDR's State Planning Commission (*Staatliche Plankommission*) developed a New Economic System of Planning and Management, which aimed to close this economic and material gap through the use of advanced technology and reliable forecasts. Under the slogan "The Productive Power of Science" (*Produktivkraft Wissenschaft*), science became widely accepted as a central authority in the GDR, a panacea that would solve the problems of East German society.[40] The scientific development in the West and in particular in West Germany served as a benchmark, even though the GDR distanced itself from an inhumane capitalistic West and presumed itself as the better Germany.[41] Capturing this spirit, "Surpass Without Catching up" (*Überholen ohne einzuholen*) became the slogan at the end of the decade. Coined by the Soviet cyberneticist Victor Glushkov (1923–1982), Ulbricht employed the catchphrase to proclaim that it was not just a question of catching up with international technical and scientific standards, but of finding new and superior technical and scientific solutions.[42]

The result of this trend—an official demand for a growth prognosis of the sciences—had profound implications for the development of human genetics in

[36]Herrmann, 1988, 3–4; Knapp and Machill 1974, 275.

[37]See, for example, the presentation in the popular scientific television broadcast "Vererbung nach Maß" in the TV series "Fernseh-Urania," 19 February 1975, DRA, Berlin, 001513.

[38]Löther 2010, 81–95.

[39]Laitko 1997, 35–57.

[40]Rapoport 2002, 52; Malycha 2002, 39.

[41]See, for example, Mette 1967, 3–4. See also Laitko 1997, 38–41.

[42]Roesler 2006, 126–127.

the GDR. Most notably scientists used the altered, more open atmosphere toward scientific innovations to advance their research interests in life science. At the initiative of biochemist Samuel Mitja Rapoport (1912–2004) and pharmacologist Friedrich Jung (1915–1997), both outstanding scientists as well as politically ambitious members of the SED, the GDR Research Council (*Forschungsrat der DDR*) established a commission called the Biology Group (*Biologiegruppe*). Rapoport, who headed the Biology Group, had fled to the United States during the Third Reich. Returning to the GDR after the war, he aspired to introduce elements of an "American" scientific practice, such as departmental structures and interdisciplinary cooperation. He also repeatedly criticized the GDR for lagging behind in this respect.[43] The mission of the Biology Group was to envision and plan the future development of biological research in the GDR from 1970 to 1980 on the basis of the field's international standards.

The Biology Group had several subgroups. One was the Central Working Group Genetics and Breeding Research (*ZAK Genetik und Züchtungsforschung*), which included several members of the Institute for Cultivated Plant Research. They drew up plans for the further development of genetic research in the GDR, including a focus on human genetics. Thus, the Institute for Cultivated Plant Research, headed by Stubbe, once again played an important role in the development of human genetics, even though it mainly dealt with plant, zoological, and molecular genetics. The ZAK's ideas were partially integrated in the final report, entitled "The Biology Prognosis" (*Die Biologieprognose*) in 1968. At its most fundamental level, it declared inter alia genetics to be the leading science of the future.[44] This change in course was the precursor for the establishment of the GDR's Research Project on Human Genetics (*Forschungsprojekt Humangenetik*).

In May 1971, after several years of planning, the East German Ministry of Health Care, acting on recommendations from the Biology Prognosis, and the Magdeburg Medical Academy (*Medizinische Akademie Magdeburg*) eventually signed the contract to establish the Research Project on Human Genetics. Under the direction of Jörg Schöneich (b. 1934), a member of the Institute for Cultivated Plant Research, several geneticists had drafted the proposal for this research project.[45] In order to convince the responsible authorities of the importance and hence secure permissions and funding, they emphasized the advantages of a consistent and strictly socialist health-care system for a human genetics project: only under socialist conditions could human genetic research take its proper place and not be "abused" like in capitalist countries.

[43]Scheler 2002, 5–27; Malycha and Thoms 2010, 115–117.

[44]Die Biologieprognose, IEGTM Münster, Bestand: Humangenetik DDR, Box 19; Schulz, 1997, 54–55; Malycha 2016, 224–245.

[45]ZAK Genetik und Züchtungsforschung, Gutachten zum Projektenwurf "Humangenetik", Barch, DQ 109/34.

Going further still, the authors of the project's proposal proclaimed that socialist humanism had a duty to struggle against heredity diseases.[46] Such a concept fit well with the GDR's constitution, which guaranteed the protection of health and of the ability to work.[47] They even referred positively to the term "eugenics." According to their definition, eugenics should positively influence the combination of genes within a gene pool, thereby avoiding gene combinations that would produce heredity diseases. While they distanced themselves from genetic manipulations that treat human beings simply as biomass, the authors argued that the procreation of people suffering from a heredity disease should be prevented. They emphasized that this should be strictly voluntary and laid out two strategies by which compliance should be achieved: by genetic counselling and by educating the public. Further, they maintained that eugenics should be employed to protect the genetic heritage against noxious environmental factors. They even referred to a speech of chief ideologist Kurt Hager (1912–1998), who had claimed that socialist culture must go hand in hand with a beautiful environment. The authors thus employed official catch phrases to advance their own interest in researching mutagens.[48]

The fundamental objective of the Research Project on Human Genetics was to promote socialist health care through the targeted analysis of genetic information in humans and their relationship with the environment. One of its primary goals was to develop a model for genetic counselling services. The physician Bernhard Wittwer (1936–1989) became the project's director. His home institution, the Magdeburg Medical Academy, became the project's institutional base, largely because it was the only one with a department for human genetics within a clinical institution.[49]

4 Establishing Genetic Counselling in the 1970s and 1980s

After the change in political leadership in 1971, from Walter Ulbricht to Erich Honecker (1912–1994), the establishment of human genetics continued. Honecker at last dropped Ulbricht's unsuccessful New Economic System of Planning and Management. In its place, he proclaimed the unity of economic and social policy and installed an extensive social program, among it health-care programs, an initiative that was supposed to stabilize the authority of the SED.[50]

With regard to abortion, a change took place. There was a shift from preliminary population issues to an arrangement concerning the individual woman. Still in 1965, when the GDR government had eased the very strict abortion regulations

[46]Projektentwurf "Humangenetik," 21.10.1970, BArch DQ 1/3358.

[47]Gesetzblatt, 1968.

[48]Projektentwurf "Humangenetik," 21.10.1970, BArch DQ 1/3358; Stubbe, 1982, 80.

[49]Projekt—Humangenetik—Pflichtenheft 1971, BArch DQ 109/34; Forschungsvertrag, 4 May 1971, BArch DQ 109/34; Weisemann 1997, 35.

[50]On the transition from Ulbricht to Honecker, see Kaiser 1997 and Malycha 2014, 11–68.

mentioned above, the government had rejected a far-reaching reform, because it had been afraid of a decline of the birth rate.[51] In contrast, access to abortions became relatively easy after 1972, when the East German Parliament (*Volkskammer*) enacted the Law for the Interruption of Pregnancy (*Gesetz über die Unterbrechung der Schwangerschaft*), granting women the right to first-trimester abortions upon request. In case of a positive diagnosis with regard to a genetically determined disease of the fetus, the law granted the option to terminate the pregnancy beyond the first trimester, if approved by a medical panel.[52] This and the establishment of amniocentesis in the 1970s were important developments linking genetic counselling to prenatal diagnosis and the option of abortion. Nevertheless, the Law for the Interruption of Pregnancy was controversial. The Protestant and Catholic Church as well as some physicians responded with protest.[53] Moreover, it was the first and also last time in the history of the GDR that the *Volkskammer* did not enacted a law unanimously, but only by absolute majority. Members of the East German bloc party Christian Democratic Union (CDU) had voted against it. However, the law was enacted and put into practice, subsequently, leading to a sharp increase in legal abortions.[54]

The 1970s also brought improvements for the foreign affairs of the GDR. For many years, the GDR had tried to gain international acceptance as a legitimate state in order to enter into legally binding contracts with other nations. The Federal Republic of Germany (FRG) tried to prevent this by threatening every state that wanted to establish diplomatic relations with the GDR with harsh countermeasures, including the severing of diplomatic relations.

The West German position gradually eased in the context of the New Eastern Policy (*Neue Ostpolitik*) under the West German social democrat Willy Brandt (1913–1992), effectively ending the isolation campaign. In May 1973, the World Health Organization (WHO) admitted the GDR as a member, thereby accepting the GDR as a legitimate state. This was a great foreign policy success for East Germany.[55]

Despite this rapprochement, the scientific exchanges between the GDR and Western countries remained restricted. If they wanted to travel to the West, GDR scientists first had to become a member of the "travel squad" (*Reisekader*), a status that was not easy to obtain. They needed approval from several local and central state agencies, including the Ministry of National Security (STASI).[56] Thus, only a few human geneticists held this desirable status, mainly those who worked for

[51]BArch, DY 30/IV A 2/19/22; Harsch 1997, 53–84.

[52]Gesetzblatt, 1972.

[53]BArch, DC 20/16111, foil 113–116; BArch, DC 20/17257, foil 101–103.

[54]Schwartz 2008, 183–212. For the numbers, see BArch, DC 20/16111, foil 30–32.

[55]Twenty-Sixth World Health Assembly, WHO Library, WHA 26/44; Gray 2003; Niederhut 2007.

[56]For the long and difficult process to become a "Reisekader," see Niederhut (2005). A SED membership was not a prerequisite to become a "Reisekader."

universities or institutes embedded in the Academy of Sciences.[57] Even those who were able to travel needed to engage in time-consuming communication with the responsible government agency to attend a particular congress, a process that required much advanced planning.[58] Following any foreign conference, the scientists had to write a political and technical report for the government agency, which distributed it to several other institutions.[59]

Scientific knowledge gained from congress visits was not only reported to the responsible government agency, it also spread within the community of human geneticists. Thus the *Reisekader* served as distributors of knowledge and as channels of communication with the West. Similarly, Western geneticists visited GDR congresses or institutes, among them the director of the Institute of Human Genetics in Heidelberg (West Germany), Friedrich Vogel (1925–2006), who avidly maintained international connections.[60]

In the face of travel restrictions, these forms of personal, scientific exchange were of critical importance, as they also enabled unofficial imports of scientific materials. Among the most useful secret imports were "pocket-imports" of biochemicals for genetic diagnosis that friendly Western geneticists smuggled across the German-German border.[61] In addition to those "direct transfers," other socialist countries served as middle grounds for East German scientists and the West. Czechoslovakia, which shared a border with the GDR, had begun to establish a nationwide network of genetic counselling services in the late 1960s. In the early 1970s, it began to provide training for human geneticists from the GDR.[62] Moreover, the annual meetings of the cytogenetic section in Czechoslovakia were also central to fostering international scientific exchange between the East and the West. This exchange substantially stimulated the development of clinical genetics in the GDR, according to the pediatricians Lothar Pelz (b. 1934) and Jürgen Gedschold (b. 1944).[63]

Another important contribution to the further development of genetic counselling in the GDR was an international congress on genetic counselling in the East German town of Mühlhausen in 1974, where speakers of the CSSR, Hungary, the Soviet Union, and Bulgaria reported on their experiences with genetic counselling in their countries. The congress proceedings were published one year later. As GDR scientists pointed out, the congress demonstrated the gap between the GDR and

[57] All congresses visited by GDR human geneticists are listed in the *Informationsblatt* of the GDR Society for Human Genetics at the IEGTM Münster, Bestand: Humangenetik DDR, Box 11. I want to thank Heike Petermann for supporting the examination of those documents.

[58] *Informationsblatt* Nr. 2 (1981).

[59] Niederhut 2005, 115–130.

[60] Gesellschaft für Humangenetik der DDR to Generalsekretariat der Med.-Wiss. Gesellschaften, 12.10.1981, BArch DQ 101/578a/2. For Vogel, see Cottebrune (2012, 58–59); Interview with Friedrich Vogel, 2003.

[61] Interview with Jörg Schöneich, 1997, 242–256.

[62] Vereinbarung über die Internationale Kooperation auf dem Gebiet der Humangenetik zwischen der DDR und der CSSR, BArch DQ 109/35; Seemanová 1975, 35.

[63] Pelz and Gedschold 1994, 69.

other socialist countries in this field.[64] This changed as genetic counselling quickly expanded after the success of the two pilot programs in Jena and Magdeburg, which started operating the same year. The centralist structure of the GDR health system facilitated a rapid expansion: by 1985, there were 20 genetic counselling services in East Germany, and each district had at least one counselling clinic. They were located at university clinics or district hospitals and connected with laboratories. Nevertheless, there were rather big differences between the individual districts: In 1985, there was one consultation per 1.500–1.800 inhabitants in the districts Gera, Berlin, Neubrandenburg, Leipzig, and Rostock compared to one per 5.600–12.000 in the districts Schwerin, Dresden, and Potsdam or one per 32.000 in the district Karl-Marx-Stadt. In the district Suhl, there was no counselling at all due to illness.[65]

The aim of genetic counselling was to avoid the birth of disabled or chronically ill children and to encourage procreation of people with unfound fears of having disabled or chronically ill children. The intention was not to improve the gene pool—that was considered a side effect only. The primary intention was the prevention of suffering. Thereby disability was equated with suffering.[66] Genetic counselling was supposed to increase individual health and family happiness. "The happiness of the individual is of primary importance" was one catchphrase used in "Hereditary Diseases and Hereditary Counselling" (*Erbkrankheiten und Erbberatung*) by the East German anthropologist Karl Sommer. The book was published in 1978. Its intention was the popularization of human genetics knowledge.[67]

Despite this focus on the individual and the family, objectives of population genetics did not completely disappear. Although human geneticists explicitly rejected the classification of people considered inferior and superior and refuted the racial hygienic idea that the gene pool would deteriorate within a few generations, the improvement of the nation's health as well as a reduction in infant mortality served as a legitimation for the importance of genetic counselling.[68] This attitude is reflected in a statement by scientists of the Research Project on Human Genetics, Volker Steinbicker (b. 1939), Herbert Bach, Hans-Albrecht Freye (1923–1994), Regine Witkowski, Werner Göhler (1928–2009), and Jörg Schöneich, published in the medical journal *Das Deutsche Gesundheitswesen*. They wrote:

[64]Bach 1975; report of the congress, BArch DQ 101/341/3.

[65]Jahresbericht 1973 des Medizinischen Forschungsprojektes Humangenetik, BArch DQ 109/35. Informationsvorlage: Stand und Probleme der humangenetischen Beratung in der DDR mit Schlußfolgerungen, 23.12.1986, BArch DQ 1/26482/2.

[66]Projektentwurf "Humangenetik". Gezielte Analyse genetischer Informationsbestände des Menschen in ihren Wechselbeziehungen mit der Umwelt, 21. Oktober 1970, DQ 1/3358; Körner, Grauel 1974, 269; Witkowski and Prokop 1974, 12–13; Sommer, 1978; Körner and Körner 1981, 80–91; Doetz 2016, 61.

[67]Sommer 1978.

[68]Wikowski and Prokop 1974, 12; Sommer 1978; Doetz 2016, 61–62.

> As a satisfying symptomatic therapy is possible only in a few cases, a reduction of genetically determined morbidity is only available on the way of prophylaxes—that means: timely genetic counselling. A key improvement of the genetically determined morbidity of our population can only be expected, if genetically burdened persons arrange their family planning in a way that they avoid the procreation and birth of heavily genetic impaired children. This has to happen voluntarily in their own interest and in the interest of society.[69]

Sometimes the argumentation went a step further and included also cost-benefit calculations. In 1987, the authors of a "Conception For the Gradual Introduction of Genomic Diagnostics" compared the costs of the treatment of people suffering from phenylketonuria, Duchenne muscular dystrophy, and cystic fibrosis with the costs of the genomic analysis of a family including also a prenatal genomic diagnostic. While the former was in the high six figures, the latter costs 1.500 West German Mark.[70] This argument is very much reminiscent of the cost-benefit calculations of eugenicists in the first part of the twentieth century. But as other contributions in this volume reveal, other countries deployed similar arguments.

Ethical questions were also part of the Research Project on Human Genetics. In order to articulate these ideas, the Ministry of Health Care contracted the Institute of Marxism-Leninism at the Magdeburg Medical Academy. It was charged with establishing the basic philosophical principles and the relevance of human genetic measures in regard to socialism.[71] The results of this examination were published in "Human Genetics in the Socialist Society" (*Humangenetik in der Sozialistischen Gesellschaft*) in 1977, the book that provided the theoretical justification for East German scientific work in the field.[72]

While the authors did not deny the role of biological factors in human development, they stressed the importance of social factors. Building on the ideas of the Soviet geneticist Nikolai Dubinin (1907–1998), the authors claimed that the evolution of a social program was crucial for human development. They argued that this social program, which would contain the experiences of generations, could be passed down from one generation to the next by the means of education. They named the transmission, acceptance, and internalization of this program "social

[69]Steinbicker et al. 1977,179 German original: "Da eine befriedigende symptomatische Therapie nur in wenigen Fällen möglich ist, kann eine Reduzierung der genetisch bedingten Morbidität nur über den Weg der Prophylaxe, also durch rechtzeitige humangenetische Beratung erreicht werden. Eine entscheidende Verbesserung der genetisch bedingten Morbidität unserer Bevölkerung wird nur dann zu erwarten sein, wenn genetisch belastete Personen unter Wahrung des Prinzips der Freiwilligkeit im eigenen Interesse und im Interesse der Gesellschaft ihre Familienplanung so gestalten, daß die Zeugung oder die Geburt schwer erbgeschädigter Kinder nach Möglichkeit verhindert wird."

[70]Entscheidungsvorlage: Konzeption zur schrittweisen Einführung der genomischen Diagnostik in die humangenetische Forschung und hochspezialisierte Betreuung, BArch, DQ 1/26482/1.

[71]Projekt—Humangenetik—Pflichtenheft 1971, BArch DQ 109/34.

[72]Dietl et al. 1977.

heredity" (*soziale Vererbung*). The development of mankind, according to the authors, was thus a process of ever-increasing adaption through social heredity.[73] They thereby extended the concept of heredity, a term that, as Hans-Jörg Rheinberger and Staffan Müller-Wille argue, made its way into biology through "a metaphorical transfer of a juridical concept to a description of the generation and propagation of living beings" at the end of the eighteenth century.[74] In fact, the term "social heredity" was not entirely new. The American psychologist James Mark Baldwin (1861–1934) had already used it in 1895 to describe the abilities that a child inherits from society through a process of social growth, rather than by direct biological inheritance.[75] Neither the authors of "Human Genetics in a Socialist Society" nor Dubinin referred to Baldwin, however.[76]

The concept of social heredity not only integrated human genetics with Marxism-Leninism on a theoretical level, it also enabled a socialist intellectual perspective on genetic disease. The authors argued that some individuals, depending on the severity of their diseases, were not considered capable of internalizing, developing, and passing on the "social program" and were thus incapable of participating in the progress of mankind. People suffering from PKU, for example, were capable of "understanding and internalizing the social program," given the timely introduction of special dietary measures, whereas people with Down's syndrome could not.[77] They thus defined disability as the inability to fulfill the needs of social programming, which in turn served as a justification for exclusion. That was consistent with the health politics in the GDR, which, as the historian Mary Fulbrook has noted, devoted most resources to those who held positions of power and those whose work or capacity for reproduction was of crucial importance to the economy.[78] The classification of "impaired"—as the term was called in the GDR—children reflected this attitude: These children were divided in "eligible for aid" and "not eligible for aid." While the former received pedagogical promotion, the latter were excluded from school and social life and had to live in special nursing homes or psychiatric clinics.[79] Given the living conditions, which were far from pleasant in those places, it is hardly surprising that disability was equated with suffering.

A disability rights movement comparable to those in the USA or West Germany did not exist in the GDR. As the SED proclaimed that the interests of a socialist society were aligned with the interests of single individuals living in that society, the government did not conceive of any self-organization of people with disabilities

[73]Ibd., 74–97.

[74]Müller-Wille and Rheinberger 2012, 5.

[75]Baldwin 1895, 219–223.

[76]Dietl et al. 1977; Dubinin, 1974a, 115–130; Dubinin 1974b, 63–91. So far, it is not clear whether Dubinin developed the term "social heredity" independently from Baldwin.

[77]Dietl et al. 1977, 74–97.

[78]Fulbrook 2005, 95.

[79]Boldorf 2008, 446–448; Doetz 2016, 59–60.

or chronic diseases, or of their relatives, with the exception of the General German Association of Blind People (*Allgemeiner Deutscher Blinden*-Verband) and the General German Association of Deaf People (*Allgemeiner Deutscher Gehörlosen*-Verband) that were founded in 1957. The reason of this exception was that the problems of blind and deaf people were considered more specific than the problems of physically disabled people and thus should be solved by these associations.[80] Additionally, there was the parents' council (*Elternaktiv*) of the "Association to Combat Cystic Fibrosis" (*Arbeitsgemeinschaft zur Bekämpfung der Mukoviszidose*), which was not an advocacy group in a narrow sense. Nevertheless, its members were able to push for social improvements for their children.[81] In contrast to the situation in the FRG where genetic counselling and prenatal diagnosis met with criticism of the disability rights movement,[82] I have not yet found any criticism of genetic counselling by one of the abovementioned groups in the GDR.

Criticism of genetic counselling and prenatal diagnosis came from the church. It was embedded in the criticism of abortion in general. Here, too, disability was considered a suffering. One statement of the protestant church on the ambivalent situation of parents likely to expect a disabled child was:

"If the parents want to avoid suffering, they will become guilty towards the expecting human life. If they want to avoid guilt, they will get their share of the imperfection and suffering in our world."[83]

Thus, a social model of disability, which emphasizes the aspect of a society disabling people, did not play a role in the history of genetic counselling in the GDR.

5 Practicing Genetic Counselling

Considering that genetic counselling was assumed to be strictly voluntary on the one hand and had the goal to prevent the birth of disabled children on the other hand, how did its practice look like?

Although one cannot deny the power asymmetry between counsellor and counselee, genetic counselling was not just a simple top-down process. In order to be successful, it needed the active participation of the counselees and other persons involved in the process as well as the integration of various genetic objects. The

[80]Dietl 1984, 87; BArch, DQ 1/23905; Grienitz 1989, 9.

[81]*ADGKJ* Box: GfP DDR: AG Mukoviszidose, Neuropädiatrie, Ultraschall; Box: GfP DDR: Ordner 3, Arbeitsgemeinschaften 1–5, 1983–89.

[82]See, for example, Sierck, Radke 1984 and Schenk 2016.

[83]Der Mitarbeiter, 1989, 16. German original: "Wollen die Eltern sich Leid ersparen, so werden sie schuldig gegenüber dem werdenden Menschenleben. Wollen sie Schuld vermeiden gegenüber dem werdenden Menschenleben, so bekommen sie Anteil an der Unvollkommenheit und dem Leid in unserer Welt."

process of counselling included an introductory conversation, a detailed genealogic exploration, and an extensive diagnostics. When all results were available, in some hospitals, a commission of experts discussed the case and would speak a recommendation with the aid of the diagnostic findings, which the counselee received for further family planning. The whole procedure would rarely take more than 1 year.[84]

As most of the counselees came via a medical referral,[85] it was important to sensitize physicians, and in particular pediatricians, to the benefits of genetic counselling. Moreover, it was crucial to implement genetic thinking within the medical profession so that medical records and autopsy reports could also meet the standards of human genetics.

The first step in the process was to collect as much precise information as possible about the counselee's biological kinship. Thus, family and kinship relations were now organized around genetic traits. Therefore, the counselees had to collect information about their kinship, existing diseases, and former hospitalizations. In addition, they should, if available, provide diagnostic findings.[86] Subsequently, the counsellors demanded medical records and autopsy reports. In some cases, they required physical examinations of the counselee's relatives, which at times was problematic as some relatives rejected such an examination.[87] There were other uncertainties in the process, namely, the variable expression and incomplete penetrance of some dominant genetic determined diseases. In that case, the persons concerned had the defect and passed it on, but remained without symptoms.[88] While the genealogic exploration relied on the willingness of counselees and their relatives to provide information, on physicians who developed an understanding of genetics, and on the visibility of genetically determined diseases, the problems with laboratory diagnostics were manifold: genetic objects did not function properly or just went missing; amnion cells did not always grow, which required a new puncture; or the interpretation of a karyogram was ambiguous.[89] Another challenge for human geneticists was the lack of local resources: the onsite availability of diagnostics such as ultrasound or other laboratory equipment, the adequacy of spaces, and the availability of computers. As the economic situation of the GDR grew increasingly precarious, particularly during the 1980s, these resources were often limited. Moreover, they were spread unevenly throughout the country.[90] This made certain adjustments impossible. For example, lab

[84]Steinbicker and Gedschold 1977, 235–238; Seidel 1984; Janitzky 1990.

[85]Ibid.

[86]Gedschold and Steinbicker 1984, 410; Bachmann 1983, 681–687.

[87]Braun et al. 1977, 1436–1440; Janitzky 1990, 47.

[88]Bachmann 1983, 683.

[89]Weise and Gabriel 1983, 2034–2038.

[90]Bericht über im Auftrag des Ministeriums für Gesundheitswesen durchgeführte Inspektionen der Humangenetischen Beratungsstellen der Bezirke Neubrandenburg, Schwerin, Magdeburg, Gera und Erfurt, 20 May 1987 and Personelle, materielle und organisatorische Voraussetzungen zur Überwindung der Uneinheitlichkeit des Auf- und Ausbaus der Humangenetischen Beratung in den Bezirken, BArch, DQ 1/26482/1.

capacities were insufficient to guarantee cytogenetic diagnostics of pregnant women older than 38, not to mention those older than 35.[91]

At the end of all explorations, the counsellor made a recommendation. Different analytic studies conducted at the time concluded that in most cases, the counsellors did not dissuade from having own children.[92] The genetic consultation at the children's clinic in Magdeburg, for example, recommended an abortion in 29 of 1.110 cases (3.7%), a prenatal diagnosis in 109 (9.8%), and the renunciation of own children in 87 cases (7.8%) between January 1975 and December 1979. They further mentioned no concerns in 366 (33%) and no major concerns in 235 cases (23.4%).[93] The terms used in this study, "to recommend" and "to dissuade," point to a paternalistic counsellor-counselee relationship. Other articles concerning genetic counselling also use the term "recommendation," although the authors of these texts equally emphasize that a counsellor has to accept the possibility of a counselee not adhering to the recommendation.[94] With their insistence on the voluntary nature of the process, GDR human geneticists closely aligned themselves with international guidelines of the time, as established in the WHO's report on genetic counselling from 1969. They deviated from the report's suggestions, however, in their beliefs that counsellors should give a clear recommendation to their patients. The WHO's report, instead, recommended that counselling should be as neutral as possible.[95] However, as the US-American bioethicists Dorothy C. Wertz und John C. Fletcher pointed out in their international studies, the communication of information as unbiased as possible was an ideal that was predominantly spread in the Anglo-American region. They came to the conclusion that both former East and West German geneticists reported a more directive counselling practice compared to those from the USA. In addition, the responses indicated a greater directiveness in counselling among East than West German geneticists. In doing so, Wertz and Fletcher also classified the provision of slanted information as directive.[96] As far as I can see, the terms "directive" and "nondirective" were not used in the GDR until the late 1980s after the translation of Peter Harper's book on genetic counselling was published in the GDR.[97] According to Wertz and Fletcher, the reported directiveness of French geneticists was even higher than the GDRs.[98] Furthermore, the East German human geneticists were not a homogeneous group. There was no consensus on the issue of directive or nondirective counselling. In the case of an expected child with Down syndrome, 56% would have counselled nondirective, 5%

[91]Herbert Bach in a letter to Edgar Harig (Deputy Minister of Health), 10 February 1988, BArch, DQ 1/26482/1.

[92]Seidel 1984; Janitzky 1990, 48–50.

[93]Seidel 1984, 35–38.

[94]For examples, see Bach 1974, 175, Witkowski and Prokop 1974, 33, and Sommer 1978, 81–82.

[95]WHO 1969.

[96]Wertz and Fletcher 1989, 26–31; Cohen et al. 1997, 61–80; Wertz and Fletcher 2004, 38–43 and 366–374.

[97]Harper 1988.

[98]Wertz and Fletcher 2004, 38–39 and 366–374.

would have counselled positively, and 39% would have counselled negatively.[99] Thus, it is too simple to justify the more directive approach of East in relation to West German human geneticists with the circumstances of working in a party dictatorship. Instead I would stress the importance of professional reasons for the directiveness. In the GDR, just as in West Germany, there was no profession called "genetic counsellor." Counselling was performed by physicians or biologists. Thus, counselling had a different focus, and it was not primarily considered a communication process as in the USA, where the client-centered approach of psychotherapist Carl Rogers (1902–1987) had a crucial impact on the practice of genetic counselling.[100]

6 Conclusion

While lingering fears of Nazi racial policies and the promotion of Lysenko's ideas by the Soviet Union both delayed the progress of human genetics in the GDR, the idea of hereditary diseases never disappeared from medicine. After all, human genetics was able to benefit from institutional niches and Cold War competitions. Its establishment may also serve as an example how, in spite of the SED's claim of leadership, scientists could advance their own interests under the condition of a party dictatorship. Due to East German human geneticists' restricted access to the international scientific community, they were forced to rely on individuals who acted as channels of communication with the West, as well as socialist "brother states," which were more easily able to forge connections with Western countries.

Through the development of the concept of "social heredity," East German academics succeeded in linking human genetics with the idea of socialism. On a more practical, medical level, genetic counselling with its focus on prophylaxis, individual health, and family happiness fits perfectly into the self-concept of the socialist health-care system—a system that devoted most of its limited resources to those people who were important for the economy.

The main goal of genetic counselling was to avoid the birth of disabled or chronically ill children and to encourage procreation of people with unfound fears of having disabled or chronically ill children. To realize this, the liberalization of abortion and the establishment of amniocentesis in the 1970s were crucial as they made a targeted abortion possible. At the same time, human geneticists emphasized the voluntary nature of genetic counselling. Eugenic goals were a consideration for GDR scientists, but not the driving force for establishing counselling. However, limiting factors for further expansion of genetic counselling were neither ideological nor ethical reasons, but economical ones—namely, inadequate local resources.

Ethically questionable was, in my opinion, the ambiguous situation that genetic counselling should be strictly voluntary on the one hand and had the goal to prevent

[99]Cohen et al. 1997, 67.

[100]Doetz 2016, 63. For a history of genetic counselling in the USA, see Stern 2012.

the birth of disabled children on the other hand. The situation was compounded by the absence of independent advocacy groups of people with disabilities or chronic diseases: A disability rights movement that questioned the equation of disability and suffering and thus could have called into doubt the goals of genetic counselling did not exist in the GDR.

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Remarks on the History of Genetic Counselling in Czechoslovakia, 1945–1990

Michal V. Simunek

Abstract Genetic counselling was established in the second half of the 1940s, and its name emphasised more individual one-to-one relationship and targeted at the educational objective of the client. The main aim was to make more informed decisions regarding the birth defect concerns and questions after counselling than they would have been without it. In Czechoslovakia, its establishment occurred under the specific position of genetics in the 1950s and 1960s and developed mostly in the 1970s and 1980s.

Keywords Genetic counselling • Czechoslovakia • Twentieth century

1 Introduction

During the period of 1948–1989, Czechoslovakia developed a system of centralised socialist health care based on state health insurance. As part of this system, medical genetics was also established, though its development was far from straightforward. Moreover, a unified and centralised model of health care guaranteed to the entire population provided considerable space for prophylactic genetic measures that varied from that on the other side of the Iron Curtain.

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2 1940s and 1950s

Medical genetics in Czechoslovakia was introduced in 1946–1947 with the establishment of the first professorial chair at the Faculty of Medicine of Charles University in Prague.[1] Just as in many other countries, postwar medical genetics in Czechoslovakia had some important continuities—both personal and scientific—with the interwar reform eugenic movement.[2] During this earlier period, there existed premarital counselling, in which a eugenical minded physician trained in Mendelian genetics provided prospective partners with 'medical genetical advice'. At this time, there were different opinions of whether this premarital counselling should be voluntary (and eo ipso individual) or obligatory.[3] Genetic counselling as introduced by Sheldon S. Reed (1910–2003) in 1947 in the USA was, however, not reflected immediately in Czechoslovakia.[4]

Because of the political development in Central Europe during the late 1940s and 1950s, there was an official attempt to abandon genetic knowledge and banish it from the medical curriculum. In Czechoslovakia this was a period that saw increased influence of scientifically obsolete doctrines of Olga B. Lepeshinskaya (1871–1963) and also Trofim D. Lysenko (1898–1976), although this was ending by the 1960s. The exact impact on medical science was not historically analysed yet. In 1962 genetics was, however, included into the then new compendium of general biology for physicians.[5]

At the same time, there was an enormous increase of preventive medical care with huge impact especially on paediatrics and obstetrics in the newly centralised system of public health care after the communist takeover in Czechoslovakia in 1948.[6] The decisive year was 1952, when—based on the resolution of the Communist Party of Czechoslovakia and the Government—the so-called united health-care system became reality with a special focus on the care of the mother and child.

[1] Bohumil Sekla (1901–1987) studied first history and Czech language at the Faculty of Philosophy of the Charles University in Prague. Soon he started focusing on psychology and then transferred to the Faculty of Medicine. He became an assistant and member of the Czechoslovak Eugenics Society. In the mid-1930s, he conducted experimental research of both short-lived and long-lived Drosophila flies. During his stay in Edinburgh, he worked together with Francis A. E. Crew (1886–1973). After being appointed the first professor of (medical) genetics in Czechoslovakia in 1946, he became also a director of the Institute of Biology of the Faculty of Medicine at the Charles University in Prague. In the 1950s, he started to deal with immunogenetics, and after the rehabilitation of classical genetics in 1964, he actively participated in the preparation of the Governmental Resolution on Basic and Applied Research in Genetics and was also in charge of the departmental plan of a research of human and medical genetics.

[2] On the history of eugenics, see Šimůnek 2015, 127–190.

[3] See Sekla 1941, 188–189.

[4] See Reed 1974. For the definition of genetic counselling, see Fraser 1974.

[5] Sekla and Krajnik 1962.

[6] Prokopec 1975.

Further resolutions followed in 1964 and 1977. In 1966 a central law on the health of the nation (zákon o zdraví lidu) was adopted.[7]

The approach towards genetically handicapped people and their quality of life was based, generally speaking, on the idea of the so-called factual humanism (reálný humanismus) rising from Marxist theory.[8] In practice there was, however, a strong accent on their working capabilities, which was a situation quite similar to other socialist countries.[9]

At the end of 1950s, it was estimated in Czechoslovakia that for every 100,000 newborns, there were about 2000–2500 newborns with congenital anomalies. In 1957 there was established in Prague a consultation office for congenital anomalies as a part of the central Prague Institute for the Care of Mother and Child (Ústav péče o matku a dítě). In this Institute, although congenital malformations were not explicitly understood as 'genetic', genealogical and kinship relations were taken into consideration in their assessment. During the first 3 years of its existence, the total number of 251 children from Prague and neighbourhood area was examined here.[10]

3 1960s and 1970s

Beginning with the 1960s, the principles of responsibility of the State towards the family, as was required by the Law on the Family (zákon o rodině) from 1963, enabled to present the genetic counselling as a part of the State's contribution providing the family with the necessary information.[11]

Already in 1961 the unofficial registration of congenital anomalies became obligatory, and the first research Department for Medical Genetics was established at the then Faculty of Paediatrics of Charles University in Prague.[12] Since January 1964 there was official registration of congenital anomalies in all liveborn children until their 28th day of life and all dead newborns. First there were 36 registered anomalies and since 1975 their number increased at 60.[13]

As in other 'socialist' countries, the decisive year for the 'rehabilitation' of genetics was 1965 (when the Mendel-Symposium took place in Brno). Already 1 year before this symposium a research project on the medical genetics had been incorporated into the State Plan of Basic Research, which was obligatory for the all research institutions in Czechoslovakia. It was usually planned for 4 years, and for the period 1976–1980, genetic counselling was part of the subprogramme called 'A Complex Genetical Care'.[14] Parallelly medical genetics became part of the

[7] 1981.
[8] Démant 1955.
[9] Doetz 2016.
[10] Kučera 1962.
[11] Kapras 1985.
[12] Seemanová 1975.
[13] Šípek 2014.
[14] Houštěk 1981: 853.

medical curriculum and in 1967 the Czechoslovak national Society for Medical Genetics (Společnost lékařské genetiky; abbr. SLG) was established with about 130 members in 1975. As a national accreditation body it played (and plays) a key role also concerning the issues of genetic counselling.[15]

During the 1960s the Czechoslovak representative also took part in the activities of the Scientific Group on Genetic Factors in Congenital Malformations, which was established by the WHO and which significantly shifted the attention towards the causal genesis of the congenital malformation.[16]

The first real genetic counselling practice in Czechoslovakia was established in 1963 in Brno by the Research Institute of Paediatrics (Výzkumný ústav pediatrický).[17] The probands were divided into two main groups. First genetic counselling was delivered to parents who either requested it of their own accord, or who were referred by their doctors. These were mostly parents with already affected child. Second there were patients and families detected either by an active screening survey for metabolic disorders, or via other genetically determined diseases like diabetes mellitus, haemophilia, osteogenesis imperfecta tarda, cystic fibrosis, muscular dystrophy type—Duchenne etc.[18] Among the physicians involved was, for example, Professor Renata Laxová (b. 1931), who emigrated after 1968 to the US and significantly contributed to the further development of genetic counselling there.[19]

At the end of the 1960s further development of genetic counselling was foreseen by the third report—recommendations of the WHO. There was proposed, for example, a coverage by the social and health insurance scheme, specialised training in medical genetics, publicity, sharing and unification of information, etc.[20] In Czechoslovakia the introduction of medical genetics into medical practice—including the first systematically created network of the offices for genetic counselling—took a more centralised form and was realised under the supervision of the Ministry of Health. It was based on a specific programmatic document called *Conception of Medical Genetics*, which—in its very original form—was accepted and officially published in October 1969 being effective since 1970.[21] By this act, medical genetics in Czechoslovakia was acknowledged as a special branch of medical sciences, introduced into the medical and social system and provided with a specific amount of money from the state budget. In 1980 more detailed and progressive

[15]Štark 1975.

[16]Kučera 1970.

[17]Laxová 1968, 4–5. See also Seemanová 1975: 36; Mrskoš 2002.

[18]Laxová 1968: 157–160.

[19]Interview of the author with R. Laxová, Prague 2014, Collection of Interviews on History of Medical Genetics in Czechoslovakia, Institute of Contemporary History—Centre for the History of Sciences and Humanities Prague.

[20]WHO 1969.

[21]Brunecký 1972; Kučerová 1980; Kučerová 1981.

version was prepared under the supervision of associate professor Milan Macek Sr. (b. 1932) and adopted by the Ministry of Health.[22]

Following the position of the official authorities such as Professor Boris V. Petrovskyi (1908–2004), the Soviet Minister of Health in 1965–1980, who declared genetic counselling as necessary during his Prague visit in 1975 and supported the education of new highly qualified staff in this field, the publication of the *Conception of Medical Genetics* was a key moment.[23] Following the first version of the *Conception* departments of medical genetics containing also offices for genetic counselling should be created in all eight administrative districts of Bohemia and Moravia until 1973 or 1980 respectively. The ideal personal equipment of such departments were understood as follows: three specialised medical doctors, five further graduated workers (two geneticists, one anthropologist, one biochemist and one statistician) as well as an adequate number (eight to ten) of laboratory personnel.[24] Routine work at that time included chromosomal examination, examination of sex chromatin, biochemical examination, anthropogenetic examination and genealogy.[25]

For example, in Prague, the first of two offices for genetic counselling was established in 1969 at the Regional Institute of National Health (Krajský ústav národního zdraví; abbr. KÚNZ) and at the Faculty of Paediatrics. During the first 3 years of its existence, it carried out more than 3000 examinations, including chromosomal examination, sex chromatin and Y-corpuscule examination, anthropogenetic examination, IQ tests and detailed genealogical examinations.[26] One huge challenge was how to control the screening and examination of families. To facilitate this, they established a genetic register for the whole hospital area with a permanent checklist of families with a history of genetically conditioned deviations and their active follow-up by means of computer techniques.[27] The main objectives of these registers were (a) application of genetic preventive methods, (b) planning of necessary funds for social care and (c) a base for further rational population measures.[28]

Based on the experiences from Prague, the largest group of people searching for genetic counselling were parents with an affected child asking for information risks to later pregnancies. Less frequently represented were the siblings or other relatives of affected individuals. The smallest group comprised the affected persons themselves or their partners. The more common conditions to affect such people were polygenic

[22]Interview of the author with M. Macek Sr., Prague 2013, Collection of Interviews on History of Medical Genetics in Czechoslovakia, Institute of Contemporary History—Centre for the History of Sciences and Humanities Prague.

[23]Quoted in Prokopec 1975, 75.

[24]Sekla 1973, 11.

[25]Ibid., 11–12.

[26]Špale 1973, 11; see also Seemanová 1973, 225; Sekla 1973, 12–13; and Seemanová 1990, 615.

[27]Špale 1973, 29.

[28]Kapras 1973, 32.

hereditary disorders, inborn errors of metabolism, chromosomal aberrations and recessive sex-linked diseases.[29] The effectiveness of genetic counselling concerning reproductive choices was evaluated among 246 families in the period from 2 to 6 years after the genetic counselling session. Prague experienced a decrease of 66% of born children in the families with a 25% risk or more.[30]

The genetic counselling services in Prague became especially well known because of close connection to the clinics and syndromology.[31] An exact clinical diagnosis was understood as the essential condition of the genetic counselling, especially for those affected with known heterogeneity (mucopolysaccharidoses, myopathies, etc.) Therefore, close cooperation with other medical specialists was necessary, as was precisely documented genealogical data, which represented the basis for the genetic counselling at that time.[32] A leading role in this was played by Professor Eva Seemanová (b. 1939), who, for example, discovered the Seemanova II syndrome, Nijmegen breakage syndrome or Berlin syndrome, which is a rare autosomal recessive congenital disorder causing chromosomal instability and is occurring in the West Slavic population. In syndromology Prof. Seemanová cooperated also very closely with East German and Soviet colleagues.

Cytogenetics gradually became of key importance because at least half of all probands coming to the office for genetic counselling needed chromosomal analysis. Overall, an abnormal karyotype was detected in more than in 20% of these probands with one third being unusual.[33] However, especially at the beginning of the 1970s, not every regional centre of medical genetics was adequately equipped with a cytogenetic laboratory, so the samples were sent for analysis to Prague.

Prenatal genetic diagnosis was first established at the Department of Medical Genetics of the former Institute for Child Development Research of the Faculty of Paediatrics of Charles University in Prague in 1971. From 1970 Prenatal Genetic Diagnosis was experimentally tested on amniotic fluid cells obtained from the clinical interruption of pregnancies.[34] At the same time, amniocenteses and prenatal ultrasound examination in genetic-risk pregnancies were introduced at the Clinic of Obstetrics and Gynaecology of the same faculty. During the 1980s, further accessibility of the Prenatal Genetic Diagnosis services was guaranteed, as well as increase of the age indications, which in 1983 reached about 60% of all examinations.

Thanks to the *Conception* from 1970 the basic Prenatal Genetic Diagnosis services were introduced also in regional centres. Their establishment, however, was rather a gradual process and their technical and personal equipment was not so satisfactory as was foreseen by the *Conception*.

[29]Seemanová 1973, 226.
[30]Židovská 1985, 720.
[31]Seemanová 1985, 607.
[32]Seemanová 1973, 227.
[33]Kučerová 1982, 3.
[34]Macek 1997, 495.

For example, in 1972, the office of genetic counselling was established for Western Bohemia in Pilsen, and in 1977, the independent centre of medical genetics was established there.[35] In 1975 the Laboratory of Medical Genetics was established at the Clinics for Gynaecology and Obstetrics of the Medical Teaching Faculty in Olomouc, Central Moravia.[36] In Northern Bohemia, the Regional Department for Medical Genetics was established not before 1980, and after several years, it was not equipped by laboratory at all.[37] The most underdeveloped situation was, for example, in Southern Bohemia, where the PGD services could only be delivered first in 1984.[38] In the 4-year period (1976–1980), × total 3067 probands were consulted in all offices of genetic counselling in Bohemia and Moravia.[39]

In 1975 genetic counselling was incorporated by the Ministry of Health into the population genetics, which should from 1980 onwards aim on the 'complex eugenical and social regulation of the development of our population'.[40] How relevant this aim could be is, however, unclear.

4 The 1980s

In this period, the position of medical genetics in Czechoslovakia was definitely secured, and it became irreplaceable both in the official health-care system and clinics. Its further development was outlined in detail in the official document called *Conception of Medical Genetics* (Koncepce lékařské genetiky).[41] The document described the aims and diagnoses of medical genetics, but it also explained that medical genetic centres should:

- Within the framework of preventive medical care, it aims at early diagnosis, treatment and prevention of genetic disorders and congenital defects.
- It provides and determines genetic risks in families and population and recommends suitable preventive measures and treatment.
- It diagnoses chromosomally and metabolically conditioned defects and other congenital defects in various stages of ontogenesis.
- It diagnoses teratogenic, mutagenic and other genetically hazardous factors during the preconception and prenatal stage.

Keep records of genetic disorders in the population, which means in particular (a) developing a register of genetically handicapped families and families with

[35]Lošan 1978, 37.

[36]Šantavý 1981, 65; for map see Štark 1975: 25.

[37]Kofer 1985, 682.

[38]Křesnička 1988, 470.

[39]Houštěk 1981, 853.

[40]Úvahy o budoucnosti lékařských věd. Koncepční prognostické studie [Considerations on the Future of Medical Science. Conceptual Prognostic Studies]. Praha: Avicenum 1975: 44–46.

[41]Věstník Ministerstva zdravotnictví [Bulletin of the Ministry of Health] 1980: 127–131.

increased risk of congenital and genetic disorders and actively searching out handicapped individuals and carriers of congenital and genetic disorders, (b) collecting and analysing data on the current state of genetic stress that the population is exposed to and (c) developing prognoses of further genetic development of the population and suggesting measures leading to a lowering of genetic stress.

Regarding the genetic counselling, the Conception capacitated the practical physicians and especially obstetrics for the consultation.[42] A directive approach towards the families was seen as "incompatible with the ethics of modern medical genetics".[43] Important was also the financial aspect, because in 1983, it was calculated about 50,000 Czechoslovak Kronen for the asylum and child per year.[44]

A significant consequence of this was an increase of centres for cytogenetic testing, which provided cytogenetic, chromosomal, eventually other specialised testing also for other departments of preventive medical care including pathology as well. It led to further professionalisation. In 1986, for example, a section of genetic laboratory technicians was established within the Society of Medical Health Workers.[45]

Departments of medical genetics at Type III Hospitals in select regions may establish, where needed and subject to a decision of the Ministry of Health, specialised departments of medical genetics serving a larger area. Such departments could include (a) a specialised section of prenatal genetic diagnostics (app. one department per two to three million inhabitants), as found at Departments of Medical Genetics of Teaching Hospitals (Fakultní nemocnice) in Prague (capital city and Central Bohemia), Brno (Southern Moravia), Hradec Králové (Eastern Bohemia) and Olomouc (Middle Moravia) and (b) a specialised section for diagnostics and treatment of congenital metabolic disorders or other congenital disorders. Other specialised sections could also be established, e.g. for the testing of alpha-fetoprotein at the Department of Medical Genetics of Teaching Hospitals in Prague and in Brno, or a preconception section of genetic care at select obstetric departments (clinics) of Type III HwPs of teaching hospitals. In order to use resources rationally, specialised departments of medical genetics were located—based on mutual agreement—located at relevant research institutes of the Ministry of Health or other departments and institutes. In 1980 these were, for example, (a) a specialised section for the testing of mutagens with the Institute for Hygiene and Epidemiology in Prague and (b) a specialised section for prenatal genetic diagnostics—foetoscopy (and others) with the Institute for Care of Mother and Child in Prague.

In order to provide inpatient care for the diagnostics and treatment of genetically affected families and families with genetic risk, hospital beds were provided by the relevant specialised departments (such as internal medicine, paediatric, obstetric wards of the Type III hospitals).

[42]Kapras 1985, 586–591. See also Rubín 1981.

[43]Kapras 1985, 586–591; see also Rubín 1980.

[44]Šantavý 1983.

[45]Křížová 1987.

5 The International Cooperation

Concerning international cooperation, Czech experts were in close touch with colleagues both from the so-called socialist and Western countries. In 1975 a Standing Commission on Health of the Comecon countries was established, which adopted a special research plan for the period 1976–1980.[46] Under this Commission, Czechoslovak experts should be responsible for the genetic counselling.

Close contacts existed especially with the East German colleagues. They were invited, for example, to participate at the final meeting of the project on human genetics sponsored by the Ministry of Health of the German Democratic Republic in 1974 and took part in first Symposium of Socialist Countries on PD of Genetically Caused Diseases in Rostock in 1982.[47] But the cooperation occurred also on very practical level, as, for example, in case of Northern Bohemian centre for medical genetics and the Institute of Medical Genetics at the Faculty of Medicine of the University in Greifswald in 1983 (Prof. Hermann), which concerned the analysis of Lesch-Nyhan syndrome (LNS). On the other hand to this, geneticists from Eastern Germany could meet their colleagues from Western Germany and Europe at the meetings organised in Czechoslovakia especially in the 1980s. These meetings were designed as occasions for inviting the leading experts in medical genetics and genetic counselling from Western Europe or the USA and thus played an important role in the history of medical genetics at the end of the Cold War.

6 Conclusion

The development of genetic counselling in postwar Czechoslovakia depended on the general position and professional endorsement of medical genetics. While during approximately the first two decades after 1945 the focus was on a rehabilitation of genetics as such, a pragmatic approach, supported by numerous discoveries and progress in basic research, has been prevalent since the 1960s. In the Czechoslovak case, the constitutive period came in the 1970s, a time which saw the introduction of a centralised and above all complex conception of medical genetics, including genetic counselling, as well as a spread and improved accessibility of diagnostic methods. Thanks to such and other measures Czechoslovakia in many ways served as a positive model for other countries of the Socialist Bloc, whereby closest collaboration was established with the former German Democratic Republic.

[46]Semrádová 1987; Prokopec 1981; Schiavone 1981, 111.

[47]Zwinger 1982, 73.

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The Establishment of Human Genetic Counselling in Austria in the 1970s in Between the Establishment of Human Genetics and the Eugenic Indication of Abortion

Katja Geiger and Thomas Mayer

Abstract The chapter focuses on the establishment of genetic counselling in Austria, especially in Vienna during the 1970s. It is our assumption that the emergence of genetic counselling services during the 1970s benefited from the eugenic indication of abortion, which was enabled in Austria in 1975 due to the reform of the penal law. Counselling offered mainly abortion as a solution to genetic or chromosomal aberration, because no genetic therapy was available at that time. We focus on Vienna's tradition on eugenic counselling as in 1922 the first municipal marriage counselling service was established in "Red Vienna". However, the practice of this early counselling demonstrated that not eugenic but sexual advice was sought by its visitors. While eugenic counselling was obligatory during the National Socialist reign, concepts of counselling outlived World War II in the Catholic marriage counselling.

Furthermore, the relation of the professionalization of human genetics, the establishment of genetic counselling and the relaunch of eugenics during the 1960s and 1970s are examined. Concepts of counselling in Austria supported the individual and the society at the same time. While the term "eugenics" seemed to vanish from the scientific discourse during the 1970s, we have demonstrated that some counsellors like the child neurologist Andreas Rett still supported eugenic reasoning in the early practice of counselling. The eugenic indication of abortion remained an important possibility for the outcome of a counselling interview.

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In our chapter we will focus on the establishment of genetic counselling in Austria and especially in Vienna during the 1970s. Furthermore, we intend to investigate the relation between the professionalization of human genetics, the establishment of genetic counselling and the relaunch of eugenics during the 1960s and 1970s.

In 1974 human genetic counselling started in Austria. Four services were established in this year alone: one in Graz at the human genetic department and three in Vienna, one at an intern clinic, one at the first women's clinic and one at the department of anthropology at the museum of natural history in Vienna. At least in one case, the genetic counselling service in Marburg (Federal Republic of Germany) that was established in 1972 by the human geneticist Gerhard Wendt served as a model.

However, it is our assumption that the emergence of genetic counselling services in 1974 went hand in hand and benefited from the eugenic indication of abortion, which was discussed at the time in Austria and finalized in January 1974. Genetic counselling offered diagnostics on cytogenetic level but offered mainly abortion as a solution to genetic or chromosomal aberration, because no genetic therapy was available at that time.

1 The Eugenic Indication of Abortion

As in other European countries, the eugenic indication of abortion was discussed in the 1960s and 1970s due to the reform of the penal laws. Eugenic indication meant that abortion was not to be punished if the foetus was to be severely damaged. In older versions of penal laws, abortion was considered a major crime in Austria and in many European countries, and women were endangered to be convicted as murderess in case of aborting a foetus. Due to liberalization of social life in European societies in the 1960s, this view had changed, and new approaches to deal with abortion were considered.

The eugenic indication was first an issue of discussion after World War I, when in the young Republic of Austria, social values were debated due to the loss of the war and the end of the Austrian-Hungarian Empire. As abortion was still forbidden, liberal forces demanded a change in legislation to allow or at least not punish abortion to a certain extent. So also the eugenic indication of abortion was discussed in Austria since the 1920s, especially among social democrat-oriented doctors.[1] However, the definition of the eugenic indication remained rather vague as no precise understanding of what diseases or aberration existed. In Vienna the social

[1] Lehner 1989; Mesner 1994.

democrats ruled the city, and its representative for health and welfare, the well-known professor for anatomy Julius Tandler (1869–1936), advocated openly the eugenic indication of abortion as one measure to control and improve the genetic quality of a population, next to other measures like sterilization. As most eugenicists in Austria, he agreed that abortion was not to be left to women's choice but to the state's programmes of birth control.[2] Thus, in 1924 at a meeting of social democrats physicians, Tandler argued for the medical, the social and the eugenic indication for abortion and for a commission of doctors that was to decide on abortion. Although his proposal on the doctoral commissions was not shared by all participants, Tandler's suggestion on the three indications for abortion was included in the platform of Social Democrat Party in 1926. However, in 1927 the bill of the new penal law proposed by the Catholic and German-National coalition only opted for a medical indication. Interestingly, half a century later, in 1974, Tandler's suggestion was considered, when the penal law was revised by the social democrat government in power, resulting in the legalization of the eugenic indication for abortion. Even though Tandler and the Social Democrat Party advocated a liberalization of the law on abortion, the penal law (Österreichisches Strafgesetzbuch) was even restricted in 1937 by the conservative and Catholic government.

Eugenic abortion was only enabled during the period of the terror regime of National Socialist rule in Austria between 1938 and 1945, when the Nazi law "Gesetz zur Verhütung erbkranken Nachwuchses" (Law for the Prevention of Hereditarily Diseased Offspring) permitted the abortion of a pregnancy. Based on this legal premise, abortion was enforced on the so-called unfit women.[3]

Since the 1950s, the eugenic indication of abortion was discussed again in the drafts for a reform of abortion in Austria by various groups, continuing the debates of the 1920s. The debates focused on an expected "damaged" or "severed" child. In 1956 the social democrats stated their position on legalizing abortion and opted for three kinds of indications: the medical, the social and the eugenic indication, following their party platform of the 1920s.[4] In the ongoing discussion with the other major party in Austria, the Catholic conservative ÖVP, the "Volkspartei" (People's Party) and the social democrats agreed to a compromise and discarded the eugenic indication in 1957. During the 1960s several plans for the reform of the law on abortion included a eugenic reasoning. The social democrats included the eugenic indication in their party platform in 1969.

When in 1971 the social democrats ruled Austria, they started their programme of a health reform, which encompassed the reform of the law on abortion. Since 1971 the eugenic indication of abortion was part of the intended new penal law. It also remained in the proposal, when the women's right movement strongly

[2]Mayer 2015b.

[3]Spring 2010; Czech 2007, 269–274. Although legally the pregnant woman had to permit the abortion, in practice an abortion often was enforced against the will of the woman.

[4]Mesner 1994, 96–97.

advocated the so-called "Fristenlösung", an abortion permitted within the first 3 months of pregnancy.

The eugenic solution was also supported from the political opponent, the right-winged FPÖ, which embraced many former Nazi members. However, it is quite remarkable that a member of parliament of the FPÖ argued in 1971 that there is too little genetic knowledge to justify eugenic abortion whereas another member of the same party justified only 2 years later, in 1973, eugenic abortion with the notion that human genetic knowledge was advanced enough to distinguish between genetic and nongenetic causes.[5]

At the turn of 1973 and 1974, abortion was legalized in Austria, and next to the "Fristenlösung", a medical and a eugenic indication of abortion was permitted. When the "Fristenlösung" allowed an abortion during the first 3 months of pregnancy, the medical and eugenic indication of abortion enabled abortion until birth. The implementation of a eugenic reasoning in the penal law was an important condition for the "therapeutic" effect and the success of the genetic counselling, all the more, when prenatal diagnostics like amniocentesis worked only after the first 3 months of a pregnancy. Genetic counsellors in Vienna argued during the 1970s that eugenic abortion was a key to the success of genetic counselling.[6]

2 Family Policy, Genetic Counselling and Biopolitics

At the same time of the establishment of the genetic counselling and the legalization of eugenic abortion, other legal initiatives in the field of public health care were taken[7] that supported the practice of the genetic counselling: First, the "Law of the Promotion of Family Counselling" ("Familienberatungsförderungsgesetz") and second, the "Mutter-Kind-Pass", a screening programme for the pregnant woman and her newborn child. We argue that those initiatives had different impacts on genetic counselling, namely, to spread eugenic and genetic knowledge among people and counsellors alike and to provide scientists with data.

The "Law on the Promotion of Family Counselling" enabled the establishment of family counselling services, which were to enable rational family planning and to spread eugenic ideas alike. In 1974 eugenic thinking was lectured by medical experts like the child neurologist Andreas Rett (1924–1997) at instruction workshops for family counsellors.

The voluntary examinations of the "Mutter-Kind-Pass", which started with the pregnancy of a woman and ended with the first year of the child and were paid generously, and, as a side effect, provided human geneticists with data. The child

[5]Stenographisches Protokoll, 10. Sitzung, 9. December 1971, 674–675; Stenographisches Protokoll, 84. Sitzung, 27 November 1973, 8013.

[6]Vormittag 1974; Vormittag 1980.

[7]Mesner 1994, 228–229.

neurologist Andreas Rett put the "Mutter-Kind-Pass" in the tradition of biopolitic measures of the 1920s "Red Vienna" government. Rett considered the biopolitic importance of the "Mutter-Kind-Pass" as a preventive measure that worked on individual level as well as on the level of the whole population. It was Rett who supported the introduction of the "Mutter-Kind-Pass" due to his excellent connection to the Austrian minister of health, Ingrid Leodolter (1919–1986). One main idea of the "Pass" was to reduce infant mortality. In 1978, 99% of pregnant women participated in the "Mutter-Kind-Pass"-screenings - certainly encouraged by a high premium of 16,000 ÖS that was rewarded to the women afterwards. However, the child neurologist Rett was not entirely satisfied, since the collecting of data was not uniform and did not contain all developmental disorders. In his opinion the data was not entirely useful for genetic counselling.

To sum up, the social democrat government modelled the health reform of the 1970s on the public health reforms of the 1920s of Vienna's social democrat government and its supervisor, the anatomist and health politician Julius Tandler, in four aspects: Firstly, the eugenic indication of abortion; secondly, the intended collection of data of the population for reasons of public health care; thirdly, the establishment of counselling services that had to determine the health of the offspring; and finally, at least by some actors, like Andreas Rett, it was argued that the eugenic sterilization of mentally disordered girls was a need of public health care.

In evolving those programmes, the government was cooperating with medical experts among which were some of the genetic counsellors. One of the key actors was Andreas Rett, who was well connected within Austrian health politics and the Austrian and German genetic scientific community as well. He was dealing with children with developmental disorders. Rett performed genetic counselling since the end of the 1960s, but did not run an official counselling service at this time.[8] His audience was the families of his patients, who wanted information on the genetic risk of further offspring. At Rett's clinic 100 chromosome analyses were performed each year. Since the beginning of the 1970s, Rett's clinic cooperated with anthropologists from the Department of Human Biology. Rett claimed the uniqueness of this cooperation in Austria and presumably in Western Germany.[9] In 1974 the genetic counselling of the First Women's Clinic cooperated with Rett's clinic by asking for clinical diagnosis. According to Rett his clinic was the only institution in Austria that collected data on trisomy 21 on a broad base, dealing with 50–60% of all cases of trisomy 21 in Austria.

Rett's Nazi past was mentioned in some papers, when he joined the Hitler youth as early as 1932 and the Nazi party in 1942.[10] Furthermore, he used the material of

[8]Since 1967 a cytogenetic laboratory existed, where research was conducted and diagnostics were made. In 1976 Rett's clinic was listed as one of the five existing genetic counselling services. See Schnedl 1976, 62.

[9]However, Vormittag also cooperated with the Vienna anthropologist Margarete Weninger (1896–1987) on genetic issues of epidermal ridges ("Hautleisten"). See Vormittag 1974.

[10]Schönwiese 2012; Ronen et al. 2009.

the Nazi child euthanasia programme for scientific publications and cooperated thereby with the collaborator of Nazi child euthanasia, the physician Heinrich Gross.[11] Rett's support for eugenics was only mentioned briefly yet. As recent as 2009, a paper argued that Rett supported and encouraged "parental consented sterilization of their retarded daughters", which "was already controversial at the time".[12] In addition, we want to argue that Rett advocated the idea of eugenics in various ways: he supported the eugenic indication of abortion by arguing for eugenics as medical expert during the process of legalizing abortion in 1972. In cooperation with women clinics, he ordered the sterilization of mentally disabled girls and the interruption of their pregnancies during the 1960s and early 1970s, when those measures were illegal by law.[13] He advocated eugenic sterilization of families of lower social class in case of genetic risk. He considered eugenic thinking to be an essential part of a rational family planning. Rett used the reform of the penal law for scientific research, when his student finished a dissertation on the impact of the eugenic indication of abortion on families.[14] And Rett advocated eugenic advice as a proper outcome of a counselling interview.

2.1 From Eugenic to Genetic Counselling

Vienna's tradition on eugenic counselling started in 1922 when the first municipal marriage counselling service in Europe was established in "Red Vienna". The marriage counselling centre (Gesundheitliche Beratungsstelle für Ehewerber) in Vienna was the first of such institution in Europe. The Vienna Counselling Service became an immediate model institution within Austria and abroad. It provided advice on a voluntary basis for marriage candidates and married couples. The centre was established in 1922 under a social democratic city government and implemented at the communal level by Julius Tandler, at the time Vienna's councillor for public welfare (Wohlfahrtswesen). It became in fact the first practical eugenic institution that functioned in Austria before 1938. Tandler intended to determine one's chance of bringing forth mentally and physically healthy offspring. Located at the communal health centre in Vienna, the service provided people with advice as to whether or not they should procreate. Attendance at the clinic was voluntarily, and, as records show, people wanted primarily sexual advice. However, the practice of this early counselling demonstrated that not eugenic but sexual

[11]Neugebauer and Schwarz 2005, 230–231.

[12]Ronen et al. 2009, 124.

[13]Rett 1979; Rett and Seidler 1981, 277–286; Sterilization of an incapacitated person was legalized by the decision of the Austrian Supreme Court in 1977, when sterilization was permitted, if the guardian agreed. See OGH (Oberster Gerichtshof (Austrian Supreme Court)), 12 December 1977, 1 Ob 735/77.

[14]Bublitz, Peter-Michael (1977): Struktur und emotionelle Integration von Familien mit mehr als einem hirngeschädigten Kind, Diss., Univ. Wien.

advice was sought by its visitors. Until 1934, when the new Catholic totalitarian government closed down the centre, just 5000 individuals benefitted from its services. While eugenic counselling was obligatory for those seeking marriage during the National Socialist reign, concepts of counselling outlived World War II in the Catholic marriage counselling. The tradition of marriage counselling reemerged in Vienna in 1956, when marriage counselling service at municipal level was reestablished and interpreted as successor service of the 1920s.[15] To what extent Catholic marriage counselling included eugenic and genetic counselling remains an open question. However, the renowned Catholic gynaecologist Albert Niedermeyer (1888–1957) advocated eugenic counselling as a part of Catholic marriage counselling in his influential textbook on pastoral medicine in 1950.[16]

Until up to the end of the 1960s, genetic counselling was often termed eugenic counselling. For example, on the occasion of the establishment of the European Society for Human Genetics in 1967, human genetics was perceived as applied knowledge for the goals of "eugenic counselling" among Austrian physicians.[17] And in 1965 the eugenic society "Association for Voluntary Hereditary Care (Human Genetics)" ("Verein für freiwillige Erbpflege (Humangenetik)") encouraged the establishment of genetic ("erbbiologische") counselling services in all nine capitals of Austrian federal states.[18] However, during the 1970s, "eugenic counselling" was termed more often genetic or human genetic counselling—now labelling the new discipline and shaping distance to the past: all genetic counselling services established in 1974 named themselves genetic or human genetic, but not eugenic.

3 The Practice of Genetic Counselling in the 1970s

What objectives did practitioners themselves pursue with genetic counselling? The origins of the genetic counselling service were seen differently by their practitioners. While some directors of counselling services emphasized only recent developments in human genetics and society and especially the new counselling service of Gerhard Wendt in Marburg, others argued for the continuation of a tradition of social democrat counselling of the 1920s, and finally some anthropologists emphasized the Nazis as their forerunners. The latter happened at the "Human Genetic Family Counselling Service" that was established in July 1974 at the anthropological department at the *Museum of Natural History* in Vienna.[19] The service could be visited for free and lasted at least 2 years. It was part of the "anthropological-hereditary-biological counselling service", where

[15]Österreichische Ärztezeitung 1956, 11 (1): 32.

[16]Niedermeyer 1950.

[17]Österreichische Ärztezeitung 1968, 22: 338.

[18]Mayer 2010.

[19]Szilvássy 1982, 133.

genetic research was conducted and where next to genetic paternity tests for civil courts, still racial science of genetic traits of African people was performed.[20] The genetic counselling service was seen in tradition of genetic counselling of the Nazi period in Vienna, when the visit of the service was enforced for those seeking financial support for marriage.[21]

All counsellors agreed that the services were part of preventive medicine and rational family planning when the birth rate was declining.[22] Like in Germany[23] the talk of risk prevention was widespread among early Austrian counsellors.[24] Some argued that some genetic diseases were not as rare as thought, and therefore genetic counselling was needed.[25] The number of 4% of damaged offspring was used to legitimize the existence of the genetic counselling service.[26] As no genetic therapy was available, rational family planning was the aim of genetic counselling.[27] Some counsellors promoted genetic counselling even to the "cornerstone of preventive medicine".[28] Others disagreed in this respect. Walter Vormittag, genetic counsellor, internist and clinical geneticist at the second medical clinic in Vienna, considered genetic counselling to a lesser extent responsible for preventive medicine, but to serve the individual case. Therefore, genetic counselling was rather a task for physicians than for geneticists.[29]

All consulters agreed that the relation of consulter and counselee was one of a doctor-patient relation, although some counsellors reflected the fact that some counselees were not sick themselves, but just carrier of genetic aberrations.[30] According to the service in Graz, counselling was aimed for people who were afraid to have a disabled baby or a genetic disease themselves.[31] Rett considered the aim of counselling to determine the "Wiederholungsrisiko" (risk of repetition) within a family of a "damaged" child. The procedure of counselling included an initial talk with family anamnesis, clinical diagnostics, prenatal testing, cytogenetic analysis and the final counselling interview. This final talk was considered the most important, because it was to guide the counselee to the right decision, even though the final decision remained with the counselee.[32] However, the final talk included

[20]Anonymus 1975, XL; Anonymus 1976, XLVII.

[21]Mayer 2015a, 324.

[22]For example, Rosenkranz 1974, 18; Vormittag 1974, 880; Schnedl 1976, 61–62; Schnedl 1977, 326–27; Rett 1977, 510.

[23]Waldschmidt 1996.

[24]For example, Rett 1978; Vormittag 1980; Schnedl 1977, 327.

[25]Schnedl 1977, 326.

[26]Schnedl 1977, 326.

[27]Schnedl 1977, 326.

[28]Schnedl 1976, 94.

[29]Schnedl 1976, 94.

[30]Vormittag 1974; Vormittag 1980.

[31]Österreichische Ärztezeitung 1978, 33: 60.

[32]Rosenkranz 1974, 20; Schnedl 1977, 326.

advice on abortion or the abdication of children: For example, the Graz service advised to get no more kids when the genetic risk was higher than 10%.[33] Rett advised abdication of further offspring in case of already born "severe-damaged" children. He also demanded from the counselee that she must agree to an abortion before conducting an amniocentesis, when it was not clear whether Rett was authorized to make this request.[34]

Different opinions existed on the aims of the genetic counselling in regard to the individual and/or society. Which one was the service to serve? Rett, for example, considered genetic counselling to serve the individual and the society at the same time, while Walter Rosenkranz in Graz emphasized the importance of the health of the individual and its family over enhancing the quality of a population.[35] Rosenkranz headed the first genetic counselling service in Austria, which was established in July 1974 with the support of the Austrian Ministry of Health.

The counselling services adopted special fields of counselling according to the research focus of the attached departments. For example, cases of expected trisomy 21 were mainly sent forward to the clinic of child neurology of Andreas Rett, when diagnostics of inborn errors of the metabolism were done at the internal clinic, and women aged 35+ were directly sent to centres for amniocentesis.[36]

The emergence of genetic counselling supported the view of human genetic knowledge as applied knowledge, rather than theoretical. We want to argue that this view indicated the importance of human genetic knowledge for the needs of modern society. The above-mentioned eugenic indication of abortion was acknowledged as an important precondition to counselling.[37] The law on abortion enabled plenty of scope for interpretation: an abortion was indicated, when "a serious threat exists that a physically or mentally severe-damaged child would be born" ("eine ernste Gefahr eines schweren körperlichen oder geistigen Schadens für das Kind besteht, wenn es geboren würde"). In the expectation of the counsellors, and obviously also in their experience, this scope was to be defined by the practitioners themselves.[38] A public discussion on the topic, which diagnosis was to be affected by the eugenic indication of abortion, did not occur during these years.

The experience of counselling during the 1970s showed that many counselees accepted prenatal diagnostics with the chance of interruption of the pregnancy, while only few, religiously motivated people refused this option.[39] Explicit advice was given for abortion in certain cases. Vormittag, for example, gave advice for

[33]Rosenkranz 1974, 20; Schnedl 1976, 94.
[34]Rett 1978.
[35]Rosenkranz 1974, 18.
[36]Vormittag 1980.
[37]Vormittag 1974, 880.
[38]Vormittag 1980, 1334.
[39]Vormittag 1980, 1334.

abortion in case of teratogenic errors. Abortion was considered indicated in the case of alcoholism as there was a 40–50% chance for embryopathy. Abortion was also advised in cases of exposure to radiation of 5–10 rem during the first trimester of pregnancy or in case rubella during the first 4 months of pregnancy. No teratogenic effect was seen by smoking during the pregnancy.[40] Abortion was also advised in cases of X chromosomal disorders, when there was a 50% chance that sons were affected.[41]

However, during the first years of existence, the counsellors were not quite satisfied with the attendance in the beginning: Graz handled 350 inquiries during the first year and advised in 25% of the cases to get no further children.[42] The service at the Vienna internal clinic realized 400 inquiries during 5 years of service.[43] Rett counselled approximately 300 cases each year in 1979. In the Western part of Austria, chromosome analyses were performed on 1800 people from 1972 to 1981, when the genetic counselling centre finally was established in Innsbruck.[44] Amniocentesis was established as a standard procedure in reproductive medicine in Austria by the end of the 1970s[45] and advanced to one key method for genetic counselling. In Graz human geneticists and gynaecologists argued in 1981 that amniocentesis saved many pregnancies as well as the method saved society from the costs for the care of disabled people. However, the scholars neither mentioned abortion nor the eugenic indication of abortion as a key tool for genetic counselling.[46] Possibly, the more genetic counselling was performed and socially accepted, the less it was labelled as eugenics.

3.1 A Story of Success? Human Genetics as a Discipline at Austrian Universities

Academic counsellors considered the rise of human genetics as a discipline of medicine to be connected with the rising need for genetic counselling.[47] So, the rise of genetic counselling raised the need of institutionalization of human genetics at university. In the beginning of the 1970s, Austria's only human genetic institution was based in Graz at the Department of Medical Biology. The eugenic solution of abortion and the expectation of applied human genetics as genetic counselling services encouraged in 1973 several initiatives to establish academic departments

[40]Vormittag 1980, 1332.
[41]Vormittag 1980, 1334.
[42]Rosenkranz 1975.
[43]Vormittag 1980, 1335.
[44]Schröcksnadel 1982, 204.
[45]Wolf 2008, 615.
[46]Zierler 1981, 79.
[47]Schnedl 1977, 326.

for human genetics at Austrian faculties of medicine. At the faculty of medicine at the University of Vienna, Austria's biggest faculty, the establishment of a Department of Human Genetics failed in the mid-1970s, maybe due to competition among the involved human geneticists. Ironically, the Ministry of Education had encouraged the foundation of human genetics already 10 years earlier in the mid-1960s, when the Vienna faculty of medicine was asked to name proper candidates. Probably the Austrian ministry followed the Western German example, where the Atomic Commission emphasized the establishment of human genetics at academic level to study genetic mutations of atomic testing during the Cold War.[48] However, in Vienna in 1965, the faculty abandoned the idea as unnecessary, because animal genetics was established at the faculty, and therefore no special human genetics was needed. Issues of academic competition had played a major role for this judgment. Finally, the establishment of genetic counselling did not account for new departments of human genetics in the 1970s.

4 Conclusion

The emergence of genetic counselling services in 1974 benefited from the eugenic indication of abortion, which was enabled in 1975 due to the reform of the penal law. Counselling offered mainly abortion as a solution to genetic or chromosomal aberration, because no genetic therapy was available at the time. Since the 1950s the eugenic indication of abortion was discussed in the drafts for the penal law in Austria by various groups, which continued debates from the 1920s. In the 1970s some counsellors considered the implementation of the eugenic indication in the penal law as an important condition for the "therapeutic" effect and the success of the genetic counselling.

Vienna's tradition on eugenic counselling started already in 1922, when the first municipal marriage counselling service was established in "Red Vienna". However, the practice of this early counselling demonstrated that not eugenic but sexual advice was sought by its visitors. While eugenic counselling was obligatory during the National Socialist reign, concepts of counselling outlived World War II in the Catholic marriage counselling. After 1945 genetic counselling in Austria was understood as eugenic counselling until the end of the 1960s.

Furthermore, we argued in this chapter for a close relation of the professionalization of human genetics, the establishment of genetic counselling and the establishment of the eugenic indication of abortion during the 1960s and 1970s. Concepts of counselling in Austria supported the individual and the society at the same time. While the term "eugenics" seemed to vanish from the scientific discourse during the 1970s, we have demonstrated that some counsellors like the child neurologist Andreas Rett still supported eugenic reasoning in the early practice of

[48] Kröner 2002.

counselling. The eugenic indication of abortion remained an important possibility for the outcome of a counselling interview.

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Genetic Counselling in Belgium: The Centre for Human Genetics at the University of Leuven, 1960–1990

Joris Vandendriessche

Abstract This chapter traces the history of the Center for Human Genetics (CHG)—the first centre of this kind in Belgium—at the University of Leuven from the 1960s to the 1990s. In 1960, a laboratory for diagnostic chromosomal research was set up by the physician and geneticist Herman Van den Berghe. In 1966, this laboratory was turned into the Center for Human Genetics (CHG), which combined a service of genetic counselling with genetic research. The paper discusses the evolution of the Leuven CHG in relation to developments within the Faculty of Medicine and the University of Leuven, to government policies on genetics and to wider social debates. The CHG's expansion in the 1970s, 1980s and 1990s was paralleled by governmental attention to the field of human genetics and the life sciences. State support was allocated to eight genetic centres, which—following the Leuven model—were integrated into the Belgian academic hospitals, resulting in a decentralised model. This system of financing contributed, it will be shown, to the multidisciplinary nature of genetic research and counselling in Belgium. The paper also pays attention to contemporary ethical debates about medical technologies, of which genetic diagnoses were part. While these debates were conducted nationwide, they were particularly present at the University of Leuven, as the institution struggled to reconcile its Catholic heritage with its modern research ambitions.

Keywords History of medicine • Genetic counselling • Academic medicine • Medical ethics • Science policy • Belgium

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1 Introduction

Genetic counselling in Belgium is thoroughly integrated in the country's academic hospitals. The state support for genetic clinics and research that developed since the 1970s was allocated to eight genetic centres, located at the Belgian universities. They received governmental subsidies to conduct genetic tests—a measure that was coupled to strict quality control, the need to conduct scientific research, to offer psychological guidance to patients and to inform the public about the implications of genetic testing and research. Unlike in other countries, where 'genetic counsellors' developed as a professional medical subgroup, genetic counselling in Belgium is performed by physicians, often paediatricians. From the start, and increasingly with the availability of new genetic tests, they collaborated closely with other (medical) specialists, such as gynaecologists, psychiatrists, psychiatric nurses, psychologists, remedial educationalists, etc. As a result, Belgian genetic counselling has gained a strong multidisciplinary outlook—a second particular feature—which has reinforced its integration within academic health centres.

The recent history of genetics in Belgium remains to be written.[1] This chapter is intended as a first step in unravelling the field's historical trajectory by focusing on one centre—the *Centre for Human Genetics* (CHG) at the University of Leuven, which played a central role in the development of the integrated and multidisciplinary Belgian model. In 2016, the Leuven CHG celebrated its fiftieth anniversary with lectures from international researchers, a formal academic session, a conference on 'Genetics and Society', a series of postgraduate lectures and a visit of the Belgian queen.[2] If today around 350 people work at the CHG, in particular in its major research and diagnostic laboratories, only a handful of members made up the staff 50 years earlier, as a *Laboratory of Cytogenetics* was established as a separate unit within the *Department of Human Biology* in 1966 and a separate clinical service was started in the Leuven academic hospital. Its history, however, may be traced back even further. In 1960 already, a one-man laboratory, run by Herman Van den Berghe, provided diagnostic chromosomal research to patients transferred from other clinical services.

The focus of this chapter is not so much on the considerable expansion of the scientific research and genetic tests performed at the *Centre for Human Genetics* in Leuven, which have been treated in several retrospective brochures.[3] My aim is rather to place the centre in a wider historical context and discuss its trajectory in

[1] While the post-WW II history of genetics in Belgium has been hardly studied, some work has been done on genetics in the interwar years: De Bont, 2007; De Raes, 1989.

[2] For an overview of the activities organised to celebrate the CHG's fiftieth anniversary and the current organization of the centre: http://gbiomed.kuleuven.be/apps/cme/.

[3] Three retrospective brochures have been published by the Leuven *Centre for Human Genetics* which allow a look into the centre's developing research activities. The first was published in 1987, when the CHG moved to the new medical campus of Gasthuisberg; the second in 1998, when Herman Van den Berghe retired as the centre's director; and the third in 2016, when the CHG celebrated its fiftieth anniversary.

relation to developments within the Faculty of Medicine and the University of Leuven and more generally to government policies on genetics and to wider social debates.[4] In this light, the embeddedness of the CHG in a Catholic academic institution has been essential. Particularly since the (late) nineteenth century, the University of Leuven emphasised its position as one of the major Catholic academic centres worldwide. This also meant that the ethical questions raised by genetic testing and research were answered in a particular setting, even though the influence of the Catholic Church over the University—in a secularising society—has equally been greatly diminished since the 1960s.[5] To open up this history, several interviews with (former) members of the CHG and with colleagues from other clinical services were conducted.[6] These have been combined with articles from the university's journal and from the Belgian general press.

2 The Politics of Genetic Research

The first support for genetics in Leuven after the Second World War was part of the University's programme of cancer research. Gerard Van der Schueren (1908–1978), originally a Professor of Anatomy who had become head of the *Radiotherapy Service* in 1952, followed the international research on the relation between cancer and heredity in the 1950s and 1960s closely. For his private archive, he cut out an article from *Time Magazine*, which reported of President Kennedy's support for researchers of mental retardation.[7] Another article he collected, now from a scientific journal, the *Belgian Review of Experimental Pathology*, was authored by Leonell Strong, who had presented his hereditary theory on cancer at a conference in Brussels, funded by Belgium's National Cancer Foundation.[8] While Van der Schueren himself had little time for research—he was also the director of the academic hospitals in those days—he seems to have realised the potential of genetics for cancer research and supported two young physicians, Herman Van den Berghe and Herman Verresen, who were interested in the subject. With them, he published several papers to promote genetics in Belgium. In of these papers—titled 'the promising morphological branch of

[4]The research for this chapter is part of a larger book project on the history of the Leuven academic hospitals, which will place the evolution of these hospitals since the 1920s within a wider social and cultural context.

[5]For a recent history of the University of Leuven: Tollebeek and Nys, 2006.

[6]These interviews were conducted together with Liesbet Nys in the course of 2015 and 2016. Among the interviewees were Herman Van den Berghe, Bart De Strooper, Eric Legius, Gerry Kiebooms, André Van Assche, Paul Schotsmans, Bassem Hassan, Peter Marynen, Vanessa Morais, Annemarieke Sierksma and Iryna Voytyuk. In 2007, Peter Harper also conducted an interview with Herman Van den Berghe which is publicly available on the website of the Genetics and Medicine Historical Network: www.genmedhist.org/interviews.

[7]University Archive of Leuven [from here on: UAL], Archive of Gerard Van der Schueren [from here on: AVdS], N. 159, clipping: 'Chromosomes & the Mind', 1962.

[8]UAL, AVdS, N. 159, clipping: Strong, 'Une théorie génétique', 1949.

cytogenetics'—they explained the recent advances in the research on human chromosomes and called for clinical research within paediatrics (to trace hereditary disorders) and oncology (to study the effects of radiation on tumours).[9] Both topics would become central to genetic research in Leuven.

While Herman Verresen would develop his medical career at the university's campus of Kortrijk from the late 1960s, Herman Van den Berghe became the key figure in the development of genetics in Leuven. Born in Geraardsbergen in 1933, Van den Berghe obtained his medical degree at the University of Leuven in 1958. During the 1950s, the most talented students were often recruited to work voluntarily in one of the laboratories at the Faculty of Medicine. Van den Berghe was also encouraged to assist the staff at the *Vesalius* and *Rega Institutes*, where he learned the techniques of tissue culture and virology (e.g. titrating viruses). He also worked voluntarily at the Cancer Institute in Amsterdam, assisting in research on the role of hormones in breast cancer by conducting experiments on mice. As scientific work on human chromosomes seeped through into the Leuven medical world, Van den Berghe familiarised himself with chromosomal analysis and set up a small laboratory in the basement of the Vesalius Institute.

At the Faculty of Medicine, such new research laboratories were increasingly encouraged. A new generation of Flemish (Dutch-speaking) faculty members, including the internist Jozuë Vandenbroucke (1914–1987) and the microbiologist Pieter De Somer (1917–1985), who had founded the *Rega Institute* in 1954, supported the development of new subdisciplines. For Vandenbroucke, subspecialisation formed a means to strengthen the interplay between clinical work and research in the medical sciences; De Somer, who had a background in virology, called for more fundamental research at the Faculty of Medicine. Both agreed that the existing structures within the faculty were unfit to enable innovative research. They advocated the creation of 'departments', through which the research funds of the Faculty of Medicine would be better divided.[10] Both also merged this agenda of reform with the ambition of improving the position of Dutch-speaking physicians in the Leuven research laboratories, which hitherto had been dominated by their French-speaking colleagues. Linguistic tensions would eventually lead to the splitting-up of the University of Leuven in 1968—a major event in Belgium's political history and one in which the Leuven physicians played a key role.[11] De Somer became the first rector of the now Dutch-speaking university. The French-speaking physicians moved to the newly built hospital complex *Saint-Luc* in Sint-Lambrechts-Woluwe, near Brussels, where an independent service of genetic counselling for French-speaking patients would later be developed. Their gradual departure from the buildings on the medical campus in Leuven's city centre

[9]UAL, AVdS, N. 159, draft paper: Van der Schueren, Gerard, Van den Berghe, Herman and Verresen, Herman, 'De cytogenetica: een veelbelovende morfologische tak'.

[10]Nys 2016, 94–100.

[11]Vandendriessche and Nys 2017.

provided Van den Berghe with additional space to expand the Dutch-speaking CHG.

Parallel to the more positive climate for specialised research in the Faculty of Medicine, the clinical function of chromosomal research within the Leuven academic hospitals became clear. By 1960, Van den Berghe became regularly contacted to assist in the diagnosis of disorders related to the sex chromosomes from the service of endocrinology and gynaecology. Other types of patients that were referred to him from early on were those treated for myeloid leukaemia by the Leuven haematologists. Moreover, also outside of Leuven, the laboratory's diagnostic work became known. Blood samples from patients in regional hospitals were sent to Van den Berghe for analysis. This clinical potential made that in 1966, when the departmental structure was finally introduced in the Faculty of Medicine, Van den Berghe's *Laboratory of Cytogenetics* was not only integrated in the *Department of Human Biology* as a research laboratory. Simultaneously, an independent clinical service was set up within the hospital, which would be expanded by Jean-Pierre Frijns. Both components together formed the *Centre for Human Genetics* (CHG).

The further expansion of the CHG followed swiftly as the centre sought affiliation with geneticists on the international level. Van den Berghe himself had conducted an internship at the *Galton Laboratory* in London with Lionel Penrose (1898–1972) in 1962 and received further training in genetics in Paris and Seattle. In the 1970s, several staff members, including Jean-Jacques Cassiman, Fred Van Leuven, Guido David and Peter Marynen went abroad for research stays (e.g. to Stanford University), bringing back expertise in different types of genetic research to Leuven. This allowed the research activities of the CHG to expand rapidly in different directions (e.g. cancer research, somatic cell genetics and forensic genetics). Robert Vlietinck developed a research group in population genetics, which cooperated with the University of Ghent in studying identical twins. It was 'a golden speedway for genetic research', in Vlietincks' words.[12] Since the middle of the 1960s, a twin register had been kept in Ghent, which was later continued by the Leuven CHG. In the genetic clinic, much research was conducted on X-linked mental retardation by Frijns and later Eric Legius, for which they cooperated with numerous institutions for the mentally disabled, examining thousands of patients over the years. On the basis of these data, they were able to identify and describe several new chromosomal syndromes.

The expansion of these research activities did not only increase the centre's academic staff. From its early years, laboratory assistants—mostly women—made up the largest group of personnel. This gender balance between the technical and academic personnel would only gradually alter in the 1980s. The psychologist Gerry Kiebooms became the first woman to be appointed officially in the centre's academic staff in 1987. Most of this personnel was paid with research funding from the Faculty of Medicine. In addition, the CHG also became a successful candidate in different competitive research programmes, both on the level of the university

[12] 'Tweelingenonderzoek', 1987.

and on the national level (e.g. the National Cancer Funds). Even more important was the Belgian Fund for Medical Research, which had been established in 1957 as part of the country's health politics and which was financed by the Ministry of Public Health. Increasingly, the existing mix of public and private (industrial) funding for science became complemented with larger topical research programmes from the state—it meant the hesitant introduction of 'Big Science' in Belgium. Medical research was among the strategic areas, in which the state ambitioned to take a leading role. It formed an important stimulus for the CHG's expansion.[13]

Shifts in Belgium's science policy since the late 1980s further impacted the expansion of the CHG's research activities. The Belgian state reforms of 1988 transferred most of the decision-making on state support for science to the regional level (e.g. the Flemish government). Even though the mentioned Funds for Medical Research provided certain means, subsidies for scientific research still compared poorly with similar policies abroad. When new budgetary cuts were proposed in 1992, a national march was organised in Brussels, to which 10,000 researchers participated. The protest was organised by Research Focus, an organisation founded in 1986 that aimed to promote scientific research in Belgium. Two young medical researchers—Bart De Strooper, a post-doc at the CHG, and Patrick Callaerts—played a key role in the initiative, which was supported by the university board.[14] The 1992 march proved a turning point in Flemish science policy. More funds were now allocated to the Flanders Research Foundation, the regional successor of the National Science Foundation of which the Fund for Medical Research had been part. More attention was also paid to the support of biotechnological research, which became one of the strategic areas in which the Flemish government invested. In 1990, a first programme (VLAB) had been started to support research in the life sciences. In 1996, a much larger initiative was taken with the foundation of the VIB, a life sciences research institute, heavily funded by the Flemish government. Van den Berghe was one of the advocates of this new institute, which, it is important to stress here, did not develop as an independent organisation but was fully integrated into the universities.[15] The CHG became one of the two Leuven 'core departments' of the VIB.

The funding from the VIB marked a new phase in the development of the Leuven CHG. Government attention for the life sciences had stimulated researchers such as Bart De Strooper to shift their focus to diseases as Alzheimer and Parkinson, for which major research programmes were now set up. The bibliometric and result-based evaluation that came along with VIB-funding, some researchers have indicated, equally caused shifts in the centre's scientific culture. The merit-based system, it has been said, broke down the hierarchical structures and increased the autonomy of researchers. Another development that was reinforced by VIB was the

[13]Halleux, 2015, 110–113.

[14]De Strooper and Callaerts, 1992. For a more elaborate discussion on the topic: Nys, 2016, 189–200.

[15]Ibid., 238–241.

internationalisation of the community of researchers at the CHG, which became an attractive work place for promising foreign researchers, some already with a high reputation. They, in turn, contributed to grant proposals on the European level, setting in motion a further process of expansion.

3 Integration into the Hospital

One of Van den Berghe's favourite sayings about the position of the CHG within the Leuven academic hospitals was *pour vivre heureux, vivons caché* [to live happily, live hidden]. It seems no coincidence that this phrase surfaced in many of the interviews with the centre's (former) members, not just as a recollection of Van den Berghe's style of leadership but also as a reflection of the particular nature of the genetic clinic. Unlike other clinical units, the CHG did not have its own hospital beds—an essential marker in hospital organisation. Neither did genetics constitute an essential part of the training of medical students. As many recalled afterwards, in its early years, the activities of the CHG were little known by other physicians in the hospital. Its original location, hidden in the basements of the *Vesalius Institute*, did not contribute to the centre's visibility either. The CHG would later expand to old buildings of the University's technical services in the De Croylaan. In the late 1980s, it moved to the research buildings 'O&N' on Gasthuisberg, the new medical campus outside the city centre.

While the CHG's 'hidden' position in the hospital was given multiple meanings by the interviewees, the argument of the centre's financial autonomy often recurred. The centralised hospital administration originally had little control over the budgets of each clinical service, giving these services considerable autonomy—it was the time of the 'far west' in the academic hospital, as some recalled. The genetic clinic indeed secured its own financing through different governmental channels. Since the late 1960s, a politics of subsidising genetic tests and research into hereditary diseases was launched. In 1968, a law was voted that recognised and supported centres that conducted diagnoses of phenylketonuria (PKU), a hereditary metabolic disorder. It was inspired by the fact that PKU 'entailed a serious mental retardation if it was not traced timely.'[16] Five years later, in 1973, a *High Council for Human Genetics* was founded which was to stimulate the development of genetics in medical and social ways. It was also to add to programmes of prevention and registration.[17] The director of the division 'social medicine' at the Ministry of Public Health became president of the council and Van den Berghe its vice president. Together with this council, an *Interfaculty Institute for Human Genetics* was set up through which additional governmental subsidies were divided to seven

[16] 'Opsporing van fenylcetonurie', 1968.

[17] 'Hoge Raad voor de Antropogenetica', 1973.

(later eight) genetic centres, located at the Belgian universities.[18] Moreover, the different laboratory tests conducted at these recognised centres were reimbursed by Belgian social security.

The isolated position of the CHG within the Leuven hospitals, at the same time, has to be nuanced. Van den Berghe's saying was most of all applicable to the centre's early years. An evolution towards more collaboration between different clinical services, for example, by organising multidisciplinary consultations—an evolution not limited to the field of genetics—paralleled the growth of the CHG in the 1970s and 1980s. Jean-Pierre Frijns started joint prenatal consultations with Kamiel Vandenberghe (1940–1997), a member of the *Gynaecology and Obstetrics Service*, who became known for having introduced and spread ultrasound imaging in the hospital. Many couples with anxieties over hereditary defects, who had often already been informed by the CHG, found their way to these consultations. Techniques such as ultrasound and amniocentesis became more common—the latter practised at the *Gynaecology and Obstetrics Service*, but always in the presence of a laboratory assistant of the CHG, who transported the amniotic fluids to the laboratory for analysis. Other multidisciplinary consultations were set up in collaboration with paediatric neurologists and cardiologists, for example, on neuromuscular disorders and on specific hereditary diseases such as neurofibromatosis, on which Eric Legius was the centre's specialist.[19]

With the availability of new genetic tests since the 1980s, the patient potential of the CHG increased dramatically. More and more physicians were added to the staff of the genetic clinic (which today consists of eight full-time positions). Moreover, a network with ten regional hospitals was set up, the Leuven geneticists travelling to each of these hospitals once a month to organise consultations. By 1993, almost 5000 patients got into touch with the centre each year. Among them are older couples, who desired to have children and inquired after possible risks of pregnancies at a later age; patients of whom a family member carried a genetic defect and who sought to know whether or not they carried the gene as well; and, with the discovery of genes that increased the risk of breast cancer or heart disease, patients interested in all sorts of risk assessment. With these new tests, as Jean-Pierre Frijns explained in an interview, the practice of genetic counselling developed as well. The international guidelines of 'non-directiveness' in assisting patients' decision-making were stressed at the Leuven centre. Good counselling was needed, Frijns concluded, in order for 'our technology to be at the service to those who wish to make a decision.'[20]

[18]In a budget report of 1973 of the Belgian senate, such an interfaculty institute is mentioned: http://www.senate.be/lexdocs/S0642/S06421172.pdf (Consulted on January 29, 2017), p. 16. A subsidy of 25.000 F was granted. On the website of the Antwerp Centre for Human Genetics, a brief history of the centre is included: http://www.uza.be/over-het-centrum-medische-genetica-cmg (consulted on January 19, 2016).

[19]For an overview of these consultations in the 1990s: Centrum voor Menselijke Erfelijkheid, 1998, 72.

[20]'Klinische genetica', 1993.

The psychological effects of genetic testing on patients, from the possibility of taking them to the prospect of future illnesses, became itself also subject to research. In 1977, Gerry Kiebooms had joined the CHG and since the late 1980s developed a unit of 'psychosocial genetics' within the centre. Clinical work formed part of this. The availability of new genetic tests, for example, for Huntington's disease, Kiebooms recalls, generated a fear of possible suicides by those who would test positively, hence, the need for psychological guidance—a necessity for which cooperation was sought with patient organisations. In collaboration with the Leuven psychiatrists, a shared consultation was set up on this subject, informing families about the effects of such tests and counselling them also after the results were made available. Using questionnaires, here as well clinical work was coupled to research.

Belgian legislation on genetic counselling stressed this need for psychological guidance and correct information. In 1987, a Law on Genetic Centres was voted in the Belgian parliament, which expanded the older system of subsidies and established the criteria genetic centres had to meet to receive financing. Offering counselling to patients, allowing them to make informed decisions, and informing the public about the developments of genetic research were part of this. The combination of clinical service and medical research was also stressed: the different centres were to study whether or not abnormalities, both mental and physical, were hereditary or not, concerning the nature of these defects and all elements related to being a carrier of these hereditary features.[21] The law was realised by Wivina De Meester, a politician from the Christian People's Party who was State Secretary of Public Health between 1985 and 1988. De Meester knew Herman Van den Berghe well, who advised her in debates over abortion. When the Leuven CHG moved to the new campus of Gasthuisberg in 1987, the same year as the new legislation, a solemn academic session was organised during which De Meester explained her policy and suggested the need to invite certain high-risk groups for genetic screening in the future.[22]

After 1987, the CHG employed a more proactive strategy, investing more means and energy in 'genetic education' and in explaining its workings to the outside world. It regularly organised an open house, during which one could visit its laboratories on the sixth floor of the Gasthuisberg research building. Educational films, books and booklets were released in which questions of genetic effects were explained. Members of the centre, moreover, also increasingly appeared in the media. In debates about AIDS, fertility, paternity tests among other subjects, the CHG clearly fulfilled a public role. This more public presence of the centre also illustrates the increased social importance of genetics in recent years, of which the ethical boundaries have equally become subject of public debate.

[21] See the text of the Law on Centres of Human Genetics of December 14, 1987: http://www.ejustice.just.fgov.be/cgi_loi/change_lg.pl?language=nl&la=N&cn=1987121432&table_name=wet.

[22] 'Grote belangstelling', 1987.

4 Ethical Debates

Such ethical discussions were by no means new. At the Faculty of Medicine, much attention had gone to medical ethics since the 1960s. It was a means of engaging with contemporary clerical views on matters of reproduction, which seemed at odds with modern medical techniques. In 1968, Pope Paulus VI (1897–1978) had condemned 'artificial' forms of birth control in his encyclical letter *Humanae Vitae*. Despite this condemnation, the prescribing of contraceptive pills was continued in the academic hospitals and defended by the Leuven physicians as means to prevent abortion. In 1975, a *Commission for Medical Ethics* was founded—the gynaecologist Marcel Renaer (1913–2006) became its first president—that was to formulate advice on ethically sensitive issues such as sterilisation and artificial insemination. The ambition, which was supported by the Belgian bishops, was to establish a responsible, yet modern, 'Catholic' position on these matters. The commission's advisory role was later supplemented with the need for research in medical ethics and its inclusion in medical education. Following an American model, a *Centre for Bio-Ethics* was founded at the University of Leuven in 1986. Paul Schotsmans became the centre's first director and the first full professor of medical ethics at the Faculty of Medicine—courses in medical ethics having previously been taught by philosophers.[23]

Many of these ethically sensitive issues were foremost a matter for the Leuven gynaecologists. One of these was abortion, which—until a Law on Abortion was voted in 1990—was considered a criminal offence under all circumstances, following Belgium's penal code of 1867. In the 1970s and 1980s, Belgian women seeking an abortion hence travelled to the United Kingdom or the Netherlands, where legislation had been voted much earlier than in Belgium. The 1990 Law was voted in the Belgian parliament without the support of the Christian People's Party and made abortion legal until 14 weeks of pregnancy if performed in a recognised abortion centre. Against the background of an ongoing public debate on such legislation in the 1980s, prenatal diagnoses and fertility treatments were topics that evoked considerable attention in Catholic academic circles.[24] As historian Liesbet Nys has recently shown, the introduction of in vitro fertilisation—the first Belgian test-tube baby was born in Leuven in 1983—caused a disagreement between the board of the Faculty of Medicine, who feared to offend the ecclesiastical authorities and the gynaecologists who had used the technique.[25] If not always in a direct way, the *Centre for Human Genetics* was nevertheless involved in these debates. The chromosomal analyses conducted in its laboratories, such as the examination of amniotic fluids since the early 1970s, it was rumoured by some, led to an increasing number of abortions, for which women often travelled abroad. In case of severe genetic defects, or when the mother's health was under threat,

[23]Nys 2016, 143–145, 209–211.
[24]Witte 1993.
[25]Nys 2016, 147–150.

abortion was practised in the Leuven academic hospitals, also before 1990. Some of these rumours, according to which the Belgian bishops were furious about certain 'unethical' practices in the CHG, reached Van den Berghe, who remembered gaining their trust after a meeting in which he explained the centre's functioning.

From the late 1980s onwards, the CHG became itself more involved in these ethical debates. At different occasions, Van den Berghe emphasised the social challenges that arose from the availability of all sorts of new (prenatal) genetic tests, pleading for debate and public education on these matters.[26] Within the *Centre for Bio-Ethics* as well, the implications of genetic testing were increasingly investigated, certainly after the publication of another encyclical letter, *Donum Vitae*, in 1987. The new letter stipulated that prenatal diagnoses could only be conducted if they were oriented towards healing and with respect to the 'integrity of the human foetus'. They were 'in opposition to moral laws' if they provoked abortions. In 1988, members of the University of Leuven's leadership, including rector Roger Dillemans and Guido Maertens (1929–2002), who taught medical ethics at the university's campus in Kortrijk, visited the Vatican as part of a delegation of Catholic universities to explain the medical procedures in their hospitals. While they did not succeed in altering the Church's view on the topic—in particular the creation of surplus embryos was regarded highly problematic—no conviction by the Church followed either.[27]

In the early 1990s, shortly after the mentioned Law on Abortion, the activities of the CHG were nevertheless looked at with Argus' eyes. In 1992, the papal nuncio, a diplomatic representative of the pope in Belgium, contacted the Leuven rector Dillemans after hearing rumours that genetic tests led to abortions, reminding him of the encyclical letter *Donum Vitae*. Dillemans assured the nuncio that no clear genetic advice was given, but only information, and that genetic counselling was done in 'a warm, humane and non-directive fashion' to make sure that the patients understood all aspects well. When asked for their opinion, Dillemans argued, the geneticists emphasised the respect for human life, including the life of the disabled. At the end of his letter, however, he added that 'Unfortunately, the analysis made by people with this information [. . .] is no longer in line with this traditional Christian value that gives meaning to suffering and sacrifice'. With the secularisation of Belgian society came indeed a shift in morality, of which Catholics such as Dillemans were critical. With genetic testing, hereditary defects were easier to prevent, contributing to a society in which the Christian-inspired meaning of suffering was eroded. Those with such hereditary defects, it was feared, might be less cared for and understood. For this reason among others, Paul Schotsmans pleaded for an 'ethical framework' for genetic counselling.[28]

Schotsmans did not stand alone with these views. In the 1990s, questions of genetic testing featured prominently in ethical debates. This was, for example, the

[26]Van den Berghe 1986.

[27]Maertens 1988.

[28]Schotsmans 1998.

case at the *Overlegplatform Christelijke Ethiek* [Platform for Christian Ethics], of which Maertens was president, and in the organisation's journal *Ethische Perspectieven* [Ethical Perspectives]. One of the most delicate issues was the matter of surplus embryos, created during in vitro fertilisation, and their possible use for experimental research. For Maertens, such use was out of the question (a view he had also defended during the meeting with Cardinal Ratzinger in Rome in 1988).[29] Van den Berghe, however, in a double dialogue with Maertens in the University's journal, which was picked up in the general press, had declared that such research for him was justified. The fabrication of human embryos for scientific research he too condemned fiercely.[30] Van den Berghe's statement caused a storm in the media, during which he, as the director of the CHG, epitomised the 'modern' genetic research and its far-reaching consequences. The public debate showed how, after the abortion debate, genetics moved more into the centre of the public debate on medical ethics. The new techniques of pre-implantation genetic diagnosis (PGD) in the 1990s, which allowed genetic testing of embryos prior to implantation, further fuelled these debates.

5 Conclusions

This brief survey has indicated some links between the development of the *Centre for Human Genetics* at the University of Leuven and wider shifts in government policy, clinical care and ethical debates in Belgium. While it does not allow drawing any definitive conclusions, it has rendered some insight into the particularity of genetic research and counselling in Belgium. Both on the level of science policy and public health, it has been argued, the Belgian—and later Flemish—government invested considerably in the life sciences and opted for a decentralised model, in which subsidised centres were integrated within the country's academic hospitals. New types of multidisciplinary consultations exemplified this ongoing process of integration. The embeddedness of genetic counselling in academic hospitals furthermore made that clinical service and scientific research were easily combined, a feature which has been regarded a major strength of this model. At the same time, this brief overview has made clear that the centre's growing public role—a function insisted upon by the Ministry of Public Health—paralleled ethical debates about medical technologies in a secularising society, of which genetic diagnoses were certainly part. While these debates were conducted nationwide, they were particularly present at the University of Leuven, as the institution struggled to reconcile its Catholic heritage with its modern research ambitions.

[29] Maertens 1988.

[30] 'Goochelen met chromosomen', 1993.

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Genetic Counselling for Mediterranean Anaemia in Post-war Greece

Alexandra Barmpouti

Abstract During the twentieth century, haemoglobinopathies and Down syndrome were the most frequent hereditary diseases in Greece. Until the 1950s, medical knowledge concerning the mechanism of hereditary transmission was inadequate, thus making the work of physicians very difficult. It was only in the 1960s when the improvement of medical technology and genetics provided physicians with accurate diagnosis of the most widespread anaemia in Greece, Mediterranean Anaemia or beta-Thalassaemia. In Greece, as in the majority of Mediterranean countries, there was a growing concern for this particular disease, because of the high percentage of carriers in the region. As was often expressed at that period of time (1950–1980), Mediterranean anaemia was the prime social and medical problem.

A carrier of Mediterranean anaemia does not have any apparent symptoms, but the defective gene can be easily diagnosed with a simple blood test. Moreover, a defective gene is expressed only when the person inherits it from both parents. Due to the simplicity of the procedure and the safety of the result, many physicians found themselves obliged to recommend preventive measures, such as a simple blood test.

While some considered the counsellor's involvement in the decision-making of the parents paternalistic, others thought it imperative. Discussion about genetic counselling prevailed during the period under examination and doctors' opinions varied. Value-neutral information about the risk of the disease and a non-directive approach were most of the time impossible. However, the dominant view among the doctors was that each prospective parent should take their own responsibility towards this problem.

Since the 1960s, there have been numerous epidemiological studies and abundant statistical data dealing with the incidence of the disease. The first Centre for the

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prevention of Mediterranean Anaemia was established in Athens in 1975. The blood examination which revealed if someone was a carrier was free of charge. The Greek state funded this centre in Athens and smaller units, incorporated in big hospitals, in the rest of the country.

Although some Mediterranean anaemia experts participated in the meetings of the Hellenic Eugenics Society, the prevention of thalassaemia in Greece was not associated with eugenics. Instead, it became part of the preventive policies of the Greek national healthcare system.

Keywords Mediterranean anaemia • Thalassaemia • Haemoglobinopathies • Preventive medicine • Eugenics • Greece

1 Introduction

Public awareness for the prevention of Mediterranean anaemia in Greece was accomplished by a twofold effort of the Greek physicians with the support of the Greek Ministry of Health; on the one hand, they approached the general public by disseminating information about the disease, its symptoms and methods of transmission; and on the other hand, they provided the carriers with the service of genetic counselling. A reconstruction of the preventive programme in Greece will be attempted by the presentation of the clinical picture and methods of treatment of the disease, the contribution of the Greek physicians in the charting of the incidence of the disease throughout the country and their efforts to inform the public about it.

The diagnosis and treatment of the disease improved along with the advance in medical genetics and epidemiology; prenatal diagnosis took place earlier in gestation, and the treatment became more efficient. The Greek field studies were carried out in many parts of the country because of the variety of incidence rates in lowlands, islands or mountainous areas. As the studies continued to verify the alarming state of the incidence rate of Mediterranean anaemia in Greece, the involved physicians requested the aid of the Greek Ministry of Health to control it. Recognising the severity of the situation, the Greek state funded the initiative to implement a prevention programme and established the Centre for the Prevention of Mediterranean Anaemia in Athens. The treatment of thalassemic patients was free of charge at a public hospital, as well as prenatal diagnosis and carrier screening accompanied by genetic counselling.

The genetic counsellor was either a physician or a social worker. Although there were no official guidelines to follow, it was argued that the general rule was that counsellors should discuss with the patients about their health condition without trying to influence their decision-making. The possibility of two carriers to give birth to a child with Mediterranean anaemia was one in four, but the risk to reproduce was completely theirs; the counsellor should not intervene. However, in the absence of a controlling body to monitor each counsellor's behaviour, violations of this unwritten rule cannot be excluded.

The Greek prevention programme did not include a nationwide carrier screening or a legal constraint from marriage and reproduction of the carriers. It was a public health campaign that could not be identified or associated with eugenics. The results of the programme were successful, and a sharp decrease in the number of patients with Mediterranean anaemia was accomplished by the 1980s.

2 Clinical Features

Mediterranean anaemia is a hereditary blood disease which provokes haemoglobin deficiency. Haemoglobin includes four types of chains: a, b, c and d. Haemoglobinopathies are a group of hereditary disorders regarding haemoglobin's synthesis by these chains. They are categorised to quantitative, which correspond to the reduced production of one or more globin chains, and qualitative, which correspond to the production of pathological globin chains. Mediterranean anaemia or homozygous b-thalassaemia is a severe chronic haemolytic anaemia due to an inherited defect of the b-chain synthesis, and it belongs to the first category.[1] Thalassaemia is a single-gene disorder, inherited in an autosomal recessive character. Consequently, if one parent is a carrier and the other healthy, their child may be a heterozygous, asymptomatic, healthy individual. If both parents are carriers, they have 25% possibility to give birth to a homozygous child with Mediterranean anaemia, 25% possibility to give birth to a totally healthy child and 50% to give birth to a carrier.[2] This means that only the child who inherited the trait from both parents-carriers will develop the disease. Following the progress of biotechnology, during the post-war period, a carrier could be accurately identified by a simple blood test.

The first symptoms of Mediterranean anaemia are apparent as early as after the first 6 months of life. Without treatment, the spleen, liver and heart become enlarged. The child is usually sluggish, pale, has poor appetite, cannot play or move for long time. Additionally, the child's growth is slow, and often there is abdominal swelling due to splenomegaly or hepatomegaly that is associated with the disease. Progressively, there are bone deformities and hormonal disorders affecting the overall development of the child.[3] Furthermore, thalassemia's treatment includes frequent blood transfusions from an early age, which unavoidably influence heart function due to iron overload. Frequent blood transfusions also cause cirrhosis of the liver; endocrine complications and diabetes.[4] The treatment often includes a splenectomy and the use of an iron-chelating agent to control the iron overload.[5]

[1] Kattamis et al. 1970, 502–505.

[2] See Weatherall 2010.

[3] Cao 2010, 62.

[4] See Farmaki and Galanello 2011, 272–280.

[5] WHO Working Group 1982, 655.

Nowadays, however, timely treatment in combination with blood transfusions could provide a decent survival and lifestyle of the patients. Of course, they are obliged to have blood transfusions once a month, to take medication on a daily basis and be absent from school and work. However, they could have a social life, study, work and in some cases children as well. Until the present day, unfortunately there is not an effective treatment to eliminate the disease. As a result, it remains a chronic illness which provokes misery and anxiety.[6]

3 Mediterranean Anaemia's Incidence in Greece

In Greece, haemoglobinopathies' incidence was very high during the twentieth century. Particularly Mediterranean and sickle-cell anaemia were important medical problems in Greece, justifying the special attention that was given to these diseases. Extensive epidemiological studies were carried out by Greek researchers throughout the country and confirmed that Mediterranean anaemia occurred more often in lowlands, such as Karditsa in Central Greece, Arta in Western Greece and in some big islands, such as Corfu and Lesvos. There the incidence rate reached 20%, whereas in regions, such as Macedonia and Thrace, in the northern part of the country, the incidence rate was 2–3%.[7]

In 1936 and 1938, the physician and Director of the Medical Laboratory at the Hellenic Pasteur Institute, Ioannis Caminopetros (1898–1963), firstly observed and published in two papers[8] that Mediterranean anaemia was inherited in an autosomal recessive character by healthy carriers and suggested medical counselling to parents who had already gave birth to a child with anaemia.[9] Caminopetros' publications were popularised much later though, because they coincided with the outbreak of the Second World War. However, he is an internationally acclaimed researcher on both sickle-cell and Mediterranean anaemia.[10]

Although thalassaemia was endemic in Mediterranean countries, it was hardly traceable before or during the wars, because there were numerous contagious diseases, malnutrition and harsh living conditions which impeded the detection and prevention of the disease. Moreover, children with Mediterranean anaemia died very young due to the aforementioned conditions and lack of treatment.[11] However, after the Second World War, when the living conditions improved, the contagious diseases eliminated and medical genetics advanced, physicians became interested in the study of Mediterranean and sickle-cell anaemia.

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[6]Ibid. and Koutelekos and Haliasos 2013, 101–112.

[7]See Fessas and Stamaloyannopoulos 1964a; Fessas and Loukopoulos 1964b; Loukopoulos 1965.

[8]Caminopetros 1938a, 27–43; Caminopetros 1938b, 104–125.

[9]Malamos 1962, 5–13; Cowan 2008, 192.

[10]Caminopetros 1952, 687–693.

[11]Loukopoulos 2011, 572.

Italians and Greeks instigated the study of Mediterranean anaemia almost simultaneously, yet independently. Italian research studies on Mediterranean anaemia were begun by Ezio Silvestroni (1905–1990), Ida Bianco (b. 1917) and later Giuseppe Montalenti (1904–1990). Similarly to Greece, some regions of Italy had a high incidence of the disease, while others had a low one. In Italy it was also observed a high incidence in lowlands and in the southern part of the country.[12] Interestingly enough, Italian researchers embraced the theory of the 'J. B. S. Haldane hypothesis' suggesting that heterozygous carriers of Mediterranean anaemia were resistant to malaria and demonstrated a link between the two diseases.[13] According to Stefano Canali (b. 1963) and Gilberto Corbellini (b. 1958), there were studies on this association in all countries where malaria was endemic, including Greece. In Greece, it was widely supported the opposite suggestion that the areas of Macedonia and Thrace in Northern Greece had a low rate of Mediterranean and sickle-cell anaemia patients due to the high rate of malaria.[14] However, the high incidence of malaria, which persisted for many years, does not seem to be the reason why researchers became interested in Mediterranean anaemia.

Most probably the growing interest in Mediterranean anaemia occurred due to the numerous admissions of children with anaemia in Greek hospitals. In particular, paediatricians were more concerned with the high incidence of the disease because they encountered cases with children who died very young due to the lack of diagnosis and treatment. The increasing number of thalassaemic children admitted to hospitals was the motivation to study Mediterranean anaemia.[15]

The historical course of scientific research on Mediterranean anaemia can be approximately divided in three periods. During the period from 1925 to 1950, there were the first clinical observations of Mediterranean anaemia resulting in the confirmation of its clinical picture. The beginning of this period is marked by Cooley's research on the disease. Thomas Cooley (1871–1945) was the first to provide scientific evidence for the incidence of this type of anaemia, beta-thalassaemia, based on studies on children of Greek and Italian origin. This is also the reason for referring to this disease as 'Cooley's anaemia'.[16] After Cooley's observations in 1925, the Greek scientists were more concerned about defining the clinical and haematological heterogeneity of the disease. As far as the name of the disease was concerned, in Greece, the term 'Mediterranean anaemia' prevailed, while the term 'thalassaemia' was rarely used. Caminopetros' study in the 1930s confirmed that the disease was hereditary in an autosomal recessive character[17] and until the 1950s the phenotypic characteristics were also identified.[18]

[12]Canali and Corbellini 2006.

[13]Dronamraju 2006, 2–4.

[14]Canali and Corbellini 2006, 58.

[15]Angastiniotis and Eleftheriou 2011, 314.

[16]Cooley and Lee 1925, 29.

[17]Caminopetros 1938a, b.

[18]Kattamis et al. 1973; Kattamis 2011b, 330.

The second period, from 1950 to 1975, was characterised by important scientific advances, particularly in biochemistry and studies in protein structure, which permitted the use of more accurate biochemical and blood examination methods of diagnosis. Furthermore, during the second period, many demographical studies took place in the broader region of the Mediterranean Sea revealing the high incidence of the disease. Moreover, the definition of the normal structure of haemoglobin triggered the study for the pathophysiology of Mediterranean anaemia.[19]

The third period, from 1975 until the present day, is characterised by the rapid progress of genetics and molecular biology which resulted in the application of innovative molecular diagnostic methods and treatment.[20] The pivotal point was the introduction of marrow transplantation in the treatment of Mediterranean anaemia in 1982.[21] This became the most effective treatment for patients who could find a compatible donor. Furthermore, the establishment of state-funded prevention programmes in the Mediterranean area, including Greece,[22] Italy[23] and Cyprus,[24] was essential to the decrease of the incidence of haemoglobinopathies. The most important coordinated effort to control haemoglobinopathies on a global level was a meeting of the World Health Organization's working group in November 1981, which comprised of well-known scientists representing their countries, such as Bernadette Modell (b. 1935) and David Weatherall (b. 1933) from Great Britain, Dimitrios Loukopoulos (b. 1935) from Greece, Antonio Cao (1929–2012) from Sardinia and many more. Their report included the most recent statistical data, diagnostic criteria and methods of treatment for hereditary anaemias.[25]

Caminopetros' first scientific research on haemoglobinopathies set the basis for the next generation of researchers, such as Vassilios Malamos (1909–1973), Phaedon Fessas (1922–2015), George Stamatoyannopoulos (b. 1934), Christos Kattamis (1933–1998), Dimitrios Loukopoulos and others, who became the protagonists in Mediterranean anaemia's research, treatment and prevention. The majority of the researchers belonged to the University of Athens' medical clinics, which were the source of funding for the epidemiological surveys. As mentioned before, the research studies were carried out in different regions of Greece due to the differentiation of incidence rates of the disease in the country.[26]

The first results from epidemiological surveys in Greece were published in the British Journal of Haematology in 1962 by the physicians Vassilios Malamos,

[19]Kattamis et al. 1979; Kattamis et al. 1982.

[20]Kattamis 2011a.

[21]Thomas et al. 1982.

[22]Loukopoulos 2011.

[23]Silvestroni and Bianco 1975; Silvestroni and Bianco 1983.

[24]Ashiotis et al. 1973; Angastiniotis et al. 1986.

[25]WHO Working Group 1982.

[26]Choremis et al. 1963; Fraser et al. 1964; Barnicot et al. 1963; Barnicot et al. 1965.

Phaedon Fessas and George Stamatoyannopoulos.[27] They collected data regarding the haematological and biochemical abnormalities of the carriers in order to gather information about the frequency of the trait. They conducted research in 1600 young males serving in the Greek Air Force, who came from different regions of Greece. One hundred and nineteen of the examined individuals were carriers of the trait, which means an incidence of 7.44%. Although this was not a population-scale study, this sample represented individuals from different parts of Greece. The research indicated a high rate in some regions of the country, such as the region of Epirus in mainland Western Greece and the Ionian islands. According to the researchers: 'the high incidence of 7.44% in an unselected group indicates not only the wide distribution of the abnormal genes, but also that in certain restricted areas the percentage of trait carriers may be extremely high'.[28]

During the 1970s, the number of severe cases of Mediterranean anaemia admitted to the Greek hospitals increased a fact which alarmed physicians about its frequency. In 1974, Kattamis conducted a research in the First Paediatric Clinic of the University of Athens regarding the number of children suffering from congenital diseases who were hospitalised, the days of hospitalisation and the number of the beds that they used. The results showed that 2071 out of 9664 children with congenital diseases, which correspond to 21.4%, suffered from Mediterranean anaemia. The percentage was extremely high and showed the gravity of the problem. The second most frequent disease was sickle-cell anaemia with 138 children (1.3%) and the third was cystic fibrosis with 20 children (0.2%). Kattamis was convinced that the medical advances could be better appreciated with the cooperation of other sciences and the sympathy of the entire population in order to prevent the incidence of the disease.[29]

Furthermore, statistical data revealed that the life expectancy for these patients was very short in the 1960s; without blood transfusions, patients would survive only the first decade of their lives.[30] It is remarkable that until the end of the 1970s, teen patients were considered as 'patients at high risk of death'.[31] Added to this, the need for blood supplies was growing, because frequent blood transfusions were an indispensable part of the treatment. In parallel with the campaign for the prevention of Mediterranean anaemia, a campaign for blood donation was prominent. Based on a number of scientific research studies, the control and prevention of the disease became a matter of emergency. Therefore, a public health policy to effectively tackle the disease was deemed necessary. The fact that Mediterranean anaemia could be safely diagnosed by only a blood test was the motive to expand the research and promote preventive medical measures and genetic counselling.[32]

[27]Malamos et al. 1962.

[28]Ibid. 11.

[29]Hellenic Eugenics Society 1976, 136.

[30]Angastiniotis and Eleftherieu 2011, 313.

[31]Modell 1977, 495.

[32]Hellenic Eugenics Society 1976.

To this end, certain preconditions before the implementation of a health policy were required. On the scientific level, the involved physicians had to gather the demographical surveys and field researches to accurately determine the incidence rate of the disease and acquire valid statistical data to work upon. In addition, they had to calculate the approximate cost of the laboratory examination, the haematological analysis, because a high cost would have been a serious obstacle to their initiative.

After validating the data and evaluating the cost, the most important step was to persuade the state to fund their activities and the authorities to implement a national preventive programme. The Greek health system was completely disorganised and poor in the beginning of the twentieth century, and the Greeks relied on philanthropy and private health institutions. During the interwar period, including the influx of 1.5 million refugees from Asia Minor (1922), the health system was inextricably linked to external help, mostly coming from international health institutions, such as the Rockefeller Foundation and the League of Nations Health Organisation which undertook the health and hygiene protection of the Greek people.[33] After the Second World War (1946) and the Civil War (1946–1949) that followed, the Ministry of Health was reorganised, and a national health system was established, which continuously improved.[34] Therefore, the initiative to implement a campaign on the prevention of Mediterranean anaemia targeted exclusively at state financial support.

Moreover, it was crucial to obtain the agreement of the Greek Orthodox Church, because it was the leading religious body in Greece and its approval would benefit the popularisation of a preventive health policy. Last but not least, they had to find ways to reach the wider public and convince the Greeks to take the blood test.

The first attempts were begun by Phaedon Fessas, George Stamatoyannopoulos, Christos Kattamis and Dimitris Loukopoulos in the 1970s, who were the founders of the Centre for the Prevention of Mediterranean Anaemia. It was established in Athens in 1975, and it was due to the function of this Centre—and the smaller units which were incorporated in large hospitals of the country—that Mediterranean Anaemia's percentage was reduced. The ultimate goal was not only to improve the treatment of the disease but also to popularise its prevention. It was crucial to inform the Greek adults about the possibility to give birth to a child with Mediterranean anaemia before reproduction. The actual success would be to eliminate the disease before conception, namely, to identify the carriers and advise them about their condition by genetic counselling.[35]

[33]Weindling 1997.

[34]Dardavesis 2008.

[35]Loukopoulos 2011, 573.

4 Dissemination of Information

Aiming at public awareness, Greek physicians who were preoccupied with Mediterranean anaemia instigated public information about it in many ways. One of them was the circulation of posters and leaflets. The Greek Ministry of Health undertook the cost of publications and supported the members of the Centre for the Prevention of Mediterranean Anaemia in Athens to reach the public.[36] Thus, the involved physicians wrote short leaflets including information about the prevention of Mediterranean anaemia labelled with the question: 'Now is the time to have a child! Did you take a blood test for Mediterranean Anaemia?' (Fig. 1). Distributing information about the disease in simple language and in a large group of people, aiming at young adults and newly married couples, was one of the most effective methods to implement preventive medicine.[37] People, who read these leaflets acquired an overall image of the method of transmission and symptoms in order to be convinced to take the blood test and find out whether they were carriers or not. The simplicity of the procedure was the most persuasive factor in controlling the disease before marriage and conception.

Moreover, the members of the Centre for the Prevention of Mediterranean Anaemia disseminated an informative leaflet of 24 pages to be distributed to family doctors (Fig. 2). The fact that all physicians should have been aware of such an important medical problem was not overlooked by the initiative to eradicate Mediterranean anaemia. Although physicians, such as haematologists, microbiologists, gynaecologists and paediatricians, were directly involved with the manifestation of the disease, other specialisations, such as general practitioners and pathologists, were not always informed. At the same time though, physicians having these specialisations played a critical role in reaching the public; thus, it was essential to study to help the prevention of the disease. In fact, general practitioners had the opportunity to reach more single adults who formed the primary target group of the campaign. On the contrary, gynaecologists and paediatricians, for instance, got in touch with specific groups of people, mostly married couples and pregnant women. Therefore, physicians of all medical specialisations had to be informed and educated about Mediterranean anaemia, in order to become a part of an effective and fruitful campaign.

In addition, information for the disease was disseminated in high schools by lessons on haemoglobin disorders and posters and special events in order to introduce the disease to the pupils. This was also a method of educating teachers and parents. Associations of parents having a child with Mediterranean anaemia and associations of patients were also informed about potential methods of treatment.[38] The first official patients' advocacy group was established in 1980 in Athens under the name 'Pan-Hellenic Association of Patients with Mediterranean

[36] Ibid. 575.
[37] Ibid.
[38] Ibid.

Fig. 1 Informative leaflet for parents by *Centre for the Prevention of Mediterranean Anaemia, Greek Ministry of Health* (Photo: D. Loukopoulos)

Anaemia' (Πανελλήνιος Σύλλογος Πασχόντων από μεσογειακή Αναιμία) which was exclusively led by the patients themselves and their families.[39]

Furthermore, an important factor which aided the spread of knowledge for the disease was that people suffering from it looked very sick. Before the period when the treatment of the Mediterranean anaemia became effective, it was obvious when someone is suffering from it, making him a living example of the manifestation of the disease and thus sensitising the public. The patients' appearance was unusual to the public, because they had very pale skin, severe bone deformities and abdominal swelling. The image of a sick person with Mediterranean anaemia was used by physicians and genetic counsellors to provide the counselees with a visual

[39] www.paspama.gr.

Fig. 2 Informative leaflets for physicians by *Centre for the Prevention of Mediterranean Anaemia, Greek Ministry of Health* (Photo: D. Loukopoulos)

manifestation of the disease. Gradually, in parallel and as a result of the progress in medical genetics and the improvement of treatments, such as the use of iron-chelating agents which controlled iron overload, the appearance of the sufferers from Mediterranean anaemia improved as well.[40]

After a cost-benefit analysis,[41] comparing the cost of preventive measures with the cost of hospitalisation and treatment of patients, the state financially supported prevention by information and examination and included the treatment of Mediterranean anaemia in the public healthcare system. Thus, the Greek state implemented a national programme for the prevention of Mediterranean anaemia by safe and free tests for identification of carriers, prenatal tests and genetic counselling for couples at risk, individuals and newly married couples. Furthermore, the state financed the publication of propaganda materials, mostly leaflets. Without the Greek state's aid, the success of the preventive health policies in tackling the disease would have never occurred. Greece, Cyprus[42] and Sardinia[43] were some of the regions in the

[40]Ibid.

[41]Ibid. 573.

[42]Angastiniotis et al. 1986; Ashiotis et al. 1973.

[43]Cao et al. 1981.

Mediterranean Sea, where a systematic preventive programme on a national basis was instituted.[44]

Equally important was the fact that the Greek Orthodox Church agreed with the initiative for the prevention of the disease. Orthodox Christianity, the dominant religion in Greece, favoured activities aiming at the alleviation from pain and health restoration. In every Church, there were informative leaflets for the prevention of Mediterranean anaemia. However, there was the thorny issue of abortion which was regarded as the ultimate sin, because it was equated to murder. A prenatal test which would result in induced abortion was not ethically acceptable to Orthodox Ethics. Although the Church was absolutely against abortion, medical information and examination to avoid a disease was desirable.[45] Given that during the service of marriage a Greek Orthodox priest prays for 'the fruit of the womb' and the 'beholding of sons and daughters', it is unlikely to suggest the option of not having children.[46] As a result, the Church finally decided to stay uninvolved with the campaign and let physicians to deal with it.[47]

5 Methods of Genetic Counselling

All these activities aiming at the wide distribution of information for the prevention from the disease transformed diagnosis and counselling from retrospective to prospective. The retrospective method of counselling followed prenatal diagnosis during pregnancy, whereas the prospective method followed carrier identification. Before establishing a coordinated action to inform the public for ways of prevention from the disease, prospective parents were not aware of the potential risk of giving birth to a child with Mediterranean anaemia. Given that a carrier did not manifest the disease, prospective parents were ignorant of the risk.[48] Added to this, they did not know that if both were carriers, they could inherit a severe disease to their child. Therefore, there was not an obvious reason for taking a medical test before marriage. In fact, as was already mentioned, the ultimate goal of a preventive health policy was to alert the carriers to take the blood test before the conception of a child with the disease. On the one hand, the carriers would be aware of their condition and evaluate the risk of a pregnancy, and on the other hand, the experience of an induced abortion could be avoided.[49]

Progressively, there was a shift in genetic counselling for Mediterranean anaemia from retrospective to prospective, because the diagnosis of adult carriers prior

[44]Maniatis 2008.

[45]Chatzinikolaou 2002; Mantzarides 2009; Vantsos 2009.

[46]Service of Crowning or Marriage. The Great Euchologion of the Orthodox Church 2014, 199.

[47]Loukopoulos 2011, 575.

[48]Angastiniotis et al. 2011, 314.

[49]Modell 1980.

to their decision to procreate replaced late diagnosis of embryos and newborns. The gain of this shift was immense and practically led to the sharp decrease of the incidence of Mediterranean anaemia in Greece.[50]

During the 1970s, the first Prenatal Diagnosis Units appeared in Greece. In particular, the first prenatal diagnosis was performed in 1975. Until 1977 prenatal genetic diagnosis became a common practice.[51] The first laboratory for the detection and genetic counselling for Mediterranean anaemia was established in 'Alexandra' Maternity Hospital in Athens, led by Dimitrios Loukopoulos, haematologist, trained in the USA with Blanche Alter in Professor's David Nathan Department in Boston, and Aristides Antsaklis (b. 1946), gynaecologist, who was trained in the techniques of foetoscopy with Professor Fairweather in London.[52] According to Loukopoulos,[53] Phaedon Fessas, who was an expert in the study of Mediterranean anaemia and director of the Centre of Haematology at the same hospital, and Spyros Doxiadis (1917–1991), Professor of Paediatrics at the University of Athens and Minister of Health and Social Affairs during this period, were instrumental to the functioning of this laboratory and the following prevention programme for Mediterranean anaemia.

According to Kattamis' viewpoint, there was a distinction between premarital advice and genetic counselling saying that the latter follows laboratory results, while the former is only theoretical and could include advice about fertility, sterility or family planning.[54] Setting this distinction, genetic counselling was primarily performed by physicians because they could more accurately interpret the results of a laboratory examination. However, the result of a blood examination for Mediterranean anaemia did not leave any uncertainties or room for false interpretation; namely, it was a yes or no answer. Therefore, social workers were also counsellors.

The most important aspects of genetic counselling for Mediterranean anaemia were the simplicity of the procedure, just a haematological examination, and the safety of the result. Having a positive or negative result from the test was not a complicating procedure, but the identification of a carrier or the result of the prenatal examination was immediate. First of all, diagnosis was accurate and simple; a simple blood test could detect the carrier. Secondly, the method of transmission was known and verified. Only if both parents were carriers, they faced a risk of 25% to give birth to a child with the disease. Thirdly, in accordance with Mendelian genetics, the possibilities of transmitting the disease could be mathematically calculated. The genetic advisor was obliged to inform the carriers about their possibilities, one in four, to give birth to a child-patient and their options. However, the prospective parents were the only responsible to assess this risk. Based on these concrete facts, the counsellor could be confident enough to

[50]See Loukopoulos et al. 1983; Loukopoulos et al. 1988.

[51]Loukopoulos et al. 1982.

[52]Loukopoulos 2011, 575.

[53]Loukopoulos 2016: Interview with the author. Athens, Greece.

[54]Hellenic Eugenics Society 1978b, 307.

provide precise information to the carriers or prospective parents. Unlike other multifactorial genetic diseases, Mediterranean anaemia was easily detected, and its symptoms could be predicted with certainty.

The need for genetic counselling was deemed necessary in some specific occasions, firstly, in the case when one of the prospective parents was a carrier, because he/she should verify that his/her partner was not also a carrier. The couple should be aware of their potentialities regarding the health of their prospective child. Secondly, if a child with an abnormal gene was born in a family, while both parents seemed healthy, this was certainly an alarming fact, because it automatically meant that both parents were carriers. In cases when, according to medical indication, both parents have increased possibilities to give birth to a child with a hereditary disease, the counsellor should inform them about their options. These often included permanent use of contraceptive techniques for not having children, in vitro fertilisation and preimplantation diagnosis, in vitro fertilisation with a donor or prenatal diagnosis.[55]

Fessas argued that counselling should be realised in four steps, by objective analysis of the details of the medical condition, adequate information about its symptoms and treatment, explanation of the medical condition in plain words in order to be clearly understood and discussion. The counsellor should be precise, objective, educated, comprehensible and tolerant. It is critical to communicate the information to the counselee according to the counselee's social status and education. The counsellor should be flexible and adjust his language of communication to the counselee's perception in order to be understandable. Moreover, he should keep a neutral position without hiding or emphasising any aspect of the disease.[56]

Although the counsellor, who was almost always a physician, should be absolutely neutral and let the couple decide, it was very difficult to maintain a neutral position, because patients trusted physicians and often asked for their opinion to decide and because, as Fessas argued, the medical profession was, by nature, invasive.[57] Added to this, there was no method of controlling a counsellor's work. There was not a way to prove that he undoubtedly kept an impartial position. Experts in Mediterranean anaemia counselling admitted that a paternalistic approach was improper, but, in fact, there were no officially published guidelines for genetic counselling for Mediterranean anaemia. Each physician dealt with each couple or individual according to his own experience, knowledge and morality. Many times he asked assistance from a social worker or a nurse.[58] However, the general rule was non-directiveness and neutrality. The role of the counsellor was to provide ample and detailed information about the disease and let the couple or the individual trait carrier decide and be responsible of their own choice. The

[55]Loukopoulos 2011, 574.

[56]Hellenic Eugenics Society 1978b, 303.

[57]Hellenic Eugenics Society 1978b, 308.

[58]Loukopoulos 2011, 574.

counsellor should avoid taking a decision on behalf of others or manipulate them towards the decision he thinks of as correct.[59]

If the diagnosis took place before marriage or pregnancy, the genetic advisor—based on the safety of the result—was obliged to inform the trait carrier(s) about the disease, its symptoms and the method of transmission. As in other cases, couples diagnosed with the trait either decided to marry but not to procreate, to marry another person, to adopt a child, to use in vitro fertilisation with preimplantation diagnosis or with a healthy donor or to take the risk to procreate, irrespective of the diagnosis, because still they had 75% to give birth to a healthy child/carrier.

If the diagnosis took place during pregnancy, then the approach was practical, because the diagnosis from the prenatal test could lead to the decision of an induced abortion. An important detail was that in the 1970s, prenatal genetic examination took place up to the 18–20th week of gestation, whereas later in the 1980s, this could be made during the 10–12th week, using chorion villus sampling.[60] The period of gestation was often an important factor in the decision-making of the parents regarding abortion. It was more difficult for a pregnant woman of 20 weeks to decide to have an abortion than for one of 10–12 weeks.

Experience from the Centre for the Prevention of Mediterranean Anaemia showed that the majority of couples, whose embryo was diagnosed with the disease, chose abortion to secure that they will not give birth to an unhealthy child.[61] Furthermore, legislation protected the decision of the couple, because abortion due to foetal genetic abnormality was legally accepted and free of charge at a public hospital. It seems that the biomedical progress influenced the legislation regarding abortion. During the late 1970s, there was an obvious ongoing process of liberalising abortion laws across Europe. By the end of the decade, the majority of European countries had already abolished their strict laws and passed more liberal laws on abortion.[62] In Greece, in 1978 there was a significant change in the existing law which permitted abortion due to foetal abnormality. A few years later, in 1986, a new, more liberal law replaced the previous one, which on the one hand permitted induced abortion for reasons of foetal impairment up to the 24th week of gestation and on the other hand dictated that the social security fund was obliged to cover the expenses of the woman's operation and hospitalisation in a public clinic, thus safeguarding the safety of the procedure and mother's health.[63] Therefore, the legal context for abortion not only facilitated the decision of the couple but also made easier for genetic counsellors to suggest such an option.

The essential success of the Mediterranean anaemia prevention programmes was the fact that more and more people undertook the blood test before marriage and conception.[64] During the first year of function of the Centre for the Prevention of

[59]Hellenic Eugenics Society 1978b, 308.

[60]Aleporou-Marinou et al. 1980; Loukopoulos et al. 1982; Weatherall 2010, 59.

[61]Ibid.

[62]David 1992; United Nations 2002.

[63]Barmpouti 2015, 40–41.

[64]Hellenic Eugenics Society 1978b, 306.

Mediterranean Anaemia, approximately 2000 people voluntarily visited the centre and more than 6000 the second year. In a period of 4 years, the percentage of heterozygous individuals of the Greek population decreased from 21 to 17%. Gradually, people realising the danger of the disease and the advantage of knowledge visited the centre as singles, not necessarily as couples before marriage.[65]

6 Eugenics

In the context of popularising the campaign for the prevention of Mediterranean anaemia, physicians associated with the preventive health programme participated in public discussions organised by the Hellenic Eugenics Society. Although Mediterranean anaemia specialists of the 1970s, such as Fessas and Kattamis, were not members of the Hellenic Eugenics Society, they were guest speakers in some of its conferences presenting their views on haemoglobinopathies in general and Mediterranean anaemia in particular. These conferences were open to the public and often attracted large audiences. At the same time, the scientific and academic elite of the country and influential politicians also participated or attended these conferences.[66]

The Hellenic Eugenics Society was founded much later than its European counterparts, in 1953 in Athens.[67] Although one could reasonably wonder why a eugenics society was established in a European country after the atrocities of the Third Reich, this was primarily due to its association with family planning and contraception, which were then gaining publicity.[68] 'Racial purification' and social exclusion of the 'unfit members of the society' were not among its aims. Instead, these were dissemination of information about ways to found a healthy family; prevention from contagious or hereditary diseases; and problems of demography.[69] Among its numerous conferences, there were also discussions about the social consequences of medicine and medical issues, like modern-day bioethics. Briefly, it was a think tank of eminent physicians, particularly gynaecologists, academics and professionals of the time. The Hellenic Eugenics Society itself did not adopt or promote extreme eugenic policies, such as forced sterilisation, marriage restrictions or social segregation.

Given that Mediterranean anaemia was a leading socio-medical problem, the Hellenic Eugenics Society discussed it in the context of three of its conferences:

[65] See Aleporou-Marinou et al. 1980; Loukopoulos 2011.

[66] Hellenic Eugenics Society 1965; 1977; 1978.

[67] Lane 1955, 198.

[68] Bashford 2014.

[69] Louros 1955 and 1960.

Blood and Heredity (1970),[70] Antenatal Diagnosis (1975)[71] and Premarital Medical Examination (1978).[72] There, genetic counselling and popularisation of preventive measures dominated the discussions. Presumably, Mediterranean anaemia experts chose to participate in these conferences due to their popularity. This was one more way to publicise their effort against the disease, both to their colleagues and the general public.

Regarding the sensitive issue of eugenics, as a matter of fact, there is always a thin borderline between preventive medicine and eugenics. This very fact could be used diplomatically either to support the one or the other side because there is not a universally agreed definition of what eugenics is.[73] Most of the times, eugenics is identified or associated with biopolitics, state intervention in fertility and reproduction and the implementation of coercive genetic screening policies, social stratification according to a social or legal norm, racism and population management. Eugenics' utopia also envisioned a bodily and national perfection. Not only was eugenics a method for biological enhancement but also aimed at social and national improvement.[74]

Therefore, one could claim that the effort to control the spread of Mediterranean anaemia was a form of eugenics. Given that the aim of the prevention campaigns was to eliminate the birth of children with genetic defects, some accused these efforts for controlling heredity aiming at 'race betterment'. Prevention programmes for Mediterranean anaemia, such as the one in Italy and in Cyprus, were characterised as 'latent eugenic', mainly due to the criticism for compulsory genetic screening.[75] Italy was the first among the southern European countries to implement a prevention programme for Mediterranean anaemia, as early as the 1950s. Although the programme was not implemented in the same manner in the entire country, in some regions, such as Ferrara, from 1956 until 1963, a compulsory carrier screening of the school children of the area was carried out to form a complete list of carriers. The families of the children carriers received written genetic counselling on how to avoid marriage with another carrier, thus developing a eugenic 'mind-set'.[76] According to Canali, Giovani di Guglielmo (1866–1962) suggested the sterilisation of the trait carriers, while Sergio Sergi (1878–1972), in agreement with Silvestroni and Bianco, suggested compulsory screening of the entire population of Italy, providing evidence for a eugenic thinking.[77] In Greece, however, a compulsory carrier screening was never implemented, and the premarital health certificate was compulsory only during the dictatorship (1967–

[70]Hellenic Eugenics Society 1978a, 7–28.
[71]Hellenic Eugenics Society 1976.
[72]Hellenic Eugenics Society 1978b.
[73]See Bashford and Levine 2010, 3–24.
[74]See Turda 2010.
[75]Cowan 2009.
[76]Canali and Corbellini 2003, 747.
[77]Ibid. 3.

1974), but even then it was not restricting; a negative result would not refrain the couple from marriage and reproduction.[78] Moreover, none of the involved physicians and other professionals suggested including the test for Mediterranean anaemia in the premarital certificate or forced couples to undertake the test before marriage. On the contrary, Fessas argued that marriage prohibition of carriers would be a very strict eugenic measure which would undermine their personal freedom; therefore, it should be avoided.[79]

The implementation of the preventive health policies for Mediterranean anaemia in Greece was the coordinated effort of both the medical community and the Greek Ministry of Health which could not be characterised as eugenics but as preventive medicine. Although one can trace elements of eugenic thinking, such as the mentality of selection on which is often based the decision of abortion after a negative prenatal diagnosis, this was actually what the prevention programme wanted to avoid. By putting emphasis rather on carrier identification than prenatal testing, the involved agents automatically set prevention as the priority of their initiative. The much desirable shift from retrospective to prospective genetic counselling was the ultimate success of the programme, which secured fewer births of homozygous children and fewer abortions.

The non-directive approach of counselling adopted by the majority of the physicians-counsellors was also indicative of the absence of eugenics. Physicians were not meant to impose their opinion or manipulate the counselee but to help him decide by providing him with a wealth of information. Fessas considered the role of the physician and the impact of the diagnosis on the patient equally important. He argued that scientific advances influenced the function of society. People should be aware of the new technologies in medicine along with their use. Fessas also claimed that people should not be tempted to alter their genetic inheritance for eugenic reasons and that scientists ought to allow biological variety in society.[80]

As far as state intervention was concerned, public information and the examination were free and voluntary. It was not a coercive, population-based eugenics screening. The goal of the campaign was to inform, not to impose, a certain practice by legal or other means. Carrier identification was made on a voluntary basis, and the patient's privacy of his medical condition was protected by the law. Thus, the physician who performed the test or the counsellor did not have the right to reveal the name and condition of the patient under any circumstances. Therefore, a forced elimination of the 'unfit' was not the case in Greece. Furthermore, the agents of the Mediterranean anaemia prevention campaign emphasised the difference between the preventive character that a blood examination entailed and the constant control of reproduction of a nation by the state, which would form a eugenics policy. The Greek state never passed a law prohibiting people from marriage and reproduction for genetic or other reason.

[78]Greece. Law 300/1968.

[79]Hellenic Eugenics Society 1978b, 302.

[80]Hellenic Eugenics Society 1976, 124.

Mediterranean anaemia experts' goal was to prevent the proliferation of the disease by identifying the carriers and inform them about the method of transmission and symptoms of the disease. Certainly, it is unrealistic to claim that every gynaecologist or other genetic advisor neutrally advised the pregnant woman about her diagnosed embryo and did not insist to undergo an induced abortion. However, the predominant viewpoint was non-directiveness and neutrality. The decrease of the incidence rate of the disease was to be realised through information and counselling before marriage and reproduction.

7 Present Day

Public awareness and genetic counselling for Mediterranean anaemia during the post-war period in Greece was very fruitful, resulting in the decrease of the incidence of the disease in a rather short time period. On the contrary, nowadays, there is no sensitisation towards the disease for a variety of reasons.

For example, a person with Mediterranean anaemia no longer looks like a sick person at all, and the society does not have an image of the manifestation of the disease. Furthermore, in the absence of a coordinated campaign for the prevention of the disease, the dissemination of information rests to the hands of gynaecologists who might recommend a blood test for Mediterranean anaemia.

Another reason is the deficiency in monitoring the diagnosis, because many times the examiners do not know the final decision of the examined individual after diagnosis and counselling.

Last but not least, Mediterranean anaemia was not entirely eliminated due to the difficulty to reach and educate minorities who live in the country, such as the Roma people and Albanians. Increasing immigration from Albania, where the incidence rate of Mediterranean anaemia is around 15%, who mostly prefer to marry among their kin, was a fact which increased the incidence in Greece. Inadequate or non-existent information for preventive measures is the primary cause that these communities continue to give birth to children with Mediterranean anaemia. The most efficient solution would be the dissemination of information by educated social workers of the same origin and language.[81]

These are some of the reasons why the disease is not entirely eradicated which simultaneously confirm the need to reintroduce a campaign for the prevention of the Mediterranean anaemia in Greece.

[81] Loukopoulos 2011, 574.

8 Conclusions

To conclude, Mediterranean anaemia was an acknowledged threat of the health of Greek people during the mid-twentieth century. Statistical data revealed a high incidence both of carriers and patients admitted to the hospitals; thus the control of the disease was urgent.

The initiative towards the elimination of the disease began in the 1960s. The campaign for the prevention of the disease reached its peak in the 1970s when the Greek state financially supported the physicians who organised it. During the same period, the Centre for the prevention of Mediterranean anaemia was established in Athens.

Genetic counselling prior to pregnancy was the ultimate target of medical professionals engaged with the eradication of the disease. In other cases where the couple was not aware of having the abnormal gene and faced a pregnancy, prenatal examination could accurately verify the disorder. In this case, it was the genetic counsellor's duty to inform them about the disease and their options. The final decision, however, remained at the couple's disposal, because the general rule was neutrality and non-directiveness.

An obligatory carrier screening was never implemented in Greece. Instead, citizens were informed for the possibility to take a blood examination free of charge in order to detect or exclude a negative genetic disposal. As a result, the campaign to inform the Greek society was a well-organised preventive health policy, funded by the state and having a successful outcome in reducing the incidence of Mediterranean anaemia in the country. Since the first studies on Mediterranean anaemia, its treatment has been improved on many levels, but mostly because of the possibility of marrow transplantation. Hopefully, a successful gene therapy will be soon available for all patients.

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Karyotyping and the Emergence of Genetic Counselling in Mexico in the 1960s

Ana Barahona

Abstract In the aftermath of World War II (WWII), there was growing interest for international peace that gave rise to international cooperation programmes and organizations that produced important changes in the international political landscape. It was in those years, when global trends in human genetics were reshaping the field of biomedicine and when growing international interest in understanding the effects of radiation on human beings led to the formation of institutions and a proliferation of multi-centred clinical trials and inter-laboratory studies. In Mexico, the first studies on chromosomes were performed by Mexican paediatrician-turned-geneticist Salvador Armendares and his colleagues at the Mexican Institute of Social Security (IMSS) in the 1960s. Their work was based on the study on congenital malformations performed by the WHO that Mexico had participated in which was carried out by Alan C. Stevenson, one of the earliest medical geneticists in the UK. Armendares spent 2 years at the British Medical Research Council in Oxford in 1964 and 1965 under Stevenson's supervision. Upon Armendares' return from England in 1966, the first Unit for Research in Human Genetics was created at the IMSS. The Unit was created with the main objective of providing medical genetic services in a clinical setting. Armendares and the colleagues who soon joined the Unit were aware of the growing importance of chromosome studies in clinical practice, particularly concerning genetic counselling for certain diseases. Human geneticists at the Unit developed precise diagnostic protocols to provide accurate genetic information to the patients for the development of future treatments and prophylaxis (preventive medicine). In his 1968 book, *Citogenética Humana* (*Human Cytogenetics*), Armendares included a chapter on genetic counselling as being the most important practical application of human genetics knowledge. Armendares was the first to relate

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karyotyping with genetic counselling, translating test results and technical language for the patients or their parents at the hospital. He played a key role in educating physicians (creating the syllabus in medical genetics at the National University of Mexico) and the patients about the role of genetics in rare diseases such as Down and Turner syndromes. Armendares and his colleagues envisioned clinical work, medical research and educational programmes as endeavours that were needed urgently in clinical practice. This story is one of overlapping trajectories that involved institutions, physicians, practices and ideas that began to reshape human genetics that made the development of genetic counselling possible in Mexico in the 1960s.

Keywords Genetic counselling in Mexico • Salvador Armendares • Human genetics • Karyotyping • Cytogenetics

1 Introduction

In post-1945 not only were cultural and social processes reconfigured, sea changes occurred in the area of science itself, where the global circulation of scientific instruments, workforce and ideas had come increasingly into focus.

After WWII, extensive research and experimentation occurred not only in physics and chemistry but also in biology and medicine.[1] As stated by Cambrosio and colleagues, after the atomic bombs Western medicine resulted in the emergence of new practices based on the direct interaction of biology (especially genetics) and medicine. It was in those years, when global trends in human genetics were reshaping the field of biomedicine and when growing international interest in understanding the effects of radiation on human beings led to the formation of institutions and a proliferation of multi-centred clinical trials and inter-laboratory studies.[2] New techniques and practices were developed within human genetics (including population genetics and cytogenetics) as a medical field intended not only to characterize but also to understand differences among populations and their relation to the presence of certain diseases mainly in relation to radiation exposure. After 1945, there were many scientists and laboratories around the world that received public and private funding reflecting the policy concerns raised by radiation risk, atmospheric weapons testing and the rise of the nuclear power industry.[3]

As Lindee has shown, after the bombs, human genetics grew out of radiation risk and was transformed from "a medical backwater" to an appealing medical research frontier between 1955 and 1975. The production of technological knowledge was shaped by people in many different social and professional locations and resulted from the amalgamation of scientific knowledge acquired by many different people at many different times.[4] In the postwar years, physicians were taught almost

[1] Creager 2006; Krige 2006; de Chadarevian 2013.

[2] Cambrosio et al. 2006.

[3] Lindee 2015.

[4] Lindee 2005 and 2015; see also Harper 2008, Comfort 2012, Suárez and Barahona 2013.

nothing about heredity and both diagnostic capabilities and intervention were limited. By 1955, human geneticists were particularly important because of the widespread interest in the effects of radiation on human populations. In this context of political, social and industrial concerns on the genetic effects of radiation, human geneticists were seen as experts in the public debate.[5] It was during these years that Mexican physicians hosted and consolidated the emerging model of human genetics in the clinic (medical genetics) and used the knowledge and scientific practices that had recently been developed to tackle health concerns with clinical relevance to the social context of global developments.

On the other hand, human genetics underwent a profound reconfiguration following WWII, resulting in part from the horrors of Nazi eugenics and the use of human experimentation during the war.[6] To distance themselves from previous domestic eugenics programs, countries restricted their participation in genetic decisions affecting populations, focusing instead on individual medicine, and in certain cases, such as in the USA and the UK, restricting human genetics to family genetic counselling. It was also important that "in 1956, Joe-Hin Tjio and Albert Levan demonstrated that the correct number of chromosomes in humans was 46, and 3 years later, Jerome Lejeune and colleagues found that an extra chromosome (trisomy 21) was the cause of Down syndrome. Over the next years, researchers in Europe, North America, and Asia identified at least 100 chromosomal anomalies, including sex chromosome disorders such as Turner syndrome and Klinefelter syndrome. The medicalization of human genetics which accelerated in the 1950s and 1960s, helped to break human genetics out of the container of organized eugenics".[7] As we will see, in the 1960s Mexican physicians applied the knowledge and practices that had been developed abroad to establish a new medicine based on scientific principles and detached from eugenic principles.

This was a key moment when the tools and practices of medical genetics were being systematically used to attack global health problems with the support of international health organizations such as the World Health Organization (WHO). The creation in Mexico of national institutions such as the Departamento de Investigación Científica del Instituto Mexicano del Seguro Social (IMSS DIC, Mexican Institute of Social Security Scientific Research Department), within which the first unit on human genetics was founded, occurred during this international trend. In this sense, in the second half of the twentieth century the Mexican cytogenetic programme was in consonance with the international reconfiguration of human genetics and the establishment of genetic counselling.

A key player in this narrative is Spanish-born Mexican paediatrician Salvador Armendares Sagrera (1925–2010), who is considered the first Mexican physician to have undertaken graduate studies in human genetics. He spent 2 years at the British Medical Research Council in Oxford, England, in 1964–1965 under the supervision of Alan C. Stevenson (1909–1995). Upon Armendares' return from England in

[5]Lindee 2015.

[6]Kevles 1995; see also Stepan 1991, and Roll-Hansen 2010.

[7]Stern 2012, 23.

1966, the first Unidad de Investigación en Genética Humana (UIGH, Unit for Research in Human Genetics) was created at the IMSS, which had been founded in 1943 to provide medical assistance and health care to the workers at public hospitals. His former students, Colombian-born Mexican physician Fabio Salamanca (1940–) and Mexican physician Leonor Buentello (1940–), joined the recently created and promising unit a few years later. Salamanca had studied cytogenetics at the University of Minnesota in the USA and Buentello had returned from a 2-year stay in Freiburg, Germany, where she had studied virus genetics. The main objective of the research group at the UIGH was to provide specialized medical genetics services to the general public. The unit explored the effects of malnutrition on chromosome structure, child mortality, chromosome aberrations and also Down and Turner syndromes. Armendares and his colleagues transformed hospital medical practice into a medical research discipline and shared an interest in the population genetics of certain illnesses and the capacity to relate chromosomes to health conditions. The introduction of cytogenetics paved the way for the emergence of genetic counselling in the 1960s.

This story is one of overlapping trajectories that involved institutions, physicians, practices and ideas that began to reshape human genetics and made the development of human genetics and genetic counselling possible in Mexico in the 1960s. The actors involved in this narrative attempted to correlate clinical diagnosis with chromosome analysis, or karyotyping, using different techniques extensively applied to the study of human chromosomes at the time, particularly in Down and Turner syndromes, which not only allowed to visualize chromosomes, but also to professionalized and institutionalized cytogenetics. They made possible the transition from eugenics to medical genetics, from population management to clinical handling in the 1960s in Mexico, contributing to the establishment of human genetics and genetic counselling in the country.

2 Eugenic Background

During the nineteenth century, although other communities such as the botanists, zoologists and veterinarians had representation in the Mexican academic world, it was the medical community which was the most dedicated to the study of hereditary phenomena such as reproduction, diseases and malformations. The community of physicians developed the notion of heredity in the sense of appreciating certain traits of diseases that appeared repetitively in some bloodlines or as traits present in certain age groups that are considered incurable. At the end of the century, Mexican physicians looked to overturn old beliefs and false myths about mankind and its diseases. Disease became an experience accessible through observation, following the regularities of its manifestations,[8] allowing the use of new methods of study and changing the medical discourse radically. This discussion on

[8]Cházaro 2002.

heredity came before the introduction of Mendelian principles that occurred in the late 1920s in Mexico, but with the introduction of evolutionism in the 1890s in the country, the vision that heredity was the passing on of joint mental and physical qualities from parents to offspring was consolidated.[9] One of the consequences of the expansion of the empirical base of heredity on other biological phenomena was the diversity and types of conceptions of heredity. When connecting heredity with reproduction and generation, the problem of which characteristics are passed on and why emerged.

The Mexican medical community at the turn of the twentieth century accepted that what defines a disease was the combination of what was passed on and the environment, between the make-up received from the parents in conception and what occurred in the uterus and the exterior. These conceptions of heredity proposed that physical as well as moral traits can be passed on, including diseases, malformations or defects. Thus, clinical, therapeutic and prophylactic tools were designed for the study, treatment and prevention of some diseases and physical traits, with significant influence from eugenic ideas. For example, many Mexican physicians used their ideas about the disadvantage of consanguineous marriages to promote ideas of racial improvement.[10] According to this position, the State should implement a policy of selective control over reproduction in order to achieve a society free from illnesses and vices, thereby avoiding social degeneration. The "social engineering" of the first post-revolutionary governments aimed to intervene in two factors causing backwardness: social degeneration and racial diversity. For the first, physicians proposed restricted reproduction; for the second, anthropologists proposed racial mixing.[11]

Eugenic principles were as popular in Mexican medicine during the late 1800s as in countries such as Britain, France, Germany and the USA.[12] Although many Mexican physicians had been discussing social degeneration since the end of the nineteenth century, it was not until the first decades of the twentieth century that a hygiene-oriented forum for eugenics was established within the domestic medical community. Eugenics and mental hygiene were therefore two aspects of medico-hygienic thought that turned heredity into an important factor in the transformation of Mexican society. As in other countries, the protection given to women was ambiguous, because on one hand maternal and child health, sexual education, responsible motherhood, contraception and abortion were favoured, but on the other, it was thoroughly affirmed that the natural domain of women was the family and their main function was procreation.[13]

In Mexico in the early decades of the twentieth century, eugenic measures were mixed with health prevention ones, due to the lack of accurate knowledge about inheritance. Post-revolutionary governments created health institutions such as the

[9]Barahona, 2010.

[10]Barahona 2010.

[11]Urías Horcasitas 1996 and 2001.

[12]Bashford 2010.

[13]Urías Horcasitas 2003.

Hygienic Information and Education Service, the School Hygiene Service and the Infant Hygiene Centres, to reduce infant mortality, instil responsible motherhood and ensure the well-being of newborns.[14] Previously, the first Mexican Congress of the Child was held in 1921, with the second a few years later, to advance an understanding of Mexican children from a eugenic, hygienic, legislative and pedagogic standpoint. These congresses and institutions demonstrated how prevalent eugenics had become in socio-medical discourse within Mexico.[15]

The majority of physicians who formed the movement were educated at the *Universidad National Autónoma de México* (UNAM, National Autonomous University of Mexico), a place heavily influenced by the French hygiene movement[16] and with few resources for carrying out scientific research.[17] For example, in many Latin American countries, scientists and medical doctors embraced and promoted puériculture (the scientific study of the child) and eugenics, influenced by French philosophy and medicine.[18]

According to Stern, in Mexico, as in many other countries during this period, the central doctrines of nationalism and citizenship included theories of hereditary differences, reproduction control and anthropometrics. This led to the foundation of the *Mexican Society of Puericulture* in 1929, which included a section for eugenics devoted to heredity, reproduction-related diseases, infantile sexuality, sex education and birth control; the future founders of the *Mexican Eugenics Society* (1931) emerged from this organization.[19] Most of the community's members had medical knowledge and frequently mentioned the works of Mendel, Galton and Weismann, though without discussing their ideas. Knowledge of Mendelian inheritance was absent until the1940s when Mendel's laws were introduced and discussed in academic circles, so their eugenic ideas were related more to the effects of the environment and framed primarily in the field of childcare and marital and birth control.

Members originated from the inner circles of Mexican politics and public health and included biologists, physicians, judges and criminologists. As Schell has noted, "this roster of membership indicates how eugenics influenced policy and practice in law, health care, the sciences, and education".[20] With the decline of eugenics at the international level, Mexican physicians also abandoned eugenic principles.[21] In the 1930s, Mexican eugenicists tended not to support Mendelian genetics but rather the inheritance of acquired characteristics. After the international discrediting of

[14]Carrillo 2002.
[15]Schell 2004.
[16]Stepan 1991.
[17]Suárez y López Guazo 1999, 2000 and 2002.
[18]Birn 2011.
[19]Stern 2005.
[20]Schell 2010, 485.
[21]Schell 2010.

Lamarckism in the late 1930s, Mexican eugenicists began to accept Mendel's genetics and moved away from concerns with sexual and reproductive behaviour.

3 Karyotyping and Genetic Counselling in Mexico[22]

Many physicians belonging to the *Eugenics Society* actively participated in Mexican health institutions, the programmes of which were focused on increasing the level of public health standards and population density, and were instrumental in the creation of governmental institutions, such as the *Ministry of Public Assistance* in 1937 and the IMSS in 1943.[23]

The IMSS headquarters were inaugurated in 1950, and construction of clinics and hospitals soon followed. The first hospital centre, known as Centro Médico "La Raza" (*La Raza Medical Centre*), was established in 1952, and the *Centro Médico Nacional* (CMN, National Medical Centre), where medical research formally began, was inaugurated in 1963. Mexico was also undergoing a progressive movement in gynaecology and obstetrics in the IMSS, particularly in *Gynaecology and Obstetrics Hospital #1*, called *Gabriel Mancera Hospital*. Although research was not one of the original objectives of the IMSS, which was created with a strong clinical focus, research groups began to be formed in the 1960s at the General, Gynaecology and Obstetrics and Oncology Hospitals. It was in 1966 that the IMSS DIC was established.

In Mexico, the first studies on chromosomes were performed at the IMSS by Armendares and his former students Salamanca and Buentello. Their work was based on the study on congenital malformations performed by the WHO that Mexico had participated in along with other 15 countries, which was carried out by Alan C. Stevenson. It was in 1958 that a prospective study of congenital malformations was implemented in a number of countries under the auspices of the WHO.[24] This study was conceived as a step towards understanding the occurrence and types of congenital malformations found in stillborn and live-born infants in several countries. The WHO asked James Neel (1915–2000), William J. Schull (1922–2009), J. A. Fraser Roberts (1900–1987) and Alan Stevenson to prepare a document. Once accepted, the WHO asked Stevenson, then head of the Population Genetics Research Unit of the MRC, to organize and carry out the study.[25] Stevenson was particularly interested in single gene disorders from a population genetics perspective and attempted to determine their frequency in Northern Ireland

[22]This section is a revised and expanded version of Barahona 2015.

[23]Stern 2005.

[24]The countries chosen were Australia, Brazil, Chile, Colombia, Czechoslovakia, Egypt, Honk Kong, India, Malaysia, Mexico, Northern Ireland, Panama, The Philippines, South Africa, Spain and Yugoslavia (Stevenson 1966).

[25]Harper 2012.

within the context of social preventive medicine. In 1958, Stevenson was approached by the MRC to set up a population genetics unit in Oxford, where he studied the burden of genetic diseases.[26] The recording of information began in 1961 and ended in 1964. Among the findings of particular interest were "the large contribution of neural tube defects to foetal wastage in most countries and the significant correlations of frequencies of these defects over the 24 recording centres; the unexplained correlation in frequency between neural tube defects and dizygotic twinning; the marked association of consanguinity of parents with increased stillbirth rates and frequency of early death of the infant, these frequencies being highest when parents were most closely related; and the demonstration that, if malformations known to be due to the expression of single recessive gene mutations are ignored, consanguinity of parents is demonstrably associated in these data with neural tube defect frequencies only".[27] Other malformations recorded were harelip and cleft palate, malformations of the gut and urogenital tract and Down syndrome.[28]

When Stevenson visited Mexico in 1960, he met Armendares, working at that time at both the *CMN Gynaecology and Obstetrics Hospital* and at the *CMN Hospital de Pediatría* (HP, Paediatric Hospital). Armendares decided to study at Oxford with Stevenson and spent 2 years at the *British Medical Research Council* (MRC) in Oxford in 1964 and 1965. Upon Armendares' return from England in 1966, the first UIGH was created at the IMSS, with the main objective of providing medical genetic services in a clinical setting. Armendares and the colleagues, who soon joined the unit, shared an interest in the population genetics of certain illnesses and the capacity to correlate chromosomes with health and living conditions. They were aware of the growing importance of chromosome studies in clinical practice, particularly concerning genetic counselling for certain diseases such as Down syndrome. Neonatal testing through karyotyping was performed at the unit in close collaboration with medical personnel at the hospital, trying to correlate clinical observations with chromosome abnormalities.

The UIGH was the first medical genetics unit created in Mexico, and Armendares held the initial directorship from 1966 to 1976. At first, the unit was based in a small room in the basement of the HP, with personnel consisting of Armendares and two technicians, but in 1968, the unit moved to its own space in an adjoining area, where Salamanca, Buentello and more personnel could be employed and more equipment acquired. In this new institutional setting, equipped with a modern laboratory for chromosome analysis, Armendares began to provide genetic counselling, both to patients referred to him from hospitals and to walk-ins.

Soon after having established the cytogenetics laboratory, Armendares started a Medical Genetics Graduate Program within the CMN HP in 1969, endorsed at the time by the Graduate Division of the UNAM School of Medicine. The syllabus

[26]Stevenson 1961.

[27]Stevenson 1966, 9.

[28]Stevenson 1966.

included the biological basis for heredity, cellular biology, developmental biology, cytogenetics, clinical genetics and human population genetics.[29] He was convinced, after the 1962 *WHO Expert Committee Report on Human Genetics* regarding the teaching of genetics in medical schools that physicians should be trained systematically in genetics and that these courses would provide them with the knowledge and technical skills required in their medical practice. According to this report, attention will be directed towards the minimum requirement for instruction in genetics at both preclinical and clinical levels of medical education and in postgraduate training. Thus, courses should be available in basic human genetics, human cytogenetics and other subjects essential for competent genetic counselling.[30] According to this report, the development of cytological techniques for the study of human chromosomes and the consequent discovery that chromosomal aberrations cause a number of pathological conditions have led to a greatly increased demand for instruction in genetics among clinicians and research workers in essentially all fields of medicine and medical biology.[31] It's worth noticing that Dr. Fraser Roberts was a member of the WHO Expert Committee on Human Genetics, whom Armendares had been acquainted with from the study of Congenital Malformations led by Stevenson in Mexico. This could have made it easier for Armendares to be aware of the WHO reports on human genetics and be well informed of the new developments in the field.

The first generation of students at the CMN HP included Salamanca and Buentello, who became colleagues and close friends. By the time they joined the unit, Armendares was performing the cytogenetic techniques he had brought back to Mexico from Oxford, such as lymphocyte culture techniques for chromosome analysis developed by Hungerford and colleagues in the USA.[32] The combination of Armendares' clinical experience (the only one of the three who had practised medicine at the hospital), Buentello's technical skills, Salamaca's research experience and their collaboration with international networks in the circulation of knowledge, together with the population's growing need to access the public health system, facilitated research into cytogenetics in Mexico at a time when research on human genetics was becoming a medical domain for diagnosis. The data were routinely collected by the bureaucratic system at the IMSS in close collaboration with Armendares and colleagues. This system made it possible for the UIGH to carry out neonatal testing through cytogenetic analysis and, when necessary, to provide genetic counselling to the parents of the sick children. In this way, genetic counselling emerged as an integral component of medical genetics in the 1960s when scientific and technical factors converged[33] and were conceived as a service

[29]The discipline was consolidated in 1988 with the establishment of the first graduate programme specializing in medical genetics.

[30]WHO 1962, 21.

[31]WHO 1962, 4.

[32]Moorhead 1960.

[33]Stern 2012.

for couples to help them understand and solve problems related to the risk of having genetically abnormal offspring. It was based on the knowledge of genetic abnormalities that help predict the risk of an abnormal offspring. The principles of genetic counselling were those of Mendelian inheritance and population genetics that the members at the UIGH had learned abroad. For them, increasing knowledge of human genetics undoubtedly strengthens genetic counselling. Genetic information applied to assess future risks of disease has shaped its social and political meaning and its medical uses in Mexico as in other parts of the world. Armendares, Salamanca and Buentello, in a two-way traffic, were responsible for taking the samples and conducting the genetic analysis, but only Armendares and Buentelllo were responsible for talking to the patient's parents in order to give them assistance. For them, the importance of the clinic in medical practice was, in Lindee's words, "that intimate site where disease, risk, genetics and scientific knowledge coalesced around a key social actor, the patient".[34]

4 Down and Turner Syndromes

In 1964, the WHO called attention for the high incidence of Down syndrome, the frequency of which has been given as 1.5 per 1000 total births, and in some geographical areas even bigger. These differences were attributed to differences in maternal age distribution in different populations. "The incidence of the disease varies from population to population. . .. In Europe and North America, it is about one per 150 births, while in Japan the incidence appears to be as low as one per 5000 births".[35] So according to this report, attention should be given to have reliable data on this genetic condition.

According to Stevenson's study on congenital malformations, Down syndrome in Mexico was unusually common, occurring one per 500 births. The study also revealed that in Mexico City, the proportion of all pre-28th week losses that occurred between the 17th and 27th weeks was far higher than elsewhere, indicating that further analysis of the information collected might serve to identify the characteristics of a high-risk group of mothers and provide etiological clues.[36] For the Mexican medical authorities, this problem needed to be tackled by Mexican physicians in order to have early diagnosis of the syndrome as, before performing karyotypes, children with Down syndrome in Mexico had been diagnosed due to a smaller size in the iliac index of the hip.[37] With the introduction of new laboratory techniques to study chromosome structure in the late 1960s, Armendares and

[34]Lindee 2015, 51.
[35]WHO 1964, 21.
[36]Stevenson 1966, 104.
[37]Armendares 1967.

colleagues used a double approach, medical practice and cytogenetics, that combined could give a more accurate diagnosis of genetic diseases.[38] Buentello was responsible for the supervision of blood sampling to ensure the correct identification of children and for monitoring those patients whose parents had given written authorization to use the material for research. In this way, Armendares' agenda was highly suited to both global and local priorities.[39]

More attention was paid to the studies for identifying Turner syndrome, which was not yet well understood.[40] Since the 1960s, results had already begun to be published on gonadal dysgenesis in patients with Turner syndrome.[41] In collaboration with pathologist Héctor Márquez-Monter (1945–), Head of the Pathology Department at the Biomedical Research Unit of the CMN and who had studied at the Anderson Hospital in Houston, Texas, Armendares and Salamanca also found that most Turner syndrome patients showed only one chromosome in the sex pair,[42] which was in agreement with earlier results by Paul Polani (1914–2006) and Charles E. Ford (1912–1999) in England. In samples from gonadal biopsies, a series of chromosomal variants appeared in the population studied. They performed gonadal histopathologic analysis only when there was the suspicion of the presence of a part or parts of the Y chromosome or complex mosaicisms.[43]

In his monograph, *Turner Syndrome, Diagnosis and Therapeutic Handling,* Armendares gives a detailed description of the medical characteristics of the syndrome, frequency in the population, its chromosomal classification, clinical characteristics and the correlation of the phenotype to the karyotype, sexual development, intelligence quotient, treatment and counselling.[44] The book became very influential in the Mexican medical community because it helped with the early diagnosis and clinical management of patients suffering from this syndrome.

Following the 1964 *WHO Report on Human Genetics*, for Armendares and colleagues the key factors underlying advice to the patients with some genetic disease or syndrome were accurate diagnosis, the individual family history and the background of the literature. "It is the first of these factors that makes preliminary examination by the appropriate specialist so desirable."[45] They knew also the 1969 WHO report on genetic counselling that stated that the clinic (hospital) needs to be

[38]Armendares 1968; see also Santesmases 2014.

[39]Armendares 1968.

[40]Ha 2015.

[41]Ford 1959.

[42]The nuclei of human cells contain 22 pairs of somatic chromosomes known as autosomes, and one pair of sex chromosomes responsible for the development of an individual's sexual characteristics, XX for a female, and XY for a male. In Turner Syndrome, females lack one X chromosome, being XO.

[43]Márquez-Monter 1972.

[44]Armendares 1979.

[45]WHO 1964, 28.

close to laboratory techniques that human geneticists have particularly developed: karyotyping and analysis of dermatoglyphics.[46]

As well as finding the correlation between clinical observations and cytogenetic studies, Armendares and collaborators embarked on a study to measure the burden imposed on public health resources by genetically determined illnesses, evaluated in terms of morbidity and mortality. Armendares had been inspired by Stevenson's study on genetic diseases in the population of Northern Ireland.[47] From the standpoint of Armendares and colleagues, the lack of recognition for the repercussions and importance of genetic illnesses resulted from the scarcity of comprehensive studies in genetics. Without such studies, public policies to manage and treat genetic illnesses could not be established. They assumed this information would, to a certain extent, help to predict future social needs. Relevant measures could therefore be implemented, such as training the necessary number of specialists in medical genetics, preventive medicine and counselling, and, as far as possible, preventing the occurrence of genetically determined diseases through genetic diagnosis of the parents.[48]

In his 1968 book *Citogenética Humana* (*Human Cytogenetics*), Armendares included a chapter on genetic counselling as being "the most important practical application of human genetics knowledge", quoting the 1964 WHO report that stated: "A further important point is that genetic counselling may facilitate early diagnosis, and this may be a major factor in instituting successful treatment".[49] For Armendares, cytogenetics was not only of scientific interest but has stimulated medical and lay interest in genetic counselling in Mexico.

Armendares and his colleagues were the first to relate karyotyping with genetic counselling, translating test results and technical language for the patients or their parents at the hospital. They played a key role in educating physicians and the patients about the role of genetics in rare diseases such as Down and Turner syndromes. Armendares and his colleagues envisioned clinical work, medical research and educational programmes as endeavours that were needed urgently in clinical practice.

During the 1970s, genetic counselling was provided only by individual physicians and scientists with an interest in and knowledge of genetics. Since then, chromosome analyses had become more routine, which created the need for a more systematized genetic counselling. Nevertheless, this need couldn't be fulfilled since

[46]WHO 1969. It is worth noticing that Mexican physician Rubén Lisker, a close colleague and friend of Armendares', was part of the WHO Working Group on Genetic Counselling as the Mexican representative. Thanks to his belonging to this international network on genetic counselling, Lisker gave Armendares advice on this matters (Salamanca, personal communication, March 2016).

[47]Stevenson 1959. Stevenson found that approximately 26% of the beds at the hospital were occupied by patients with a genetic disorder.

[48]Armendares 1974.

[49]WHO 1964, 5; Armendares 1968, 234.

prenatal diagnosis could lead to the decision of halting pregnancies of children with genetic defects or the undesired sex. This possibility made genetic counselling practically unavailable largely due to a religious group's efforts to block attempts at modifying laws against abortion, which was illegal in the country at the time. Thus, prenatal diagnosis was poorly developed and available only in certain public hospitals. It was available in a few private hospitals in the larger cities, mostly for the purposes of cytogenetics diagnosis.[50]

From the 1990s, geneticists have generally been trained "to provide nondirective genetic counselling; however, they have little involvement in the medical care of the patients. Geneticists participate in the diagnosis of genetic disorders, provide counselling to the family on as many occasions as needed, and refer the patients to other specialists for specific medical and surgical treatment".[51] According to the 1995 report on Genetic Counselling by the UNESCO International Bioethics Committee, in Mexico genetic counselling is done mostly after the birth of a child with genetic disease or malformation. It is usually done in the outpatient clinic. Pregnant women afraid of having an affected baby, or consanguineous couples planning to marry, are also counselled.[52] "The increasing availability of genetic tests confers to the specialty of genetic counselling a fast growing place in medical practice. Genetic counselling provides the link between genetic technologies and the clinic".[53] In the last decade of the twentieth century in Mexico, formal genetic counselling was performed by trained physicians. Many counsellors have at least 1 year of previous training in paediatrics and most of them take a 2-year graduate course in medical genetics, which are available in several of the larger cities in the country. They are certified by the National Board of Medical Genetics.[54] The demand for genetic services has been growing in Mexico since the 1990s and physicians are facing ethical issues concerning counselling, but little is known about their attitudes and positions regarding genetic counselling, prenatal diagnosis and selective abortion.[55] These concerns need to be tackled and more studies need to be performed.

[50]In 2007, the Legislative Assembly of the Federal District of Mexico City approved the decriminalization of abortion at the woman's request until 12 weeks of pregnancy. However, more than half of state constitutions have been amended to define a fertilized egg as a person with the right to legal protection. Since 2009, none of these states has removed its objections to abortion. These controversies have put a brake on the development of genetic counselling as a medical practice.

[51]Carnevale 1997, 24.

[52]Genetic Counselling 1995.

[53]Genetic Counselling 1995, 2.

[54]Genetic Counselling 1995.

[55]Carnevale 1997.

5 Conclusions

With the development of cytogenetics in the 1960s and 1970s, human chromosomes were visual images that deserved medical attention. They became visual markers of pathologies and critical images in a new, broader conception of genetic disease.[56] Before the development of cytogenetics and karyotyping, genetic conditions could only be seen or detected in the pedigrees the historical reconstruction of a family's history, and the clinically abnormal body.[57]

New technologies like karyotyping were being standardized and implemented at the UIGH, paving the way for genetic counselling to be performed. Karyotyping was a central practice in the early days of human genetics in the country "to start a rudimentary form of genetic counselling emerged in response to an increasing demand for genetic risk information and risk calculations".[58] As Stern has shown for the USA, the founding of the UIGH in Mexico "elucidated how studying human heredity entered the world of academic science and medicine and introduced medical genetics to patients and physicians".[59] The community of scientists described in this work demonstrates the importance in the clinic of new medical genetics practices learned abroad. This case also reveals how important the circulation of knowledge was in the formation of Mexican scientific elites, as well as demonstrating the national and transnational concerns that shaped local practices.

Armendares and colleagues participated in the early diagnosis of genetic diseases and revealed the correlation between clinical observations and karyotyping in the Mexican health system. At the local level, they were regarded as scientific experts, yet they were also political actors whose authority derived from their belonging to international networks. Armendares and Buentello were the link between the clinic and the laboratory, between the bed and the bench. Armendares was involved in a rudimentary kind of genetic counselling due to the insufficient number of trained personnel to meet the demands for genetic services. Buentello was responsible to obtain the informed consent to perform the tests and to disclose the information in an understandable way. Salamanca, on his part, was more dedicated to the standardization and stabilization of the newly developed techniques in the laboratory setting. The work of Armendares and his colleagues at the UIGH was instrumental in the development of cytogenetics and genetic counselling in the late 1960s in Mexico.

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[56] Lindee 2015.

[57] Lindee 2005.

[58] Björkman 2015, 492.

[59] Stern 2012, 32.

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Newborn Screening on the Cusp of Genetic Screening: From Solidarity in Public Health to Personal Counselling

Margherita Brusa and Michael Y. Barilan

Abstract The chapter opens with a history of newborn screening, highlighting the factors behind its worldwide dissemination and emergent controversies. The second part of the chapter explicates newborn screening as a hybrid of public health and clinical care. The third part of the chapter delineates a framework for democratic governance of newborn screening, which is based on the value of solidarity and which is attuned to possible advent of screening tests at the genetic level. This governance aims at deep participation of the public, and the empowerment of autonomous choices of individual patients.

Keywords Public health (paediatrics) • Genomics (genetic tests and databases) • Solidarity • Newborn screening • Public participation

1 Introduction

In the last few decades, every baby born in the developed countries has been subjected to a panel of blood tests aimed at the early detection of some inborn diseases. The procedure is known as "Newborn Screening" (NBS). In the 1970s and 1980s, newborn screening was considered an uncontroversial successful public health initiative. In the past 20 years, screening has been expanding at three different levels: more conditions are screened, larger populations covered (as more countries initiate screening), and additional biochemical and genomic technologies employed. Public debate and regulative structures of NBS have been expanding as well, reflecting a growing awareness of the ethical, legal and medical aspects involved, almost exclusively in relation to the expansion of conditions screened.

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In fact, two processes have been developing in parallel to each other—the maturation of the doctrine of informed consent, and the advent and expansion of NBS. The maturation of informed consent is part of a broader turn towards human rights-based medical law, and the expansion of NBS is part of the interaction between biotechnology and information technologies. No less important are the roles of two social phenomena that loom large over the construction of NBS services—the emergence of patients' advocacy groups, and the transition from home birth to hospital birth. The temporal coincidence of these and other factors have moulded the NBS systems and its ethos of practice.[1]

In its first phase (1960s–1990s), NBS included two biochemical tests. In its expanded phase (twenty-first century), screening programmes vary considerably, some covering over 50 conditions. Some professional organizations call for genetic screening, thus heralding a possibly third phase of screening.

Because the expanded programmes posed a challenge to the established framework and guidelines, in many countries, intensive efforts at public participation in the restructuring of these programmes accompanied the second phase of NBS, mainly in the format of advisory committees. These efforts resulted in a sort of contradiction. On one hand, almost each state and jurisdiction has employed different configurations of public participation in the regulation of NBS.[2] On the other hand, academic scholars, activists and regulators have expressed dissatisfaction with the guidelines that evolved from diverse modes of participation. They actually protest against the absence of uniform standards for NBS and behold such standards a desired near-future goal.[3]

Although freedom to choose and to elect representatives satisfies liberal requirements of legitimization,[4] especially in relation to medical care and decisions affecting the person and body, there is a moral quest for evidence of significant participation. The public participation and personal choice aimed at must not be shallow and technical, but deep and respectful of personal autonomy.[5] The broad diversity among NBS programmes is a reason to suspect that, even though every programme as such has been instituted legally, deep participation and reflection on the nature of the service and on its related shared basic values has been lacking.

In this chapter, we explicate a normative model for integration of public participation and personal choice in public health policies. The model will draw from a liberal and human dignity committed conceptualization of the value of solidarity.

The enterprise involves a few steps. We begin with a historical survey of NBS. The historical overview exposes the dual nature of NBS—public health and clinical medicine. Then, we discuss the complexities of public participation in NBS. We argue that solidarity may serve as a normative template for the design and regulation of NBS in democracy. The operations of solidarity within democracy and the dual nature of NBS may explain some of the peculiar features of NBS services.

[1] Brusa and Barilan 2017.

[2] Jennings 2009.

[3] Padilla 2012; Loeber 2012; Burgard 2012.

[4] List 2006.

[5] Arenstein 1969; Needham 2011; Barilan 2011.

2 History of Newborn Screening

The history of NBS entails four conceptual transitions. The first is the rise of scientific tests that are expert domain. The second is the maturation of the responsibility of the state for "children at risk". The third is the expansion of the notion of "disaster" to health problems. The fourth and last is the expansion of public health concerns from threats to the public to risks looming over the few. Altogether, the state has a duty to mobilize public health resources in order to trace out the few at risk to a medical catastrophe and rescue them.

Efforts directed at a universal screening of babies started at the beginning of the twentieth century. It coincided with the medicalization of state efforts to protect poor babies from neglect and abuse, especially "baby farming", which were illegal businesses of foster care and adoption. In the early twentieth century, UK law required a parent to notify the public health officer about the birth of a child within 36 h (Notification of Birth Act, 1907).[6] The grass-roots humanitarian "weight charts" movement aimed at measuring the weight of every baby in order to detect growth retardation, either due to illness or deficient care. The weight chart movement was a grass-roots voluntary effort, carried out by public health nurses in clinics and home calls, aided by growing state concern for the welfare of babies as future soldiers and workers.[7] The weight chart movement was about the health of all babies.

Even though the monitoring of babies' growth had become integral to paediatric follow-up, owing to lack of standardization, already in the 1930s the medical community abandoned the screening endeavour. Too many "low weight" babies were healthy; too many sick and maltreated babies were not underweight.[8] Chart weights (growth charts) came back as an integral part of paediatric follow-up once the complexity and interaction with other indexes were understood better and interpreted holistically.

Whereas this first wave of screening aimed at a common problem (unhealthy and improperly nourished babies), using one simple index (weight), the second wave of paediatric screening aimed at very rare conditions, using technologically sophisticated means. Screening became an experts' domain.

In 1961 Robert Guthrie (1916–1995), a microbiologist from Buffalo, New York, developed a simple and cheap assay for measuring phenylalanine levels in the blood. This facilitated mass testing of babies for phenylketonuria (PKU)—a rare inborn error of metabolism—already by the third day of life, before it developed

[6]The history of NBS has a sidetrack here. Most screening services today are large-scale national (or regional) operations. Many experts believe that such large IT infrastructures are necessary for the elucidation, prevention and treatment of very rare conditions. The link between public registries of births and deaths on the one hand and paediatric public health is traceable to the English 1836 Civil Registration Act (Goldman 1991).

[7]Klaus 1993.

[8]Brosco 2001.

into a devastating disease. Although it had been already known that a special diet for the infant could diminish the neurological damage and it was possible to detect some newborns by a different biochemical test of their urine, until then, almost all babies with PKU had been diagnosed clinically, after serious harm had set in. In the 1940s and 1950s, the diagnostic criteria, natural history and impact of dietary modifications were not yet mature, owing, mainly, to dependence on clinical diagnosis made on sick children. With the new test, caregivers could initiate dietary therapy promptly. Then, it was shown that the neurological damage is fully preventable.[9] Because Guthrie's invention almost concurred with the approval in 1958 of a commercial dietary formula for PKU, the canonization of the diagnostic test coincided with the canonization of its treatment. Guthrie was also active in an advocacy organization dedicated to retarded children—NARC (National Association for Retarded Children), and even before his results were fully published in a scientific journal, NARC had campaigned for large-scale screening for PKU.

During the first half of the twentieth century, the cultural panorama in Western countries, and especially the USA, was changing in favour of taking more responsibilities over children's health and development. When the humanitarian volunteers and activists were responsible for the weight chart movement, it was about *the health of all babies*. Now the USA undertook responsibility for *the* saving of "disaster-stricken" children, which was a small minority of babies. This followed the logic of the state's response to the Mississippi flooding in the 1920s, and the conceptualization of the Great Depression as a disaster, similar to natural disasters.[10] In situations of disaster (natural disaster or man-made disaster), the State would actively intervene in order to save a selected group of affected people.

This new concept of disaster spilled over the medical realm as well. When President Kennedy announced the dedication of federal funds for research on mental retardation, a clear sign was given that catastrophic inborn diseases are not private tragedies anymore; they have become a public concern.[11] According to the new paradigm of state responsibility to "disasters", in order to be a public concern, public health does not require risk for the public anymore. Rather, public action that can reduce mortality, even at a very low scale, is a public health matter, and a state's responsibility as well, worthy of federal spending in the name of "rescue".

This evolving sociopolitical mood of responsibility for afflicted children coincided with a specific invention. For the first time, it was possible to apply a scientific test to an apparently healthy newborn child and foretell an imminent but preventable catastrophic disease. A promise of cure by means of early detection was on the horizon. The light burden of screening was measured against the alternative of lifelong institutionalization. Guthrie's invention showed the value of mass medical testing of "apparently healthy" babies. Even though PKU is quite rare, affecting

[9]AAP 2000.

[10]Landis-Dauber 2013.

[11]Kennedy 1964.

1/15,000 in the USA, the combination of easy and reliable testing in terms of costs and administration (in the 1960s, over 95% of US births took place in a hospital) rendered it reasonable to expect of the public to screen every newborn child, investing $7500 in the prevention of one case of lifelong severe mental retardation. Guthrie waived financial gains for his invention,[12] and NARC expected the public to pay the 50 cents cost of each test.[13] In the "fee-for-service" milieu of American medicine and its entrepreneurial spirit, the pioneer of NBS baptized it as an essential public good, entrusted with the agents of the state.

The advent of screening to PKU marked a conceptual revolution in the history of medicine. A universal medical procedure was targeted at a single rare condition, rather than on a common risk. NBS became mandatory in many states, even though it involved neither contagion nor other threats to the public. A third groundbreaking factor was the leadership of advocacy groups in the establishment of a public health programme. In the beginning, opposition to NBS was coming from clinical doctors and medical researchers, who expressed worries about state intervention, "socialized medicine" and the risks inherent in the implementation of large-scale testing immediately after the technology is developed and when much is still unknown about its proper use and implications. While the professionals expressed caution with a new technology, the advocacy groups contributed to the formation of NBS as a universal (i.e. aimed at every neonate in a given jurisdiction) and publicly subsidized, usually mandatory, service.[14]

In 1973, when NBS for PKU was already an established practice in the USA and many other countries, Canadian researchers developed a diagnostic test to congenital hypothyroidism, another condition that if left untreated results in severe and irreversible retardation. It was also much more common than PKU. Besides, the hypothyroidism test could be performed on the same blood sample drawn in the context of PKU screening. Other screening tests proposed requested separate blood sampling from the child and different lab techniques as well. Owing to these barriers, almost all NBS programmes were limited to PKU and hypothyroidism. Efforts concentrated on severe conditions that can be detected with very high accuracy, at low cost and for which early detection was a necessary condition for the initiation of highly cost-effective treatment. This framework fit the WHO criteria for screening programmes.[15]

In the 1990s, tandem mass spectrometry (MS/MS) was introduced, allowing the testing of numerous metabolites in a few drops of blood. With the purchase of this costly technology, the added cost for every metabolite tested became marginal. By

[12]Koch 1997, 44.

[13]Paul 1999; Paul 2008; Committee 1975, 24–28.

[14]Therrell 2001; Paul 2008; Ross 2011.

[15]Wilson 1968; WHO 1968; Committee 1975.

the late 2000s, most countries which have deployed MS/MS technology have expanded their NBS programmes significantly. Most such countries screen for more than 10 conditions and a few US states for over 40. Most of the added conditions are not as devastating as PKU and hypothyroidism; treatment is less definitive, and it depends less on early detection.

The transition from biochemical tests to NBS by means of DNA analysis seems imminent. It will allow direct access to genetic knowledge and the testing of thousands of genetic markers in a single swath.[16] One of Wilson and Jungner's criteria for screening is "acceptability to the population".[17] Many people, clinicians and ethicists have come to question the circumstances in which expanded, let alone genomic, mass newborn screening might be "acceptable", if at all. Communication with the public seems to be essential for answering this question; it is also a prerequisite to the empowerment of patients and to public legitimization.[18]

Perhaps, the most far-reaching and unexpected impact of NBS is found in the medicalization of childbirth and its financial burden.

Already in 1965, when the rate of hospital birth in America was at its peak, the American Academy of Paediatrics recommended that screening be carried out prior to the baby's discharge from the hospital.[19] Because screening for PKU (and some other conditions) can be performed only after 36 h have passed from birth, and because insurers tend to set strict limits on hospital stays, NBS has rendered two nights of hospitalization a de facto universal standard for uncomplicated birth. Undoubtedly, shorter hospital stays or home birth does not exclude the possibility of NBS, but hospital birth is evidently the only way to ensure cheap and universal screening of every newborn child.[20] Thus, NBS soaked its appeal as a public policy (rather than a fee for service offered by doctors on an individual basis) from the practice of universal hospital birth, and shaped the standard range of hospitalization in a society that has enshrined hospital birth as the standard of birth. The programme administrators integrated NBS as part of childbirth, similar to the registration of the child's birth, not as a medical intervention. When the home birth and women health movements broke out in the 1970s, NBS went unnoticed. Activists battled "unnecessary" screening tests of women.[21] The feminism-driven "women health movement" protested the alleged patriarchal conceptualization of women as wives and mothers. Claiming women's potential for non-domestic modes of flourishment, the movement tended to minimize the importance of women's

[16]Dhanda 2003; Goldberg 2012.

[17]Wilson and Jungener 1968, 31.

[18]Burgard 2012, 620; Timmerman 2010.

[19]Committee 1965.

[20]Braveman 1995.

[21]Weisman 1998, 150.

reproductive roles in society.[22] Promotion of women's privileged position in relation to NBS calls upon these downplayed roles. The Women's Health Movement was about self-determination in relation to one's body and self, not in relation to one's maternity as such.

Both NBS and hospital birth are unified packages of medical surveillance that guarantees protection of babies from rare complications; it is about a pathology-oriented cultural construction of childbirth, of gearing up the natural event of birth in preparation for the worst, as to optimize overall safety, even at the price of minor complications and occasional discomforts. This results in an apparent clash between the clinical and the public health perspectives. If a woman is about to choose for herself, she may not consider the risk of one to a thousand worthy of attention. However, when a large sovereign state assumes responsibility for every vulnerable child, attention to this level of risk saves a few people every month.

Routine physical exam by a paediatrician and hearing tests of every newborn are additional forms of screening. Some health care settings also screen for heart lesions and renal tumours. However, these tests do not involve blood sampling and retention of biological material containing DNA. The standard of 2–3 days of hospitalization also fits the recommended period of "medical observation" prior to discharge.[23] Thus, the waiting period mandated by NBS has become a screening instrument in its own right and a template for further screening tests to monitor for possible medical complications.[24] Even though all of these tests and procedures deserve the title "screening" (the application of a test on an asymptomatic population with the intent of detection of a hidden medical problem), only the heel prick sampling of blood for biochemical disorders belongs to the system and rationale of "NBS".

The French Revolutionary constitution of 1791 included a universal registration of every childbirth. The 1907 English "Notification of Births Act" rendered mandatory registration with the public health office separate from civil registration. The notification of births would allow public health inspectors to perform home calls and monitor babies' health. Human rights law of the second half of the twentieth century beholds universal registration of births a necessary instrument of public health (Szreter 2007).[25] The large-scale instruments of information technology have rendered NBS services huge forms of civico-medical registries, whose genetic future looms large over fundamental issues of personal liberties, rights and civic status.

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[22]Ruzek 1979, 193–196. This marginalization of maternal roles was in sharper contrast to the early twentieth-century emphasis, mainly in the UK, on education and inspection of mothers as key to reduction of infant mortality (Dwork 1987, Chap. 5). Interestingly, the first universal screening operation was the UK 1907 Birth Notification Act, which was not about a medical test, but the empowerment of public health officers to make home calls and inspect every newborn infant (Dwork 1987, 137). It was an operation of state inspection and control.

[23]Britton 1994.

[24]Eggert 2006.

[25]Szreter 2007.

3 Newborn Screening Between Public Health and Clinical Care

Because NBS and immunization programmes are both preventive medical interventions administered universally and uniformly to individual people, it is tempting to compare NBS to immunization. Such comparison may behold the ethics of NBS through the prism of balancing personal autonomy against the power of the state as a promoter of the common good (or defender against public catastrophes). Indeed, NBS was born as a universal, mandatory and state-directed service in the same state whose compulsory vaccination law was upheld by the United States' Supreme Court in 1905.[26] However, reflection on the meaning and policies regarding non-compliance sheds light on the fundamental differences between the services. Non-compliance with immunization differs from non-compliance with NBS in at least three substantial ways. First, non-immunization might pose risk to others. Second, because non-immunized children benefit from the herd immunity of others, non-immunization involves a "free rider" set of problems. Third, whereas massive non-immunization brings forth a substantial risk to whole communities, abolition of screening carries a very low risk (in the range of 0.1% per non-screened baby). NBS brings much benefit to the few affected babies and their families, but it has no impact on public health and other public interests. Only the construction of "saving every life at risk", as a public responsibility does render NBS a public interest.

In US law, as well as in most countries, "imminent danger" may warrant state intervention in parental decision power over their child.[27] Whereas the "imminent danger" criterion might fit PKU and hypothyroidism, it is a much less convincing category regarding the vast majority of the diseases in the expanded panel. Since the 1970s, judges have tended to apply the "imminent danger" doctrine to procedures medically indicated by experts. However, unique to NBS is the question whether a test whose purpose is finding out whether the child is "at risk" warrants the coercive power of the state, especially when the risk is very low. Moreover, in most US states, doctors do not have to appeal to the justice system in order to overrule parental choice; rather, NBS is by default compulsory.

It is also noteworthy that the success of immunization programmes has led to the shrinkage of the panels recommended (e.g. abandonment of immunization to TB) and to growing tolerance of parental choice, while the success of NBS has been associated with expansion of the programme and intense efforts to reach each and every neonate.

Health care professionals and patient advocacy ushered in the era of expanded NBS. But it has been accompanied by loud critical voices coming mainly from the direction of bioethics and citizens' rights movements. Five interacting factors are responsible for the transition of NBS from a celebrated "life-saving" service to a

[26]Gostin 2005; Mariner 2005

[27]Horwitz 1979–1980, 272.

problematic social system. The first is technical. The more conditions screened, the more questions arise regarding lab standards, incidental findings, storage and the like. The second ensues from the shift from a core practice that is limited to the most obviously beneficial and urgent, to an expansive mind-set that strives to include every potentially relevant condition. It is a conceptual transition from parsimonious service to exploration of its limits and boundaries.

The third factor is related to the IT aspects of NBS services, who have become huge data banks and biobanks. Before the era of IT, the public beheld registration of personal data as promotive of public health and human rights; today, concerns about violation privacy, exploitation and other harms seem to prevail.[28]

The fourth factor behind the emergent criticism of expanded NBS is the maturation of bioethics and rights-oriented approach to bio-law. Whereas in the 1960s a few scientists and activists were able to introduce a universal and mandatory medical service, the 2000s are marked by heightened awareness of public participation and informed consent. Some critics propound the transfer of NBS from the conceptual and regulative schemes of public health to those of clinical care.[29] However, the division between public health and clinical care might not answer key ethical problems. Rather, the key factor behind the emergent controversies on NBS seems to be the chimerical nature of NBS as fitting and unfitting both public health and clinical care.

Typically, in clinical medicine, bodies of medical knowledge on diagnosis, prognosis and care are consolidating from the teachings of basic science, clinical research and cumulative experience. Although the interactions among science, medical services and culture are complex, personal choice of patients and lay people has marginal role in the canonization of medical textbooks, guidelines and similar standards of practice. Patients' involvement takes place in the clinical encounter, which is the arena where caregivers present patients with recommendations culled from standardized knowledge, and try to tailor with each patient personal decisions of health care (e.g. selection of antihypertensive medication or choice between surgery and observant policy). Ideally, decisions are made in a process of "shared decision-making", usually in the form of informed consent to every significant intervention, and implicit consent to care overall. Not only do acts of care require consent, but the therapeutic relationship also depends on a person's choice to be a patient of a particular clinician or health care service. Clinical research is an optional addition to standard care, but the latter is never conditioned on the former. Clinical research depends on special regulation (IRB) and specific informed consent.

From this schematic description, we may discern two stages of clinical decision-making. In the first, bodies of "professional" knowledge are created independently of individual patients, whose active participation becomes crucial and detailed only later, in the actual care of each person. This individual participation constitutes the

[28]Carmichael 2012.

[29]Ross 2011.

second stage of clinical decision-making. The transition from standardized knowledge to a personal health care plan passes through the doctor–patient relationship.

However, NBS does not fit this paradigm. First, in NBS, the patient neither suffers from any symptom nor seeks medical attention. Expanded NBS involves a large number of very rare conditions, of which the ordinary person has skimpy awareness at best. Even if a health care professional is keen on and capable of elaborating patient education, the parents' mindset may not be receptive to serious contemplation of testing their newborn baby for a wide set of improbable and unfamiliar diseases. Genuine autonomous decision-making is about making one's own values bear on the choice in hand; the average parent knows too little for making value-relevant choices on NBS. In order to cope with this difficulty, recent reports and policies recommend that patient education begin early, during prenatal care, for example, and that the authorities use the media to disseminate "educational material" to render NBS common knowledge.[30] But as long as this does not happen, and we do not have evidence of appropriate patient awareness at the time of screening, we have good reasons to doubt the relevance and validity of "consent", at least in the sense of personal informed consent to clinical procedures. It might be the case that parents are neither cheated nor coerced, and yet the notion of genuine informed consent to screening for dozens of metabolic diseases is implausible. Indeed, leading professional and public bodies have doubted the practicality of informed consent to NBS.[31]

Because medical knowledge of many of the conditions screened is still evolving, there is no "standard of care" in relation to NBS. Therefore, some critics argue that in many cases it is impossible to conceptually separate expanded screening programmes from clinical research.[32] Perhaps, only through universal screening of extremely rare conditions will it be possible to trace a minimal number of "affected" people so as to allow proper knowledge of the natural history and treatment options for the disease. Put in other words, we face a circularity in which the ultimate justification of NBS might depend on its universal penetration.

Lastly, and perhaps most significantly for the public, whereas public health enterprises focus on health risks that may affect people, newborn screening is about the people tested, about inborn "errors" or genetic variants. NBS policies affect directly the classification of people as "normal"/"pathological"/"at risk"/"in need specialist follow-up"/"carrier of an abnormal mutation". Public health often poses burdens on individuals. Yet, public health is blind to personal identity. NBS begins as an impersonal, universal operation; its ultimate goal is the identification of individuals with specific birth "defects" or "differences", most notably genetic differences. This is something that concerns citizen participation and informed consent more than ordinary public health and other public services. Indeed, the future of genomic screening bodes intricate system of risk stratification, such as

[30]AAP 2000, 409.
[31]AAP 2000, 409.
[32]Ross and Waggoner 2012.

simultaneous screening of hundreds of genetic markers associated with autism.[33] Screening genes blur further the distinction between NBS, whose goal is care for the child, and prenatal screening, whose goal is almost always the prevention of the child's appearance.

In sum, while in clinical care, a patient ideally gives an informed consent in a process of shared decision-making to a scientifically sound procedure in relation to which he or she harbours value-based preferences, in expanded NBS, people who have not chosen to become patients (or render their babies patients) have little power regarding a procedure. This procedure is part of a computerized service, carrying with it a long tail of disputed benefits and evidence-based validity, and in relation to which they seem to have no relevant personal values. Moreover, the service in question touches directly fundamental issues of privacy and civil status. This low level of personal participation places the onus of the ethical burden of NBS on the structure of the service and its legitimization. In the absence of effective and personal consent, the indirect process of democratic participation becomes crucial. The public may not question the probity of screening to PKU, but the inclusion of many other conditions, the incorporation of screening into large databases and tissue storage, the transition to genetic screening and the range of parental choice within the service are leading questions worthy of public participation.

4 Deep and Shallow Empowerment of Public Participation and Individual Choice

From its very beginning, the expansion process of NBS has been accompanied by significant deliberation and public participation.[34] These may assume different shapes and consequently different outcomes. For example, Bernhard Wieser compared the regulation of NBS in the UK and Austria.[35] In the UK, screening is regional, overseen by a dedicated National Screening Committee that communicates scientific information to the public. In Austria, the programme is centralized, overseen by a medical board advising the government on a variety of biomedical and environmental issues, communicating to the public legal and political information. Drawing on Sheila Jasanoff's theory of social epistemologies in collective decision-making in democracy, Wieser concludes that each society has a distinct approach to the relationship between information and legitimization. In the UK, the authorities' appeal to public reason hinges on open communication of scientific and technical knowledge; in Austria, the authorities claim legitimacy for their NBS programme through the exposure of its legal, political and administrative foundations, structure and oversight.

[33] Tzur et al. 2016.
[34] Hiller 1997.
[35] Wieser 2010.

However, it is not clear whether any relationship exists between the cultural and social modes of planning and legitimizing on the one hand and the programmes' ultimate structure, on the other hand. Moreover, the transition from descriptive analysis of processes of legitimization to normative conclusions is still undecided. One may endorse each method of democratic public participation as equally legitimate, but an Austrian might wish to know whether the Britons (with their much narrower panel) have a better programme or vice versa. He might wish to know whether the notion of an overall "better" programme is meaningful at all. In the USA, lack of uniformity of NBS programmes across all states has been considered unethical and unjust.[36] Possibly, no other medical service is mandatory in some democratic jurisdictions and depends on full informed consent in others. The cohabitation of both extremes seems to outstretch ordinary diversity in public reason. How is it possible to know whether different outcomes of democratic deliberative processes are equally ethical, and whether the coexistence of different programmes reflects pluralism or mismanagement?

Recently, an attempt towards unification of NBS services has been taking place in Southeast Asia, Oceania and the EU as well, with much emphasis on the value of standardization of laboratory and clinical practice.[37] Whereas uniformity of lab standards is an obvious scientific and clinical goal, the ethical and legal aspects of the programme seem to call for balancing pluralism with universal norms, such as fairness, protection from harm and respect for persons.

Perhaps, some jurisdictions care only for a "minimal liberal accountability", which is a majority-based consensus on a problem in hand, not seeking underlying justification and harmonization with other, even related, regulative issues. Other jurisdictions may prefer a "comprehensive deliberative account", which requires deliberation and agreement at the level of values, reasons and legal coherence. For example, so long as regulation is the product of democratic governance, the "liberal account" might tolerate absence of informed consent in NBS despite the centrality of informed consent in clinical care and medical research. But the "comprehensive deliberative account" would insist on coherence, demanding either the application of the same standards of informed consent or justifications for the difference. The coexistence of ethically inconsistent public choices constitutes the "discursive dilemma". Societies that value pluralism and free individual choice might be more tolerant of such inconsistencies than societies that seek shared moral foundations.[38] However, the growing role of civil and human rights in bio-law pushes policymaking in the direction of the "comprehensive approach". This is so at least since civil rights groups and individuals have appealed to courts arguing that NBS programmes are unconstitutional and incompatible with human rights.[39] Rulings at

[36]See US Department of Health and Human Service website: http://mchb.hrsa.gov/programs/newbornscreening/screeningreport.html

[37]Human Genetics Society of Australia 2011; Padilla 2012; Loeber 2012; Burgard 2012.

[38]List 2006.

[39]Couzin-Frankel 2009; Laurie 2002.

the level of constitutional and human rights are clearly a matter of "comprehensive deliberation" at the most fundamental levels of value judgements in society.

In addition to the descriptive problem of social epistemologies and the normative challenge of the discursive dilemma, another pitfall of legitimization is lack of proper awareness. The gap in awareness might be found at the level of public participation, individual choice or both. We refer to it as the "saliency problem".[40] Even when the public shares the same epistemic and normative paradigms, a shift in saliency may alter the support and even tolerance of a practice. The 2000 Enschede firework disaster is a case in point. When the private bio-banking company suggested the use of NBS samples for the identification of the remains of a child victim, a public outcry ensued. Although it was not possible to maintain that the bio-banking policy was "illegitimate", it was not even secret, people were not sufficiently aware of the storage of their babies' blood samples and its potential uses. Only following this episode, the government set detailed guidelines regarding consent to storage of NBS samples. The fire incident rendered storage of NBS sample a salient public preoccupation.

In a seminal paper "A ladder of citizen participation", Sherry Arnstein observes that "participation without redistribution of power is an empty and frustrating process".[41] In a similar vein, Lain Ferguson distinguishes between "deep" and "shallow" personalization of public services.[42] While deep personalization empowers individuals to contribute to the design and governance of public services as well as to make substantial personal choices in the use of these services, shallow personalization is typically unidirectional (e.g. increased access to information about the service without opening to effective feedback) and peripheral to the essence of the service (e.g. friendly interfaces, long office hours). Shallow participation raises worries inspired by Foucault's notion of bio-power/bio-politics, according to which, by means of trivial satisfactions and distracting practices, hegemonic powers lure the public to accept the legitimacy of institutions and even to perceive them as natural and inevitable realities.[43] Consequently, high rates of compliance do not necessarily indicate genuine endorsement. This is so because participation might be shallow, people might be superficially alert to the moral problems involved, and they might see neither alternatives to conformity nor incentives to accept the price of dissent.

It follows that ethical legitimization depends on policymakers' capacity to frame questions and posit them before mindful citizens. Put in other words, participatory deliberation is not just a matter of gathering experts and stakeholders, presenting them with a regulative question and waiting for the outcome. Not every form of

[40]The concept of saliency originates in social psychology. Whereas people are aware of numerous issues, only a few are present in the forefront of people's consciousness. Each kind of choice requires a different level of saliency. Consent to a blood test is different from consent to surgery.

[41]Arnstein 1969.

[42]Ferguson 2007.

[43]Rabinow 2006.

public participation and individual frames of choice effectively bite into the ethical issues at hand. We are talking here about genetic counselling at the level of public health. Is this an oxymoron, or a meaningful goal?

5 Solidarity, Liberal Democracy and Genetic Counselling in Public Health Operations

Solidarity is a relatively young concept, a baby of the Enlightenment, whose earlier meanings connoted the abstract bonds of social cohesion and shared decision-making. The earliest use of the word is traceble to the Dictionnaire de l'Academie Frangaise (1694) in which "soidaire" was a legal term designating an indivisible collective debt. As explicated by nineteenth-century French thinkers Pierre Leroux (1797–1871) and Léon Bourgeois (1851–1925), in the absence of either a monarch or a universal religious commitment, citizens of a republic share solidarity regarding state policies and actions.

In applied ethics, solidarity is a perception of moral commitment towards some people based on a morally relevant shared trait or interest (or a set thereof). An immigrant may experience solidarity with other immigrants, willing to assist them with advice and other resources. This immigrant might be a woman sharing solidarity with other women—native and immigrant alike—willing to contribute to an organization dedicated to gender equality. Having a sister who has died from refractory anaemia, this immigrant woman may also be willing to donate blood to cancer patients. Co-workers may wish the immigrant woman to join the local labour union in solidarity of other labourers, but for some reason, she is reluctant. This imaginary and arbitrary example illustrates how much solidarities and claims to solidarity-related behaviours might combine and clash with each other. From the perspective of the individual, solidarity is a matter of personal choice in relation to incomplete duties (=the recipient is known, such as a needy child, but there is nobody designated as accountable for action). Especially in global contexts, solidarity is about a sense of commitment even in the absence of shared legal or political structure of decision-making.

It is often alleged that with modernism and globalization, solidarity has been weakening.[44] However, it might also be possible that an ever-growing awareness of diverse solidarities is responsible for this perception. As people recognize more solidarities, no one line of solidarity dominates their moral sensibilities. The contemporary notion of solidarity has never been fully differentiated from "solidarism", which is about a kind of "social debt" of the privileged to the unfortunate in society.[45] "Solidarism" is about a sort of remedying social abuses in a framework of

[44]Have 2016, 216–220. Other claim that solidarity with human beings as such is a fiction. Rorty 1989.

[45]Hayward 1959.

collective responsibility. Even though some scholars believe that even though the social phenomenon of solidarity always encompass a specific group of beneficiaries who either share something with us (e.g. class) or to whom we owe something (e.g. "solidarism", reciprocity), solidarity with human vulnerability as such is an ideal to be cultivated.[46] It might be argued, that whereas all specific solidarities contain a measure of external social pressure, solidarity with human vulnerabilities is a closer to a pure moral sentiment, and it fits integration within a moral identity.[47]

Talking about solidarity as a value in public policies, we need to explicate its nature, its standing relative to other solidarities and relative to other values such as respect for personal autonomy. In health care ethics, solidarity is focused on health-related human vulnerabilities.

Solidarity raises three key questions: the first is the identification of basic values that are related to health care and the human condition as such; the second is the relationship between solidarity and respect for personal autonomy. Solidarity promotes uniform acts (e.g. vaccination, blood donations, almsgiving) in the benefit of the needy; respect for autonomy promotes individual choice, even in disregard from the needy. Third, sometimes public policy needs to balance one kind of solidarity against the other. For example, solidarity with expectant mothers and solidarity with babies may push in opposing directions. This is one reason why solidarity is a philosophically raw concept that needs explication within a broader normative matrix. In bioethics, solidarity is a key value, which raises universal moral claims (as opposed to particular solidarities such as gender, class or ethnic-based solidarities), and which tries to avoid the challenge of "collective responsibility". The latter is guilt driven, while solidarity is a primary moral motivation for beneficial action. Moreover, many count solidarity among the core set of bioethical principles (along with justice, respect for dignity, respect for autonomy, beneficence, non-maleficence and vulnerability).

In bioethics, solidarity aims at the more basic shared human vulnerabilities, which are life, health and respect for dignity.[48] This is a crucial point in relation to childbirth policymaking. Vulnerability-based solidarity gives saliency to NBS, whereas feminine solidarity and "women's experience" do not. It brings to the fore parental responsibility as central to neonatal care.

Solidarity creates a justification for spending public moneys, burdening everybody with fungible costs. Even legal structures that tightly restrain the power of the government to spend money (e.g. USA) have accepted the notion that the public should assist victims of disasters and vulnerable people with life-saving needs. Some policies are explicable in terms of the mutual benefit of insurance and similar schemes of joint assistance. However, we invoke the value of solidarity even in the absence of risk-benefit calculations.

Solidarity creates defaults. Solidarity-based policies do not trump personal choice over body and person; the burden they place on individuals is minor and

[46]Rorty 1989, 192.

[47]See Luke 1973, 500-504.

[48]Prainsack 2011.

often not beyond the fungible. It follows that solidarity may justify opting-out, but not compulsory, schemes of organ donations from the brain dead. Solidarity may also justify laws constraining the sales of unhealthy food. Thus, society promotes solidarity-oriented choices, creating incentives and fostering publicity campaigns. Whereas it is possible to behold the encouragement of "healthy" lifestyle as "nudges" in the benefit of the common good, other policies are clearly in the benefit of the few.[49] Laws obliging wheelchair accessibility are one example. Liberal society have found acceptable the burden of making businesses accessible to wheelchair, but not the burden of medical research without informed consent. This is because we do not behold research without consent as mere burden, but offensive to human dignity.

6 A Solidarity-Based Framework for Newborn Screening

We may behold the NBS operation as comprised of two phases. The first phase is a public health enterprise in which a blood sample is drawn from every newborn baby. The second part occurs only in relation to positively screened neonates. By testing positive, they become patients in need of further follow-up. The suspected disease is their personal issue. The second part of screening is clinical in nature and it involves consultation to the parents about a particular health risk to their child.

Because of the very low level of personal risk, it makes little sense to argue that the first phase is done in the benefit of the tested baby. We do know that, if every newborn participates in screening, a few will be saved somewhere. Whereas the chances that a particular child benefits from screening directly are very low, we know that universal compliance with newborn screening will certainly save a few lives. Compliance with NBS is a personal contribution to a rescue operation. Hence, the analogy is the deployment of rescue operations. Everybody's tax money goes to rescue services that will benefit very few people. Rescue efforts are not limited to common conditions such as myocardial infarctions and road accidents. Society invests much effort in order to search for survivors of crashed airplanes and lone travellers lost in the desert the occasionally heavy costs notwithstanding.

Because personal risk is extremely low, personal counselling about NBS prior to testing seems impractical. Because people pay attention to salient risks relevant to their own lives, it is impossible to inform seriously on so many remote risks and to expect deep engagement on behalf of the public. However, public participation is the key to the structuring of the service of newborn screening as an operation covering a collection of related risks, similar to emergency services that cover a broad array of medical emergencies, some of which are pretty rare.

Because of their rarity and complexity, awareness and motivation come from either dedicated scientists-clinicians or patients' groups. Interaction between both

[49]Indeed, one does not find "solidarity" in the influential book on "nudges" (Thaler 2009).

often stimulates novel ideas, such as the Guthrie method of testing for PKU. Policymaking begins at this end. However, policymaking must diffuse into society at large, and especially potentially significant stakeholders.

On the basis of scientific knowledge and technological capacity, experts propose a NBS panel. At this phase, public participation is indirect, usually by means of representative bodies (e.g. parliamentary committees) that exercise power over speeding up and incorporating novel technologies in terms of licensure and funding.

Mixed panels (professionals and lay participants) will then translate technological competence and scientific knowledge into medical services in ways that address public's concerns and empower personal choice on ethically relevant aspects of the service. In our view, scholars in the medical humanities should participate in these panels in order to bring before the scientists and the public the historical, social, psychological and philosophical aspects of the issue at hand. This may enable the scientists and policymakers develop diverse tracts for the service (e.g. with and without tissue storage) from which the public may choose the policy that best meets its values. Precisely because the boundaries between the "normal" and the "pathological", as well as similar value-laden concepts, are culturally constructed, analytic-deliberative processes may help scientists working on applied tests and programmes to delineate categories, select conditions to focus on and set laboratory "cut-offs" so as to meet social perceptions of risk and value.[50] For example, such panels may opine that certain conditions must be detected even at the price of high rate of false positives, while other conditions are too rare or less devastating as to justify the setting of lab standards at very high sensitivity threshold. Another example might be borderline values, which the public might refuse to classify as pathologies subjected to testing, and set high the threshold of diagnosis. A third example is whether and how to represent the possibility to screen for late-onset and poorly understood conditions (e.g. Krabbe). Even if these panels decide that public policy remain silent on these conditions, researchers might still obtain ad hoc permissions to offer pilot screening programmes, whose outcome might impact future revisions of NBS policies.

The service and its structurally independent governance will include instruments for public education, as well as evaluation and feedback from its users (e.g. website, publicity officers).[51] Public education without patient empowerment is ethically shallow. Public education can serve as template for citizen empowerment at the public domain and patient empowerment in the sphere of private health care choices. For this empowerment to be effective, it is crucial that dissenting personal choices not entail service abandonment. Parents have the right to choose screening only for the core conditions (a few most common and obviously needed, such as PKU and hypothyroidism), and without having to resort to genetic testing, if a simpler alternative is technically feasible.

[50]Douglas 2009, Chap. 8 following National Research Council's report *Understanding Risk* and the Presidential/Congressional Commission on Risk Assessment and Risk Management Report.
[51]AAP 2000.

7 Conclusion

In this chapter, we have surveyed the history of NBS and key ethical and regulative challenges of expanded newborn screening, and the prospect of genetic-based NBS. In light of the values of human dignity and solidarity, we have offered a political justification and rationale for NBS services. This long way, from history, through value inquiry into specific policy issues (e.g. the structuring of informed consent) serves as a model for deep public participation and value-oriented legitimization of health care services in democracy (as well as other expert-guided public operations).

In our model, we have argued that solidarity may justify the institution of a publicly funded universal service and the creation of defaults in public health policies. The public need be informed about the service and the opting-out options so as to allow those who are interested to learn more and have personal counselling before making a personalized choice. This structure requires clear separation between solidarity-based services (those clearly connected to substantial health benefits to the patient) and related parts of the service such as controversial, experimental services and research. Participation in one part of the service must not be conditioned on participation in any other part.

Revisibility (=democratic procedures for revising decisions) by means of public representation is indispensable to democratic governance.[52] However, democratic governance also depends on set of shared and relevant values. The prospect of a transition to genetic NBS is not a technological challenge anymore, but one of public participation, especially in the choice of the ethical paradigms for approaching NBS and genomics. Even though solidarity may compel strong incentives for participation, with the possible exception of emergency and risk to others, every public health programme affecting directly the human body and person needs to have a realistic option of opting out. Services that involve issues known as public concern, such as genomics, require legitimization with appropriate saliency of public participation and effective instruments for personal choices.

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[52]Tilley 2003; Daniels 2000.

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Feminist Criticism of Genetic Counselling in the Second Half of the Twentieth Century

Shachar Zuckerman

Abstract Genetic counselling, a term coined by Sheldon Reed in 1947, was originally defined as a kind of genetic social work. Both parties to counselling sessions are usually women. Yet the socially oriented definition of genetic counselling and its feminine nature did not prevent the discipline from attracting significant feminist criticism. Feminist critics of genetic counselling regard it, first, as being complementary to reproductive technology. Women's reproductive rights groups argue that genetic counselling is defined as medical therapy aimed at alleviating human suffering whereas in fact it becomes a means of social control, especially over women. Another group of critics is disability rights advocates. They argue, in relation to the social model of disability, that genetic counsellors play an important role in the relationship between the disabled community and nondisabled society because their own moral perspectives and biases regarding disability affect decisions regarding prenatal diagnosis and pregnancy terminations. Thus, they contribute to the construction of the disabled community as people whose lives are not worth living. Thirdly, contemporary feminists focus on the genetic counselling session itself, questioning the assumptions of individual choice and shared decision making. Much of this criticism ignores the main conceptual framework of genetic counselling. The core values of genetic counselling have been designed to help women counselees make a personal decision in keeping with their moral principles and beliefs and thus truly empower them. Accordingly, this chapter concludes that genetic counselling should be encouraged rather than be criticised by feminists.

Keywords Genetic counselling • Feminism • Women • Decision making

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1 Introduction

"Each act of genetic counselling is a political act in which one woman directly influences the reproductive decision of another woman", declared Annette Patterson and Martha Satz.[1] The aim of this chapter is to critically reflect upon some of the feminist criticisms of genetic counselling over the second half of the twentieth century. By examining three major critics I shed light on specific characteristics of genetic counselling and argue for genetic counselling as an essentially *feminist* act.

Genetic conditions have captivated people since ancient times. Twentieth-century advances in biology have enabled diagnosing these conditions, and the emergence of genetic counselling has enabled laypersons to understand and benefit from these advances in making critical decisions.

The genetic counselling discipline was criticised from its very beginning, especially because of its early association with the eugenic movement. The term "eugenic", originally suggested in 1883 by Francis Galton, literally means "well-born". However, the definition of eugenics is much broader and encompasses the notion of process as well as intention: what people may be willing to do to ensure that the offspring is wellborn. As developed principally in England, the early concepts of eugenics were derived from the belief that the upper social classes were in danger of being "diluted" by the growing numbers of "inferior" lower social classes and races due to higher birth rates.[2] Galton's original concept was of positive eugenics: encouraging childbirth among members of the upper classes who possessed desirable characteristics.[3] However, the actual implementation of eugenic principles very quickly began to run along negative eugenic lines. Rather than permit the Darwinian survival of the fittest to control the gene pool, the objective was to ensure the non-survival of those considered unfit. The principal means of implementing negative eugenics was by discouraging or preventing reproduction of the "unfit" by preventing (interracial) marriage, institutionalisation and sterilisation. Added to this were quotas on the immigration of the supposedly unfit and their general stigmatisation and discrimination. Moreover, abortions were employed to prevent them from giving birth.[4]

Although all these measures were theoretically voluntary, they rapidly became compulsory in many countries and laws providing for (formally) voluntary and compulsory sterilisation were passed in more than half of the American states,[5] as well as in several Northern European countries.[6]

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[1] Patterson 2002, 120.

[2] Iredale 2000.

[3] Li 2000.

[4] Iredale, 2000; Geiger and Mayer 2017.

[5] Iredale 2000.

[6] Butler 1997; Hemminki 1997; Bjorkman 2010.

In Nazi Germany, eugenics took on a much more extreme form in the euthanasia programme.[7] This was followed by the Holocaust, with the wholesale extermination of millions of Jews, "gypsies", homosexuals, mentally and intellectually challenged people and others deemed unworthy of living and certainly of reproducing. This was to be the ultimate genetic cleansing. No longer was breeding of the undesirable to be controlled—rather, the breeders who were thought to carry undesirable genes were to be eliminated altogether.[8]

After World War II, attempts to distance genetic counselling from eugenics were made. Nevertheless, many geneticists practicing genetic counselling in the 1940s and 1950s still felt that eugenic goals were compatible with those of genetic counselling, while at the same time criticising eugenic programs based on racism and coercion.[9] As described in the following sections, the emergence of genetic counselling as a holistic process and establishing the notion of nondirectiveness as a fundamental value were thus a reaction to eugenic methods as practiced in Europe and USA.

2 The Emergence of the Genetic Counselling Profession

Genetic counselling—a term coined in 1947 by Sheldon Reed (1910–2003), a non-physician with a PhD in genetics—was originally defined not as a medical encounter but "as a kind of genetic social work aimed primarily to provide people with an understanding of their family's genetics problems".[10] This definition reacted to the history of genetic and social evolutionary ideas that had informed eugenic theories during the first half of the twentieth century as described above. In the late 1960s and early 1970s, a major shift in human genetics occurred, with the identification of some chromosomal and metabolic conditions (e.g. Down syndrome and phenylketonuria). Improved diagnostic capability together with the development of new procedures and techniques in reproductive medicine and medical genetics expanded genetic counselling beyond the tools of pedigree charting and the calculation of Mendelian probabilities to include determinations about carrier status and diagnosing conditions. Foremost among these was amniocentesis, which had been utilised by some obstetricians since the 1940s to relieve polyhydramnios patients and, on occasion, perform biochemical testing of maternal–foetal Rh compatibility. By the 1950s advances in the culturing of foetal cells allowed amniocentesis to be combined with cytogenetic techniques such as karyotyping for the purpose of chromosomal analysis.[11] Twenty years later, once

[7] Rotzoll 2006.
[8] Epstein 2003.
[9] Resta 1997.
[10] Reed 1974, 332.
[11] Cowan 2008.

amniocentesis developed as a clinical service, a new goal of the profession was defined: conveying the risks and benefits of the test or translating scientific possibilities into personal calculations. Those gatekeepers between science and social work, between epidemiology and empathy, became the genetic counsellors—members of a new allied health profession.[12]

In 1969, the first college programme in genetic counselling was established in Sarah Lawrence College in Bronxville, New York. Beyond the perspicacity and persistence of the programme's founder, Melissa Richter (1920–1974), various social, scientific and educational factors converged to this path-breaking programme possible.[13] The first was the emergence of second-wave feminism, leading to the rise of the feminist health and civil and reproductive rights movements and their struggle for acceptance of and access to birth control.[14] This dovetailed with the decriminalisation of abortions. In 1970, New York became the first state in the USA to decriminalise abortions.[15] Thus, Sarah Lawrence's first cohorts of genetic counselling students started their clinical training as abortion became available for patients who decided to terminate their pregnancy because of a genetic disease diagnosed in the foetus. And indeed, the medical advances discussed above also played into these developments. Broader trends during this period that also facilitated the professionalisation of genetic counselling were the growing importance of bioethical principles, such as patient autonomy[16] and changing attitudes towards the physician–patient relationship.[17] Finally, Sarah Lawrence's unique educational mission and geographical location were an important factor. Its Centre for Continuing Education aimed to encourage women who had abandoned college to return to complete their degrees, and one of its missions was to offer equal opportunities to women and members of racial minorities. The college's proximity to New York City with its hospitals and genetics clinics was also important to the programme in that it provided internship opportunities. Richter's recognition of these factors and their implications for patients prompted her to design the genetic counselling programme.[18] Richter's design was fundamentally important since it would prepare the groundwork for genetic counselling for years to come. Some professionals involved in the programme's design had originally believed that only PhDs or MDs could do genetic counselling and that genetic counsellors should serve as mere assistants or at most associates charged with the low-priority duties for which busy physicians had no time or inclination. Richter and her colleagues, however, were convinced that an MA programme could combine intensive science studies with training in complex counselling skills in

[12]Rapp 1999.
[13]Stern 2009.
[14]Nichols 2000; Munch 2006.
[15]Cook 1978.
[16]Dresser 1996.
[17]Farrel Smith 1996.
[18]Stern 2009.

order to enable counsellors to perform professional medical social work and at the same time distinguish themselves as independent healthcare providers.[19] Regarding the controversial eugenic legacy, the new programme had initially accepted eugenic rationales, but eventually Richter came to support a brand of genetic counselling that emphasised private decision-making and individual reproductive choice not dictated by the state. Her main goals were to provide the students with expertise in communicating genetic risk with sensitivity and scientific accuracy and in understanding the function public agencies should play in genetic testing and counselling. Following Richter's approach, Joan Marks (b. 1929), who replaced her as programme director, enhanced its psychosocial component and added a four-semester sequence titled "Issues in Clinical Genetics", which taught patients' rights, women's health, family dynamics and medical sociology.[20]

The dark shadow of Nazi ideology and other eugenic practices and the dynamic changes of the 1960s and 1970s, as illustrated by other contributors of this volume, for examples the chapter regarding the case of Thalassemia in the Mediterranean parts of Europe,[21] helped frame approaches to the goals and limits of the new profession. Genome scientist Arno Motulsky (b. 1923)[22] encapsulated the ethos of genetic counsellors as having a duty to "put the interests of the patient and his family before those of society and the state. The genetic counsellor pursues medical and not eugenic objectives".[23] In order to accomplish this complex mission, the notion of nondirectiveness emerged as one of the core values of genetic counselling.

3 Nondirectiveness

Nondirectiveness (ND) is one of the hallmarks of genetic counselling. It plays a critical role in reminding us of the past abuses of genetics in the first half of the twentieth century.[24]

It is unclear who first introduced the term into the genetic counselling literature. Reed may have been instrumental in this regard, as he had borrowed the concepts of directiveness and ND from psychotherapy and associated them, respectively, with the giving and withholding of advice by the early 1960s.[25] The term may also have been coined by J. A. Fraser Roberts (1899–1987), who mentioned in one of the first textbooks on medical genetics, published on 1959, that "The principles of genetic

[19] Stern 2009.
[20] Stern 2009.
[21] See Barmpouti 2017.
[22] Motulsky 1973.
[23] Motulsky 1973, 318.
[24] Michie 1997.
[25] Kessler 1997.

counselling, as laid down, are that advice should be given in terms of risk and that it should be nondirective".[26] Later, in the 1970s, there was a gathering momentum among geneticists towards a nondirective approach and by the mid-1980s, surveys of hundreds of medical geneticists around the world showed an overwhelming endorsement of ND in genetic counselling.[27]

In the influential text "Psychological Aspects of Genetic Counselling. XI. Nondirectiveness Revisited", Seymour Kessler[28] defines nondirective counselling as the process designed to help the patient make an autonomous decision in keeping with his own principles. Kessler emphasises the ability of genetic counsellors to promote autonomous decision-making using ND, whose expanded definition is as follows:

> ND is more than withholding advice. It is a way of interacting and working with clients that aims to raise their self-esteem and leave them with greater control over their lives and decisions... Thus, ND is an active strategy requiring quality counselling skills. ND does not happen by default or by not directing the client towards a particular decision or course of action... ND is a way of thinking about the professional–client relationship in which at each step of the way the professional attempts to evoke the client's competence and ability for self-direction.[29]

According to Kessler, ND strategies require the professional to (1) pay attention to patients' strengths, accomplishments and competencies; (2) verbally acknowledge these abilities throughout the session so that clients feel that the professional has confidence in their ability to make their own decisions; (3) remember that most clients are already experienced decision-makers: draw on their intelligence and life experiences; (4) encourage clients to talk more during the counselling session since it gives them a sense that they have greater control over the situation and (5) reward and reinforce any effort towards autonomy, self-direction and individuality.[30]

Kessler specifies the reasons for the assimilation of ND in genetic counselling including the anti-eugenics conviction of many geneticists, the changing nature of medical practice, the growing consumerism movement and the increased awareness among geneticists that they often dealt with life decisions about which they had no greater expertise than anyone else. Last but not least was the finding that women, more than men, were likely to be nondirective.[31] Thus, the feminisation of genetic counselling led to wide acceptance of ND.

[26] Michie 1997, 40.
[27] Wertz 1988.
[28] Kessler 1997.
[29] Kessler 1997: 169.
[30] Kessler 1997.
[31] Kessler 1997.

4 Feminisation of Genetic Counselling

Both parties to genetic counselling sessions are usually women. The majority of sessions in prenatal counselling are associated with pregnancy, so counselees are mostly women, sometimes accompanied by their spouse. Moreover, the primary caregiver of children is still usually the mother. In the field of oncogenetics, much of the debate regarding surveillance and prevention measures deal with genetic factors causing breast and ovarian cancer, so here too most counselees are women.

Women who come to genetic counselling usually meet a female counsellor: As was true in previous administrations of the Professional Status Survey of National Society of Genetic Counsellors, in the 2016 Professional Status Survey 96% of the respondents were female and 4% were male.[32] Percentages of women counsellors in European and Middle-East countries range from 92.5 to 100%.[33]

In the early beginning of the profession, it was dominated by male physicians. However, as described above, the first educational programme of genetic counselling attracted mainly women from the well-educated white suburban community near the Sarah Lawrence College, often wives of upper class men.[34] Sarah Lawrence's Centre for Continuing Education, the location in which this programme was located, aimed to encourage women, who had abandoned college education for reasons of marriage, motherhood, or work, to return to complete their bachelor's degree or to obtain a professional degree. Moreover, Richter believed that the Centre for Continuing Education was the ideal home for a genetic counselling program and would appeal to its core constituency of mothers in their 30s. She believed genetic counselling was a profession ideally suited for women because they were generally more concerned with health and the preservation of life and had the "female qualities" deemed necessary, such as empathetic listening. Richter's objective was to bridge the gap between the rapidly expanding genetic knowledge on one hand and lay patients on the other by training smart and caring women who could communicate effectively with patients. The feminist and civil rights movements' appeal to a widening female workforce also contributed to creating this new opportunity for women. The women applying for the programme sought to balance their intelligence and independence with the demands of family life and part-time employment.[35]

Of particular relevance to the present discussion is the fact that although the first genetic counselling programme emerged more than four decades ago, the racial, ethnic and class homogeneity of the early cohorts still characterises genetic counsellors, who remain overwhelmingly white middle-class women. According to the National Society of Genetic Counsellors 2016 Professional Status Survey, the majority of genetic counsellors (91%) reported their racial classification as white,

[32]NSGC 2016.

[33]Skirton 2013; Pestoff 2016; Israeli Association of Genetic Counselors 2016.

[34]Rapp 1999.

[35]Rapp 1999; Stern 2009.

4% as Asian, 2% as Asian Indian and very few (<2%) reported their ethnicity to be Hispanic, Latino(a), Black or African American. Two percent of respondents identified themselves as part of a disability community.[36] Perhaps inevitably, the feminisation of genetic counselling and its social aspect resulted in the profession's under-evaluation in the traditionally male-dominated medical profession in terms of both compensation and prestige.[37] They also did little to deflect the growing feminist criticism against it, to which we now turn.

5 Feminist Approaches to Genetic Counselling

Two social movement constituencies stand out among the feminist critics of genetic counselling: feminists concerned with the increasing medical control of women's childbirth experiences and disability rights activists concerned about eugenic judgments and practices affecting the stigma of physical and mental differences. Contemporary feminists refer more to the content of the counselling session itself, which they find unsuitable to the highly diverse clients population.

5.1 Women's Reproductive Rights Groups

The first strand of feminist criticism of genetic counselling, starting in the mid-1970s, condemns medical–scientific practices in general and regards genetic counselling as complementary to reproductive technology. In one of the chapters of *Made to Order: the Myth of Reproductive and Genetic Progress*, American biologist who has been working on a feminist critique of science, Paula Bradish (b. 1953)[38] argues that while new reproductive technologies are presented to the public as therapy for the infertile, they actually offer a powerful means of social control. Similarly to genetic engineering, genetic counselling is supposed to alleviate human suffering whereas in fact both are new ways of influencing and controlling society in general and women in particular. Moreover, while the new technologies of conception determine the *quantity* of children a woman will have, genetic counselling intervenes by determining the children's *quality*.

According to this view, genetic counselling is a new form of eugenics. Indeed, while its proponents admit that genetic counselling uses more subtle and scientific forms, they claim that the upshot is the same: it supports discrimination and categorisation that affect women's decision making. Furthermore, within the scientific and medical community, the main argument in favour of genetic counselling

[36]NSGC 2016.
[37]Rapp 1999, Stern 2009.
[38]Bradish 1987.

and diagnosis—behind the smoke screen of alleviating human suffering—is a simple cost-benefit analysis. Geneticists are accused of attracting more clients by employing different arguments and strategies in counselling sessions than they do in public. Shockingly high percentages of infants born with "genetic" defects are presented to the baffled client, without an adequate definition of what is included in this category. These professionals use the term "congenital defects" and its correct numbers without mentioning the inclusion of illnesses and handicaps caused by environmental factors (e.g. pollution and medications taken during pregnancy), medical malpractice or other unknown factors. Finally, the disingenuous intentions of the genetic community is proven, according to these critics, by the support of some geneticists of sterilising people carrying genetic risks, although this practice has been illegalized in their countries.[39]

5.2 Disability Rights Advocates

A second group of critics are feminists with a disability rights perspective. They argue that socially constructed attitudes of stigma and prejudice, rather than absolute biological capacities, lie behind the discrimination of disabled children and adults.

British sociologist and disability rights campaigner Tom Shakespeare (b. 1966)[40] explains that the social model of disability relies on two key elements: the distinction between disability (social exclusion) and impairment (physical limitation) and the claim that disabled people are an oppressed group. Furthermore, he distinguishes between impairment and disability, the former being individual and private and the latter structural and public. His claim is that while medical professionals seek to remedy impairment, the real priority should be to accept it and remove disability. Shakespeare suggests an analogy of disability with feminism and the distinction between biological sex and social gender. Like gender, disability is a culturally and historically specific phenomenon, not a universal and unchanging essence. Social model thinking mandates barrier removal, antidiscrimination, legislation, independent living and other responses to social oppression. Disabled people are distinguished from nondisabled people and are an oppressed group. Finally, it is often nondisabled people and organisations—such as professionals and charities—that are the causes or contributors to that oppression.

Following this view, critics from this discipline argue that genetic counsellors play a pivotal role in the relationships between the disabled community and society at large. The personal and moral views of genetic counsellors towards disability affect clinical decision making related to prenatal diagnosis and pregnancy termination and by extension affect societal values and public policies.[41]

[39]Bradish 1987.

[40]Shakespeare 2013.

[41]Patterson 2002.

Adrienne Asch (1946–2013),[42] a feminist and pioneer of disability bioethics, was a strong advocate for disability rights and social equality. In her view, genetic counsellors are ill-equipped by their own training and norms of practice to provide any insights into disability in today's society. This is a result of the fact most graduate programs in genetic counselling do not include courses in the social implications of life with disability for children and families; do not include contact between counsellor trainees and disabled children and adults outside clinical settings and do not expose counsellors to laws, disability rights organisations and peer support groups that constitute what is described as the disability rights and independent living movement.

These critics argue that genetic counsellors participate in the construction of disability by attempting to define and explain the nature of a particular condition, often without any experience of disability or interaction with a disabled person. In that, they take advantage of their professional authority, usually from a privileged and able-bodied standpoint. By providing information to prospective parents about genetic conditions, usually without any experience with disability themselves, they structure the counselee's view regarding that condition and consequently influence the future existence or oftentimes nonexistence of a person with such a condition. The information they provide becomes a frame of reference for prenatal decision making. The idea of counselling itself conveys a message since offering information about the foetus' defects and disability in a medical setting, with abortion offered as an alternative, places women on a course that may end in pregnancy termination due to suspected abnormality.[43]

According to Adrienne Asch and Gail Geller,[44] the educational materials used for training genetic counsellors is of concern since it convey traditional biases about disability and focuses simply on medical characteristics and probabilities and less, if at all, on the lives of people with disability. Mistakenly, the genetic counsellor links every difficulty of a disabled person to the condition's physiological characteristics and ignores the role played by society in framing, if not creating, the disability. This view is related to one of the prominent notions of feminism that challenges the definition of women's place in society based on their biology. This counselling approach has indirect but very real impact on able-bodied biases, as counselling regarding disabled foetus conveys the presupposition that our society and our families cannot accommodate people with a wide range of problems. Their concern is that aborting disabled foetuses will lead to intolerance of difference in a society.

These critics describe the actors in genetic counselling sessions as operating within a social hierarchy, in which the counsellors are the privileged and the disabled are the marginalised. In order to overcome these obstacles in genetic counselling, they propose incorporating the perspectives of those with disabilities

[42]Asch 1999.
[43]Patterson 2002.
[44]Asch 1996.

into the counselling process. This requires listening and relating to their histories, achievements, social relations and hopes, as well as critically examining the dominant institutional beliefs and practices that systematically disadvantage them, and reflecting on the counsellor's own biases.[45] Larger-scale change will take place when we, as a society, choose to overcome the traditional and oppressive mythology surrounding genetics: biology is destiny, difference is always bad and technology is the sole solution. The community should enable women and families to welcome all children and should aid adults with genetic and other disabilities to live as valued members in society.[46]

5.3 *Contemporary Feminist Analysis of Genetic Counselling Sessions*

Most recently, feminists have tended to focus their criticism on the character of the genetic counselling session. A leading representative of this group, American anthropologist Rayna Rapp[47] suggests that in light of the assumptions of the counselling protocol, and despite pretensions to the contrary as embodied in the ND concept, the nature of these sessions is actually directive and far from value neutral and "eugenics-free". According to the current protocol, Rapp argues, a counsellor should, as a matter of principle, support whatever decision regarding testing and pregnancy outcome a woman or a couple make. In practice, however, this is based on the twofold and unsubstantiated assumption that the woman counselled is free to make individual choices, unconstrained by kinship and community, and that her male partner is supportive regarding communication and shared decision making. Moreover, the sessions in the American context are not value neutral in that, the counsellors' thinly veiled views closely mirror dominant American values and deserve examination.[48]

To address these common shortcomings of the counselling session, feminist critics argue that in each session, we need a different, fuller context within which to situate the meaning of this particular pregnancy in light of community values, reproductive histories, and the trajectory of each particular woman and her partner. Such grounding would provide ample space for examining the contradictory social relations and limits each pregnant woman faces and the constrained agency she exercises in her reproductive choices. As Rapp vividly illustrates this point, "Each pregnant woman brings the light and shadow of her personal biography, family history and community resources into the consulting room", and this filters the new information she hears.[49]

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[45] Patterson 2002.
[46] Asch 1996.
[47] Rapp 1999.
[48] Rapp 1999.
[49] Rapp 1999: 77.

Rapp also questions the ND ethos and particularly its value neutrality aspect. In her view, it is hard to argue for the neutrality of a technology explicitly developed to identify and hence eliminate foetuses with certain chromosomes and genes.[50] The biomedical and public health interests behind the development and routinisation of the technology itself evaluate such foetuses as expendable. Parents of potentially disabled foetuses have a right to know the chromosomal status of their foetus—but also the right *not to know it*—and the information is theirs to provide the basis for any decision they may wish to make, whether that means preparing for the birth of a child with special needs or ending the pregnancy. In practice, argues Rapp, the very existence and routinisation of the technology imply directiveness and lack of neutrality.[51]

The practical problems of value neutrality are even more vexing when genetic counsellors work in a cross-cultural community. Some individuals and populations may not want what they perceive to be the burden of individual choice. To them, directing the patient towards the appropriate choice is the counsellor's job. Rapp suggests that in order to adjust the counselling to women from different backgrounds, counsellors should be able to understand community-based differences, respect different experiences among women and investigate patriarchal aspects of control over women's reproductive decision making.[52]

Other critics propose a more ambivalent approach to genetic counselling. In "Prenatal tests: Blessing and burdens", British women's liberation activist Janet Hadley[53] suggests that contemporary reproductive genetics has both blessing and burdens, in that it is simultaneously liberating, discriminating and constraining. Israeli social scientist Yael Hashilony Dolev[54] adds that women's options are always constrained both by the value-laden technology itself and by the prevalent ways of using that technology in their society. Studying the cultural premises behind pregnancy management is vital for any critical discussion of women's losses and gains as a result of undergoing genetic tests. The discussion regarding prenatal diagnosis may enhance women's freedom of choice either to refuse this procedure without being labelled irresponsible or give them the opportunity to terminate the pregnancy without feeling guilt and shame.

[50]Rapp 1999.
[51]Rapp 1999.
[52]Rapp 1999.
[53]Hadley 1998.
[54]Hashilony-Dolev 2006.

6 Critical Discussion

Feminist approaches to genetic counselling mirror feminist approaches to reproductive technology. As Charis Thompson[55] argued in her review of feminist literature on infertility, most of the early feminist writings on assisted reproductive technology (ART) expressed "moral certainty" that hi-tech reproduction was bad for women. However, since the last decade of the twentieth century, there has been a shift in the feminist literature towards "moral ambivalence" regarding these technologies, which are increasingly viewed as enabling women to exercise reproductive choice.

Perhaps the time has come to also examine the criticisms levelled at genetic counselling from a more critical point of view. Genetic counselling has been a target of feminist criticism throughout almost its entire short life. As a genetic counsellor and a feminist, I feel both qualified and motivated to respond to these criticisms.

Regarding the criticism of genetic counselling as part of a coercive, paternalistic and patriarchal scientific agenda of ART in general aimed at controlling women's fertility and health, it has long been suggested in the feminist literature that ART has advantages for some women as well as disadvantages for others.[56] Moreover, some feminists view In Vitro Fertilisation (IVF) as a means of regaining control when infertility is perceived as loss of control.[57]

In "Liberation or oppression? Radical feminism and *in vitro* fertilisation", Elaine Denny[58] criticises radical feminists for portraying infertile women as passively accepting the control of a male medical profession. According to her, the experiences of individual women have been lacking from most radical feminist literature. She offers an alternative feminist perspective by claiming that in her sample of women undergoing IVF treatments, accepting infertility was perceived as passivity, while making decisions and using the technology meant taking control.

In response to the disability rights advocates' criticism and the proponent of the social model of disability, I believe we should remember that disability may cause suffering regardless of social constraints and contexts. Not *all* the negative experiences people with disability have are because of the construction of disability by society in general and genetic counsellors in particular. When addressing the weaknesses of the model, Shakespeare himself notes that:

> The social model so strongly disowns individual and medical approaches, that it risks implying that impairment is not a problem. Whereas other socio-political accounts of disability have developed the important insight that people with impairment are disabled by society as well as by their bodies, the social model suggests that people are disabled by

[55]Thompson 2006.

[56]Shalev 2006.

[57]Haelyon 2006.

[58]Denny 1974.

> society not by their bodies. Rather than simply opposing medicalisation, it can be interpreted as rejecting medical prevention, rehabilitation or cure of impairment. . ..[59]

Physical pain, inability to participate in some activities and shortening of life span are only few examples of obstacles people with disability may have regardless of society's attitudes towards them. Many genetic counselling sessions are self-indicated by persons with disability who wish to spare their prospective offspring from suffering the same condition. In such cases, how should the empathic counsellor address the desperate wish of this fully experienced person and his perspectives on disability?

A related point concerns the fact that mothers are still the primary caregivers. For women from low socioeconomic background having to care for a dependent child for many years could be limiting if not devastating in terms of work, education and personal wellbeing for herself, the disabled child and his or her siblings.

One final issue I would like to address with regard to the second criticism is its explicit or implicit approach to the disabled community as a uniform group united in its condemnation of genetic counselling, prenatal testing and termination of pregnancy. For example, representatives of this group, Patterson and Satz, recommend that women diagnosed with a foetus with Down syndrome read books written by people with Down syndrome or their parents.[60] I feel that such recommendations are unjustified and generalising: As mentioned above, some people with disability are against genetic counselling, prenatal diagnosis and pregnancy terminations, while other people with disability will consider using these practices in some situations. The complexity of such generalisations has been illustrated in several recent studies. Aviad Raz[61] found that leaders of organisations for disability rights and support groups for people with genetic conditions in Israel were generally in favour of prenatal genetic testing as well as selective abortion, unlike many of their counterparts in North America and Europe. Felicity K. Boardman[62] interviewed people with spinal muscular atrophy (SMA) in their family and reported that some of them wished to prevent this condition in future offspring and chose to undergo prenatal testing while others felt comfortable with the possibility of their child having SMA. Addressing future parents to discuss the issue of disability with people with disability or their parents can be problematic. People with disability, as women, are not made of one piece, each one has its own beliefs, attitudes and desires.

The third criticism levelled by contemporary feminists against the presumed nondirectiveness and value neutrality of genetic counselling is directed against the very normative core of genetic counselling and thus finds fault in the discipline as a whole. In my view, genetic counselling practice today is closer to the National Society of Genetic Counsellors' definition: "Genetic counselling is the process of

[59] Shakespeare 2013, 219–220.

[60] Patterson 2002.

[61] Raz 2004.

[62] Boardmen 2014.

helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease". This process integrates the following: interpretation of family and medical histories; education about inheritance, testing, management, prevention, resources and research and counselling to promote informed choices and adaptations to the risk or condition.[63]

Moreover, an important key value of genetic counselling refers to the patient as a whole in the context of the family environment, culture, community and belief system.[64] Kessler describes quality genetic counselling as an interaction in which the counsellor contacts clients on a human level and leaves them in a more cognitively and affectively integrated place than when their contact began.[65] As for ND, he explains it as an active strategy to assist clients to achieve personal health-related goals. One of the tasks of non-directive genetic counselling is (and has always been) to help clients make personally relevant decisions by helping them think through the various options open to them, grapple with the meaning of various choices for themselves and their greater family in both the short and long term and identify and attempt to defuse the obstacles, affective and otherwise, in the way of their autonomous decision-making.[66]

ND is the core value of genetic counselling. Despite its known limitations, it is essential in helping the woman counselee make a personal decision in keeping with her moral principles and beliefs and thus truly empowering her. Indeed, the education of genetic counsellors regarding disability studies and cultural differences should be further improved, and attempts should be made in order to alter the present composition of the genetic counsellor cohort to more closely match the demographics of patients. However, in my view, above all, genetic counselling is a unique type of professional relationship in which, at each step of the way, the counsellor attempts to evoke the counselees' competence and ability for self-direction. This target can be achieved by paying attention to the strengths, accomplishments and competencies that clients bring with them to genetic counselling and reinforce them when needed.

Several researchers have addressed these issues. Attitudes of Israeli women diagnosed with mutation in BRCA1 or BRCA2 genes who underwent genetic counselling were explored and one of the main themes revealed was that following counselling, women felt that "knowledge is power".[67] In a research assessing the motivation and outcomes of carrier testing in Britain, reproductive empowerment emerged as the central phenomenon. Participants were able to make informed decisions, regain control over their reproductive risk and pass on information to family members. Counselees reported that the main motivator and outcome was reproductive empowerment.[68] Finally, interviews were conducted with adolescents

[63]Resta 2006, 79.
[64]Guimarães 2013.
[65]Kessler 2001.
[66]Kessler 2001.
[67]Dagan 2009.
[68]Lewis 2012.

diagnosed with a genetic condition between the ages of 12 and 18 years who received counselling in Canada. Findings included understanding the genetic counsellor's role, greater perceived personal control and adaptation to one's condition. Authors conclude that genetic counselling can play an important role in providing information and support to this patient population.[69]

Feminists seeking to empower women emphasise concepts of choice and autonomy and the liberating potential of knowledge. They share with other writing on medical ethics a deep appreciation for patient autonomy in treatment decision making. Genuine autonomy requires that decision makers possess both sufficient knowledge and options to make careful informed choices about their lives.[70] In their daily work, genetic counsellors educate women counselees regarding the genetic condition under discussion, introduce their options and promote informed choices. Given the profession's strong ethical backbone, I feel that genetic counselling should not be the target of feminist criticism but rather be celebrated or at least encouraged as context—admittedly still unique in our society—in which one woman supports another throughout the challenging and complex process of making an autonomous reproductive decision.

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[69] Pichini 2016.

[70] Asch 1996.

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The Evolving Concept of Non-directiveness in Genetic Counselling

Angus Clarke

Abstract Debates about the core values of genetic counselling have drawn on various conceptions of 'non-directiveness' as a point of reference throughout the second half of the twentieth century. The use made of this concept has varied over this period, reflecting an evolution of the identity of the genetic counselling profession (in the broader sense of the practitioners of genetic counselling, thereby including many clinical geneticists). The term was used in the early phase of genetics clinics (up until about 1970) as a way to stress the difference from the former eugenics clinics. It became established as a key aspect of professional identity and was readily applicable to genetic counselling for decisions about reproduction and about predictive testing for essentially untreatable disorders (notably Huntington's disease).

More recently, as clinical genetics has become more widely relevant in medical practice and decisions about treatment, as in oncology and cardiology, the concept has seemed less relevant as it is good practice to make recommendations about genetic testing and surveillance for complications of disease. Some practitioners have wanted to move on from the term, suggesting it has been superseded, but it is still widely used in discussions of the ethos of clinical genetics and genetic counselling. In short, an assessment of (non)directiveness in the self-concept of the genetic counselling professional allows us to track the evolution of professional identity over more than six decades.

The nature of medical genetics services has changed from being primarily concerned with reproduction and dysmorphology to a much wider role in health care and the assessment and management of the risk of developing diseases and disease complications. Along with this, there is now a much greater scope for professionals to make direct recommendations on appropriate decisions and

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behaviours, so that a blanket approach to questions of (non)directiveness would now appear simplistic. Furthermore, the ethically grounded recommendation to patients that they share information within the family and take into account the interests and wishes of other family members when making decisions about genetic testing need not be in conflict with an appropriately nuanced conception of non-directiveness.

A focus on non-directiveness can illuminate many social processes, from the research evaluation of interactions within a consultation along the spectrum of directiveness to the pattern of provision of clinical services and wider aspects of social life including stigmatisation and the processes of resource allocation in health and social care. Many of these areas have received inadequate attention and (non)directiveness has great potential as an analytic tool to examine them.

One of the principal philosophical values upheld in much medical ethics is that of respect for autonomy, but discussions of (non)directiveness often employ a shallow notion of autonomy, especially when non-directiveness is regarded as equivalent to neutrality. Discussion of (non)directiveness may therefore have implications for debates on informed consent more generally, as the depth of understanding required for someone to give consent to a test or treatment varies with the context. A patient's unwillingness to engage in hypothetical discussions about possible future scenarios could lead to inappropriate decisions to deny them genetics services, if the professional assessed the quality of their informed consent as being inadequate.

Keywords: Genetic counselling • Non-directiveness • Neutrality • Patient-centred • Autonomy

1 Introduction

The term 'non-directiveness' has served as a key concept in the self-understanding of those engaged in genetic counselling since this developed as an activity after World War II. As clinical genetics and genetic counselling have emerged and evolved into distinct professional groups, this term has remained in use although its meaning and attitudes towards non-directiveness have changed, as the conceptual landscape of science and medicine has changed around it. The term itself is somewhat unsatisfactory as a key organising concept because it is framed negatively, as *NON*-directiveness: should the profession not choose a more positive concept around which to organise its identity?

This essay relates the term 'non-directiveness' to the philosophical concept 'autonomy' and aims to chart the reasons for the persistence of non-directiveness through some 60 years of major change. We ask whether it is still a helpful term and whether it has uses or applications that remain valid today, suggesting that it may indeed still have some uses for genetics health professionals and perhaps as an analytic tool.

2 Origin of 'Non-directiveness'

First, how do we define 'non-directiveness'? We will define this as an approach to genetic counselling that aims not to guide the patient (or client) to an outcome predetermined by the counsellor or the genetics service but instead to support the patient in reaching their own decisions. We will also refer to the client as a patient, whether or not they currently have a disease or any related health problems, as the term 'client' seems too close for comfort to the term 'customer' and denies the extensive blurring of the boundaries between being affected by a genetic disorder as a patient, being at risk of developing such a disorder in the future, being at risk of having an affected child or being affected less directly through a close relationship to someone who is clearly a patient.

The early history of genetic counselling was given by Sheldon Reed (1974) and that account makes clear the reasons for the negative framing of non-directiveness as a deliberate reaction against—a distancing from—the social and scientific movement of eugenics. Many of the early eugenicists, in the nineteenth and early twentieth centuries, were clearly idealistic, if with hindsight misguided, and they had the welfare of future generations at heart. The movement attracted support from across the political spectrum and their activities were generally benign although paternalistic. The terrible abuses committed by nationalist socialist ideology in post-1933 Germany—with its twin doctrines of racial superiority and racial hygiene—shocked a generation, so that almost anyone working in this field after WWII had to distance themselves from that dark past, although there were some unrepentant eugenicists in Germany itself.[1] What is even more shocking, however, is that harsh forms of eugenic practices continued in many Western countries, as with compulsory sterilisations in North America and Sweden, and illegitimate—unethical, even actively harmful—research persisted, without patient consent, as if no lessons had been learned from the horrors of Nazi medical experimentation. The Tuskegee syphilis study is merely one of the best known examples but there are others. The need for the Helsinki Declaration and for strict ethical oversight of medical research has been clear ever since.[2]

The term 'genetic counselling' was coined in 1947 by Sheldon Reed, who saw genetic counselling as a type of social work intended for the benefit of each family rather than for the state or the population.[3] Many early practitioners, in the 1940s–1960s, provided 'genetic advice' rather than 'genetic counselling', and, indeed, many of these early practitioners in both the USA and UK were non-clinical scientists, just as many in pre-war Germany had been physical anthropologists and not physicians. The benefits to be gained from genetics clinics were often framed in terms of benefits to society and the population and perhaps a rather long-

[1] Müller-Hill 1988; Harper 1996.

[2] World Medical Association 2013.

[3] Reed 1974.

term benefit to future generations.[4] The psychotherapeutic and counselling aspects of the genetics clinics developed rather slowly, drawing inspiration from the client-centred movement in psychotherapy and counselling, associated specially with the work of Carl Rogers (1902–1987) in California. His ideas, as part of the milieu of the 1960s, were drawn upon by those working in the early genetics clinics in the 1960s and 1970s. Publications describing the experience of these early clinics were often rather hybrid, with some attention to the emotional experiences and needs of the patients and at the same time some focus on the more objective 'effectiveness' of the genetic counselling, by which was usually meant some impact on a couple's reproductive plans or their decisions about future pregnancies.[5] The essential distinction is whether the benefits of genetic counselling are seen as accruing to the individual and their family or to the population more broadly. In principle, perhaps, there need be no conflict between these two perspectives, but in practice they encourage different patterns of service provision and very different styles of practice.

3 Reasons for the Persistence of 'Non-directiveness'

Professionals gain something from using the term 'non-directiveness'. They not only distance themselves from eugenics and can feel virtuous in being different from 'those bad people from the past', but, in addition, they gain emotional distance from their current patients' decisions. Furthermore, this helps the practitioner to assert that the legal responsibility for any decision made by a patient lies with the patient. In a litigious society, that clarity may be helpful. Using the term 'non-directiveness' may also allow professionals to align themselves with those who reject the use of cost-saving arguments for antenatal screening (see below).

Through the collective assertion of these aspects of non-directiveness, the genetic counselling profession has helped to reinforce these benefits of being non-directive and actively reproduces them through training student genetic counsellors in this approach. However, there are possible disadvantages that come packaged with this concept. We will turn to consider them once we have considered how genetic services changed with the legalisation of abortion in many jurisdictions.

[4]Carter 1951.

[5]e.g. Morris and Laurence 1976.

4 Abortion and Antenatal Screening Programmes

The widespread legislative changes in the 1960s and 1970s, permitting the termination of pregnancy on medical grounds including the finding of foetal abnormality, led to changes in 'genetic counselling', broadly understood. Alongside the legal changes, and partly driving them, were technical developments in obstetrics that made possible the prenatal diagnosis of neural tube defects and, later, of Down syndrome, at first using amniocentesis but then increasingly a combination of maternal blood samples and foetal ultrasound scans.

While some forms of neural tube defect (NTD), and Down syndrome with complex congenital heart disease, will often be lethal—still today but more so in the 1970s—other cases of NTD and of Down syndrome will often have a good (i.e. lengthy) life expectancy and may therefore be seen as 'costly' disorders. In the amoral world of health economics, a programme of antenatal screening to identify and then terminate pregnancies with an affected foetus had the potential to save money through avoiding the future health and social care costs of those affected children who would no longer be born. This potential justification of an antenatal population genetic screening programme was promoted by at least some clinical geneticists in many countries as well as by those who would be actively engaged in setting up or delivering the screening. There were numerous studies of antenatal screening that addressed the question of the cost of a programme in terms of the cost per 'expensive' foetus terminated[6]; few such papers faced up to the ethical challenges as clearly as did Hagard and Carter (1976).[7] These authors showed that the costings could work out in favour of screening-plus-termination but then called for a debate as to whether such a policy was wanted or would be ethical. That public process of deliberation has been largely side-stepped in favour of individuals making choices within a social context that encourages such screening. Abandoning parents to make their own decisions and then live with the consequences is what autonomy has sometimes come to mean; for the Thatcherite neoliberal, respect for autonomy means, 'Stand on your own two feet!'

The extent to which genetic counsellors as a profession are involved in antenatal screening programmes varies between countries. In the USA, they are often key players in the information-discussion-and-consent process before a patient chooses whether to accept screening. In the UK, they are hardly involved at all, unless an anomaly is found or suspected, as midwives undertake the initial discussions; other countries and even centres differ again in their practices and in their recognition of genetic counsellors as a distinct professional group. This engagement in antenatal screening is important for the profession's relationship with non-directiveness as the routinised nature of the offer of screening, and the clear expectation of many antenatal clinic staff that patients should participate, may exert a strong social

[6] e.g. Gilbert 2001.

[7] Hagard 1976.

pressure that patients should agree.[8] The choice is not neutral: the decision not to participate is dispreferred, often being seen as deviant so that it requires justification, while a decision to participate is accepted without demur. This asymmetry may be appropriate in the context of many of the routine newborn screening tests but inappropriate antenatally, as for non-therapeutic newborn screening, so that changes to the clinical process may be needed to help patients make a considered choice in accord with their values.[9] The overall pattern of organisation of the antenatal clinic may lead patients to make particular choices—being strongly directive—even though no one is told what to do. This is a structural form of directiveness that it may be difficult to counter.[10] It may require the professional to work hard with each patient to persuade them that they (the professional) are making a service available without promoting it or recommending it. This takes active, emotionally charged, work as many patients will think, 'Why would they make this available to everyone if we were not in fact expected to take part?' A commitment to non-directiveness as a value can then help the professional to counter the assumptions generated in patients by the systems of care in place. This will benefit both the counsellor and her patients.

Where genetic counsellors play a role in antenatal screening, as in one Australian centre where they see the cases identified as at increased risk of Down syndrome, the clinic appointment focuses very much on the transfer of information and not so much on the patient's thoughts and feelings and the *counselling* aspects of the setting.[11]

The underlying reasoning in favour of screening draws upon an administrative or bureaucratic rationality that ignores the value of human life and merely counts costs. This is one of the two contemporary Western forms of eugenics, the other being a consumerist eugenics discussed by Troy Duster (1990).[12] One problem with this bureaucratic approach to screening programmes is the inbuilt tendency to maximise the 'efficiencies' of the system. In some contexts that may be desirable but in antenatal screening it means that any birth of an affected infant triggers mechanisms that monitor and audit the performance of the screening programme; this very fact could well reinforce professional practices designed to minimise the chance of another case arising: i.e. to strengthen the structural directiveness of the whole system.[13] A more detailed review of this issue is found in two chapters from 1997.[14] Contemporary eugenics takes different forms in the Islamic Middle East and in East and South-East Asia, but we do not have space to expand upon those phenomena here.

[8]Rothman 1988; Rapp 2000.
[9]Parsons 2000.
[10]Clarke 1991.
[11]Hodgson 2010.
[12]Duster 1990.
[13]Clarke 1990.
[14]Clarke 1997a; Clarke 1997b.

5 Potential Difficulties in Screening

There are several ways in which screening programmes can challenge professional efforts to be non-directive, in addition to those considered above. One is the question of what information to give about the conditions that screening is intended to 'avoid'. How can one give 'balanced' information?[15] Is that notion coherent, when the goal of screening is to reduce (or even minimise) the birth incidence of the condition? 'Society' has in some sense taken a decision that the condition being screened for is undesirable, despite protestations that the goal of the programme is to maximise 'informed reproductive decisions' rather than achieving a fall in the birth incidence of a disorder or a fall in the cost of health and social care.[16] However, simply increasing 'informed reproductive decisions' is not a coherent goal and begs too many questions relating to the broader social context. This is addressed from a legal perspective by Dunne and Warren (1998),[17] who describe the directiveness of the framing within which antenatal screening is offered. As a remedy, they suggest that information about Down syndrome should be provided by members of the Down syndrome support group to help counter this, although they fail to recognise that such an intervention may well have unanticipated consequences. This remedy may itself be perceived as also being directive, in the sense of attempting to manipulate the patients' decisions. In addition, it might well also cause distress to those who exhibit their private lives as the parents of children with Down syndrome, when at least some of those they meet will then choose to accept screening or to terminate an affected pregnancy.

The disability rights critique of antenatal screening is highly relevant as it refers to the offensive nature of such screening, from the perspective of affected individuals who consider their lives to be worthwhile but who feel disrespected by society, even without being a target of antenatal screening.[18] To promote screening in the face of such opposition suggests that supporters see real benefit in screening and wish to promote it. On the other hand, not to make screening available may also lead to anger and distress and can be seen as a denial of basic health care. The decisions made by society in setting up antenatal screening are difficult, as are the choices to be made by patients: no single policy will suit everyone. Indeed, the choices offered in antenatal screening may be most unwelcome, as the mere offer of screening imposes a choice with which people may prefer not to be confronted.[19]

The possibility that screening might lead to the offer of a termination of a wanted pregnancy needs to be discussed at entry to screening for a patient's decision to be grounded in reality and for the potential seriousness of screening to be appreciated. Such discussions will be difficult for many patients and will be draining for staff

[15]Lippman 1992; Hippman 2012.

[16]Modell 1993.

[17]Dunne 1998.

[18]Parens 1999.

[19]Dworkin 1988; van Berkel 1999; Hildt 2002.

when repeated many times a day: it is all too easy to see why a *full* discussion may be delicately avoided by both parties, so that the talk remains on the level of information transfer as education. This is a failure to address the emotional and ethical issues, i.e. to keep the discussion in clinic fixed on an educational level and not deepen it by orienting to the counselling aspects, which would be required for professionals to counter any systemic, structural directiveness.[20]

6 Making the Decision

Any assessment of the process of making reproductive decisions must address the contrast between the often routinised and superficial discussion about screening when the question of abortion is remote and hypothetical, and the more focused discussion about a decision that has to be made 'for real' about whether to interrupt or continue an affected pregnancy.[21] The hypothetical talk may be brief or it may cover a wide range of topics—concerning the family and society, politics and religion, identity—while the discussion in the face of a 'for real' decision, about whether to terminate that particular pregnancy, is much more focused and pragmatic, not usually influenced by those more theoretical considerations that might appear crucial if discussing abortion in the abstract. This leads us back to the question of the information to be provided about Down sydrome, spina bifida and other disorders: what would a 'balanced' account look like?

The opportunities for miscommunication in the antenatal clinic are increased substantially when the professional(s) and the patient/family come from different cultures or linguistic communities. The failure of a professional to recommend a genetic test, for example, may be misinterpreted as suggesting that the test would be inappropriate and should not be performed.[22] Equally, the social role of the health professional in a society may entail taking responsibility for recommending a particular course of action. Not to do so, in that society, would suggest incompetence or a lack of concern. Of course, the same difficulty could also sometimes arise within a society, simply because of contrasting personality types.[23]

Recent studies of decisions about antenatal screening show the complexity of such decisions and report on the different styles of making decisions. For some, the decision about whether to accept screening is relatively simple, but, for others, it is complex and difficult.[24] At least in Scandinavia, the reasons underlying a decision to decline screening often relate to the parents' philosophy of life and the value they place on disability and diversity.[25] But there are often burdensome, practical

[20]Hodgson 2010.
[21]Erikson 2003.
[22]Browner 2003.
[23]Wüstner 2003.
[24]Wätterbjörk 2013.
[25]Gottfredsdottir 2009.

consequences when a patient makes the dispreferred decision in a screening programme, i.e. that she declines the offer of screening. This decision will then have to be voiced and justified at every contact with health professionals throughout the pregnancy. Even if they do not mean to challenge the woman or try to persuade her to change her mind, this repeated call for explanation may easily be experienced as coercive pressure.

7 Contesting Autonomy

We have now looked at the origin of 'non-directiveness' and the reason why many genetics health professionals have chosen to continue using this negatively framed, process-oriented term as a key principle. We have also looked at programmes of antenatal screening that may involve genetic counsellors and we have considered the marked similarities between such genetic screening programmes and the population-oriented goals of the eugenics movement.

We will now pause to consider the most helpful approach to take in reporting the shifts of understanding and opinion concerning the concept of autonomy and how these relate to 'non-directiveness'. The initial position of medical paternalism persisted after WWII through the 1950s and into the 1960s but then weakened, as respect for autonomy gained in strength and recognition. The growth in respect for autonomy led to a shallow form of respect for the choices made by patients as if they were choices made by a shop customer choosing between different loaves of bread; their choice could have been carefully considered but could also have been mere whim. This shallow respect for autonomy may be understood as the professional stance of neutrality, perhaps an overreaction to the previous paternalism. Attention then began to focus on the quality, not of decisions made, but of the making of the decisions. One could say that the Anglo-Saxon utilitarian philosopher John Stuart Mill (1806–1873) was challenged by Immanuel Kant (1724–1804). Kant's notion of autonomy is more complex than Mill's, in that a Kantian genetic counsellor might attend to how a patient was making their decisions rather than simply accepting what they said. They might wish to challenge the patient and try to help them improve the clarity of their thinking, whereas a follower of Mill would ensure only that the patient's decisions would not damage the interests of others. This process of considering the quality of the making of decisions was reported from around 1980 and has continued ever since. As Shoshana Shiloh says, genetic counsellors (should be) '[. . .] helping clients reach a decision wisely, rather than reach a wise decision'.[26]

It would be difficult to give this history as a linear sequence of events. It will make more sense to present several points along this moral journey or perspectives that illuminate it. The developments have not all been unidirectional and nor has

[26]Shiloh 1996.

progress been made in all areas at once. The points and perspectives we have chosen are:

- Addressing patients' understanding of risk assessments and probabilities
- Influences on reproductive decisions
- The unsatisfactory nature of medical paternalism and the shift to non-directiveness
- The confusion between non-directiveness and neutrality
- Empty autonomy: ethics in a vacuum
- Assessing genetic services: studies of process and the shift to patient-centred outcomes
- Making recommendations about content and about process
- Beyond non-directiveness

8 Probabilities

Prior to any consideration about the impact of genetic information on patient choices comes an awareness that this will be shaped by how the information given is understood. The need for this was discussed by Pearn (1973)[27] and was then opened up more substantially by Abby Lippman.[28] More recent work has expanded on this, but this framework set the scene and showed that a client centredness was essential to any genetic counselling practice.

9 Reproductive Decisions

We have already seen that population-based programmes of antenatal screening have often been developed by enthusiasts who have seen the goals of the programme as at least partly related to its impact on the population and, therefore, the costs of public health care. To what extent have these population goals played a part in the acceptance of the need for genetic services by government? In the UK, I am sure that the explicit link between the provision of services and future health care costs played an important part in the establishment of antenatal screening programmes. It is less clear that this was true for clinical genetic services more generally, although I strongly suspect that this was the case; at the very least, I am certain that some involved in promoting the new clinical specialty presented it to government in that light.

[27]Pearn 1973.

[28]Lippman-Hand 1979a; Lippman-Hand 1979b; Lippman-Hand 1979c.

Considering the impact on patients and families, however, is very different from looking at the effects on the population. In the setting of prenatal diagnosis for a high-risk Mendelian disease, the factors that influence a patient's decision about a pregnancy have been studied in surveys and in smaller, interview-based studies. Thus, a survey of those who had utilised genetic counselling services shows that patients do not generally experience being pushed or led to make particular decisions by their genetic counsellor (e.g. to terminate a pregnancy).[29] Such evidence is reassuring for genetic counsellors that, in contrast to the situation in antenatal screening, patients do not generally feel directed or coerced by their experience of genetic counselling. Petra Frets et al. (1999) showed that the availability of prenatal diagnosis makes a very substantial difference to the reproductive plans of couples at high risk of transmitting a serious disorder, so these potential effects have increased as prenatal diagnosis has become available for ever more conditions. The strength of the desire for children and a lack of direct experience of the disorder in the family are also associated with the decision to have a child.[30]

One interesting development in the UK of the 1990s had interesting implications for clinical genetic services: the Royal College of Physicians Confidential Inquiry into Counselling for Genetic Disorders. One example of their work was a study of families with two siblings affected by cystic fibrosis,[31] which set out to review whether families in which there had been a recurrence had been offered genetic counselling that could have enabled them to avoid a second affected child. This is naturally of interest to a clinician, but the modelling of this programme on the confidential inquiries into maternal deaths did convey a sense that recurrences had to be avoided (prevented) and that a recurrence represented, in some way, a failure of practice. While the Inquiry was clear that a recurrence, where the family had chosen not to pursue the possibility of prenatal diagnosis, was not seen as a problem, the borrowing of the format and framing of the project from the confidential inquiries into maternal deaths had the implicit effect of reinforcing the idea that genetic services were all about disease prevention, just as the primary purpose of obstetrics is to help mothers to deliver their babies without dying in the process. The initiative helped to maintain professional attention on the population aspects of genetic services as well as seeking to identify inequitable provision. Whether it was designed specifically to emphasise the cost benefits of genetic services, and thereby to help to justify support in government and policy circles, is unclear.

[29]Wertz 1986.

[30]Frets 1990.

[31]Lane 1997.

10 The Shift from Paternalism to Non-directiveness

This is the easiest of the transitions to recognise and appreciate. The rejection of eugenics did not instantly transform the patient–professional relationship in genetic consultations, but there was a slow process of change as 'non-directiveness' came not only to be accepted in theory but was also integrated into practice. This required a change in manner of the professional and of expectations in the patient and neither happened overnight. The essential point of non-directiveness in genetic counselling is for the professional not to predetermine the outcome but to enable the patient to make the best adjustment and the best decision(s) s/he can from their perspective.

Similar changes were happening in medicine as a whole, but the changes in genetic services were perhaps accelerated by the need for genetic counsellors to distance the profession from the medical authoritarianism associated with eugenics. The changes within medicine led to the Patient-Centred Medicine movement in the 1980s,[32] by which time the client-centred approach derived from psychotherapy had long established itself within genetics. Seymour Kessler stated that non-directiveness was entrenched within genetic counselling in the USA by the late 1960s and internationally by the late 1980s.[33] Patient centredness of course developed unevenly across medicine, earlier and more thoroughly in some specialties than others: earlier in family medicine than internal medicine, and earlier there than in surgery, where the very nature of the specialty makes meaningful patient involvement in some of the decisions to be made rather more difficult to achieve.

11 The Confusion Between Non-directiveness and Neutrality

The distinction between non-directiveness and neutrality then becomes important as the words have sometimes been used as synonyms although the concepts are very different. 'Neutrality' suggests that the professional would provide information but then allow the patient to make their (informed) decision on the basis of whatever factors were important to them. With neutrality comes a danger of professional indifference and patient abandonment: the counsellor may be hoping not to pressure the patient, but s/he is experienced as unengaged, excessively detached from the patient and indifferent to them. While it is good for the professional to avoid over-involvement, as the distress of some families in the face of their genetic disorder may be profound and emotionally very challenging, but, if patients feel abandoned rather than supported by the professionals, then there has clearly been a failure of professional practice.[34]

[32]Weston 1995; Brown 1995.

[33]Kessler 1992.

[34]Quill 1995.

Non-directiveness is rather different from neutrality. The professional still has no particular outcome in mind—is not attempting to sway the patient to make one decision rather than another—but is using an active approach to the patient–professional relationship. The non-directive professional shows concern for the patient and their welfare—they are not indifferent—and they are interested in how the patient makes their decision. In fact, they are more interested in that than in the decision that is made. The non-directive professional values the patient and is concerned to provide information and explanation, to help the patient understand their situation and weigh the information they have been given. This allows them to make the best decision they can, a decision with which they, and not the counsellor, will then have to live.

Arthur Caplan (1993) drew attention to the distinction between neutrality and non-directiveness, arguing that neutrality had to be abandoned as it was clearly an inadequate anchor for professionals in supporting the making of decisions by patients.[35] Alex Huibers and Adriaan van't Spijkers (1998) pointed out that giving more information was not always the best way of supporting patients and that some types of information about the future might even undermine a patient's autonomy. Furthermore, conflicts might arise between a patient's autonomy and their other interests or between the autonomy of the patient and that of other members of the family: these conflicts of interest are not easy to solve.[36]

The experience and insight of a genetic counsellor, who has seen how other families make decisions and the aftermath, can lead them to challenge the initial judgements of a patient without wanting to supplant their right to make the eventual decision. They may be able to see that the patient has misunderstood some fact, or has not recognised the relevance of some aspect of the social context within the family, so that the counsellor then makes remarks that are direct although not 'directive'. They may recommend that the patient considers some potential consequences of their decision without this meaning that the professional is wanting to make or impose the decision.

Three interesting studies were published in 1997, all relevant to this debate. Susan Michie et al. (1997) coded statements made by genetic counsellors in consultations and rated them on a scale of directiveness. Unfortunately, the rating of these statements was decontextualised and they were rated on the basis of being direct (e.g. making a clear recommendation) and directness was then confused with directiveness. The basic error is to suppose that directiveness is intrinsic to the words used in a statement, rather than emerging from the use of statements in a particular context: the coding and analysis can be ever so 'objective' and 'reproducible' at the same time as being irrelevant. A genetic counsellor may well wish to make recommendations about the process of arriving at a decision without thereby be trying to steer or coerce the patient to make *this* decision rather than *that*.[37]

[35]Caplan 1993.
[36]Huibers 1998.
[37]Michie 1997.

van Zuuren (1997) studied neutrality in genetic consultations and, similarly, found that counsellors made statements that could be construed as 'directive', but these also were often directive in relation to minor aspects of the process rather than the key decisions to be made.[38] Finally, Dianne Bartels et al. (1997) asked genetic counsellors in the USA to rate their professional activity for its directiveness and found that some declared that they were, at times, 'directive'. This study was remarkable for demonstrating how finely attuned the professionals were to the subtle gradations between directing the process of genetic counselling and defining its outcomes. This paper suggested, indeed, that it might sometimes be appropriate for genetic counselling to be 'directive'. We will return below to the question of making recommendations.[39]

12 An Empty Autonomy: (Pure) Ethics in a (Social) Vacuum

A simple respect for autonomy might be thought to lead the genetics professional to accept, and to act upon, any decision or request uttered by a patient. This sense of autonomy is empty, in that it conceives of autonomy as a 'right' but in a (social) vacuum, as if we could make decisions on a whim, or out of caprice, and without having to consider the implications of the decision for those around us or even for ourselves in the future. One task of the genetic counsellor is to protect us from making such irresponsible and ill-considered decisions.

We have already considered, briefly and in caricature, the concepts of autonomy of Mill and of Kant. Neither philosopher would have accepted the mere indication of a choice as an autonomous decision to be taken seriously.

Controversies have raged about whether society should respect a person's choice deliberately to bring into the world a child with substantial impairments, in circumstances where they could equally bring into existence a child without such an impairment (as in the operation of preimplantation genetic diagnosis, PGD). This is not the place to rehearse these arguments, but they raise the question as to what type of impairments we might allow a patient to select for their child, at PGD or even at prenatal diagnosis. In the future, we might imagine facing questions about what impairments we would permit a parent to introduce into their child through genome editing.

Would it be acceptable to select for genetic deafness in a child? Or to deafen a child postnatally with gentamicin treatment? Or to use gene editing technology to achieve the same end? How do we respond to this point with arguably more serious impairments such as achondroplasia that can be progressively disabling and is

[38] van Zuuren 1997.

[39] Bartels 1997.

sometimes lethal? Or limb reduction defects, such as those resulting from maternal thalidomide ingestion?

In short, how do our discussions about non-directiveness in genetic counselling relate to these debates about the value of lives lived with such impairments or diseases and the right of parents to make choices about the type of children they wish to have?

We rapidly end up in a fairly abstract realm of ethics, which can be very different in tone from the discussion about real-life scenarios in the genetics clinic. A practitioner may find it difficult to take seriously some of the ethicists' dilemmas, as these feel as if they have been—and indeed they have been—contrived so as to illustrate a point of argument. I would simply counsel that clinicians should remain in touch with these debates as they may become relevant sooner than we can imagine and it is also useful to keep ethicists engaged with issues as close to the real world as they will let themselves be confined.

One particular point that is highly relevant to genetic counselling practice, and that has been raised for discussion by ethicists on several occasions and proposed as a contribution to practice, is the idea of raising questions of morality for discussion within the genetic counselling clinic. We are told that we should help patients to discuss their moral and ethical views and values and then help them to implement these in the decisions they make in clinic.[40] While the subtlety of these suggestions varies between the papers, a quick rejoinder is that it would be immensely difficult for the practitioner to raise the issue of ethics with the client without it being highly manipulative and potentially coercive. Indeed, how would one respond to an interrogation about one's values in relation to genetic testing, knowing that one's response would then be used as a standard against which you would be judged? Most people, other than professional philosophers, will not derive their ethical judgements from their values and then articulate them. Rather, statements ascribing values are likely to follow a person's intuitive ethical assessments, and serve to convey their sense of values to others.

It is quite different if a patient raises the question of ethics or religion or simply vague intuitions of what actions are (not) acceptable. Then the practitioner can engage with the patient's thoughts and feelings and help them explore what is most important for them: that is completely different (and entirely acceptable).

It is interesting that a study of patients making antenatal screening decisions has found that many do refer to moral principles but only after they have made their decision, once they are looking for reasons to justify, support and explain their decision.[41] While beliefs and commitments form the person who is making the decision, the role of these principles in making—i.e. in arriving at—their decision is much less clear.

[40]Stone 1999; Wüstner 2003; van Berkel 1999.

[41]Garcia 2008.

13 Assessing Genetic Services: Studies of Process and the Shift to Patient-Centred Outcomes

By the time Timothy Quill and Howard Brody (1996) were arguing for a balance between physician power and patient choice in medicine at large, the importance of recognising the new patient–professional relationship in genetic services had long been recognised.[42] Furthermore, the importance in the setting of goals for a service had been appreciated, as such goals can drive the ethos of a service and are therefore important influences on the patient experience and the measures of outcomes available at the time were completely unsatisfactory.[43]

One of the major developments to arise from the ferment of ideas about the evaluation of genetic services at this time was the concept of 'perceived personal control', which Shiloh and colleagues built into a scale for assessing the helpful effects of genetic counselling.[44] This work led to further, collaborative work on outcomes with Marion McAllister and her construction of 'patient empowerment', developed from qualitative research with patients and then consolidated as a quantitative scale that was validated and is now used as a patient-reported outcomes scale for clinical genetic services. Although this was not completed until 2011, the preparation for this had begun at least two decades earlier.[45] This fits neatly into the contemporary emphasis on patient-reported outcome measures (PROMs) across health care in general, ensuring that genetic services will not be sidelined simply because of a failure to have developed a PROM relevant to genetics.

14 Making Recommendations About Content and About Process

One of the major challenges to the focus on 'non-directiveness' in genetic counselling has been the increasing recognition that it can be appropriate—even essential—to make recommendations to patients. When a genetics clinic could only attempt to make a diagnosis, and perhaps give information about the risk of a condition recurring in a family, it was not so difficult to resist the temptation to give advice or tell people what to do, at least for those professionals who had thoroughly rejected eugenics and did not want the responsibility for how others would live their lives. However, once a patient's health could be influenced by genetic information, the situation changed profoundly. It then became natural and inevitable that clinical geneticists and genetic counsellors should give information to families, explaining

[42] Quill 1996.

[43] Clarke 1990; Clarke 1996.

[44] Berkenstadt 1999.

[45] McAllister 2011.

(for example) that those at risk of familial polyposis coli should have colonoscopies to monitor the development of polyps from at least their teenage years and then, if polyps were found, undergo colectomy to minimise their risk of cancer.

Numerous other clear clinical benefits of genetic information have been identified and no geneticist or genetic counsellor would want to deny their patients these important benefits through a failure to be clear that this application of genetic knowledge was vital for their welfare.

Some practitioners have found this nuanced position on non-directiveness to trouble them and they regard it as undermining the whole principle, which then (they feel) has to be jettisoned. The more nuanced view is to recognise that there are areas of genetic practice where non-directiveness is vital but other areas where genetic knowledge can bring clear and unambiguous benefit. Non-directiveness would be seen as crucial for decisions about reproduction and about predictive genetic testing where the point of the knowledge gained is not to bring a definite medical benefit but, rather, a choice as to how to live one's life: either in as full a knowledge of what the future is likely to bring as possible or choosing not to know (for the moment) and thereby retaining the uncertainty but also the hope that the feared disorder will pass you by.

There are many areas where genetic knowledge can bring medical benefit, as in newborn screening for some metabolic disorders, many of the familial cancer syndromes, some inherited cardiac disorders and more. In these conditions, and within boundaries, we can recommend a prudent course of action to our patients. These are recommendations about 'content' or 'substance'. There is another category of recommendations that we make, however, where the recommendations are about 'process'. Here, we can also make clear recommendations but of a different sort: we make recommendations, based on collective professional experience, about the general approach to genetic information within a family.

We make recommendations about the sharing of information with relatives, where this could be relevant to their own health or to the health of children they may have in the future. We work hard to persuade patients and their families not to deny information to their relatives that may be important to them.[46] Very occasionally, we may even encounter circumstances where we feel strongly that a family is withholding information from relatives inappropriately and that we should insist upon or force disclosure against their wishes, and against the traditional medical respect for confidentiality, so as to avoid serious harm. These extreme circumstances are most unusual, however, and would have to involve prior consultation with a medical lawyer. I am thankful that I have not yet been involved in such an episode.

We also work hard to persuade parents (and sometimes our colleagues) not to generate information about their children that will be of no medical significance to them as children but only in the future as adults, if we think that they (the one who is now a child but can be regarded as a future adult) may well wish to make their own

[46]Clarke 2005.

decision.[47] There has been a lot of debate about this topic, partly because of the different legal approach to children in the USA versus the UK, as parents are often free to make their own decisions about the health care of their children in the USA (where children seem to be something like their parents' property), whereas, in the UK, parents have an obligation to make decisions about their child in the best interests of that child. However, the debate within professional circles touches on other issues too: on what basis should professionals refuse to do what the parents have asked them to do? Some professionals (myself included) will often wish to protect the future rights of the child to make their own decision about genetic testing as an adult, while others (often of a utilitarian mentality) would only wish to interfere in the freedom of the parent if there were concrete evidence that harm would be likely to result. If we do not agree that an abrogation of future rights is in itself a harm, we end up in a CATCH-22 because only if we agree to do what the parents request, and cause the harm we would like to avoid, can we gather the evidence to justify not causing the harm. And even that is only half the story, as we would need agreement in advance as to what would count as evidence of harm, and some of the potential harms might not manifest for two or three decades. This is a fundamental value disagreement about what constitutes a harm to the developing child, but perhaps this is not the place to pursue this question any further.

While the UK and European policies make clear recommendations about these aspects of managing genetic information, the policies in the USA are less clear. For example, when would we or would we not accede to a parental request to undertake a predictive test on a child to satisfy, in effect, parental curiosity? The American Society of Human Genetics gives a clear and consistent message, similar to those of the European and UK societies, whereas the joint recommendations of the American Academy of Pediatrics and the American College of Medical Genetics are less clear and give room for practitioners to do whatever parents request. The precise line that is defended will vary between clinicians.

Turning now to studies of genetic consultations, the detailed turn-by-turn analysis of what is said in clinic, there are patterns that emerge in how clinicians attempt to support their patients in the making of decisions about predictive genetic testing. They raise for consideration issues and factors that the patients might prefer to ignore. They prompt, for example, reflection about hypothetical future scenarios: 'How would you feel if . . .the result was X? or Y? Or unclear? Or if you decided not to go ahead with testing (for now)?'.[48] Some patients are willing to engage with such reflective frames while others are not.[49] These reflective frames constitute one of the means through which counsellors bring non-directiveness into operation: they hope that such conversations will allow the patient to recognise the likely consequences for them of the various possible courses of action and thereby enable them to select the most appropriate path. When a patient chooses not to engage with

[47]Harper 1990; Clinical Genetics Society 1994.
[48]Sarangi 2004.
[49]Sarangi 2005.

this reflective approach, the counsellor may feel frustrated and will try to persuade the patient to do so. If they continue to resist engagement, or if they are simply unable to engage for other reasons, the counsellor can give advice about how they should approach the making of their decision but will still not tell the patient what decision to make.

The expectation on the part of the counsellor that the patient will (should) engage in reflection on hypothetical scenarios can generate problems, including the feeling in the patient that they have to satisfy the counsellor in some way to gain access to the test. When an at-risk patient has a history of psychiatric disease, there may be some weight behind this concern: the counsellor may be reluctant to proceed with the test when the patient is in an especially vulnerable mental state and/or socially unsupported. In such a situation, the counsellor may maintain contact with the patient but not proceed in the usual way until the patient's mental state and social situation are both more stable. In most situations, however, the counsellor will make explicit to the patient that it will be their (the patient's) decision whether to have the test, and they do not need to jump through hoops of reflection for the benefit of the counsellor. This explicit giving of control to the patient will sometimes help them to engage in the process, reflect on the question of testing and prepare either to go ahead or to defer the decision.

This process can amount to something like the collaborative process of 'shared decision-making', one form of patient-centred medicine, as applied to genetic counselling.[50] There are difficulties with this phrase, as the making of a decision can hardly be shared, and in most circumstances the action based on the decision is in the hands of one party or the other: the patient decides after the consultation whether actually to take the medication prescribed by the physician, or the counsellor decides whether to process the blood sample and perform the test. So there is always some asymmetry in the decision. However, the application of the non-directive focus on the making of the decision—how it is made—represents a real advance over earlier approaches to understanding the practitioner's role in the clinic.

Additional aspects of the counsellor's role can include an assessment of the level of understanding on which it is based, and of the independence of the patient in making the decision, although these topics cannot be pursued further here. Is the patient making their own decision or are they being obliged to seek testing at the insistence of others in the family? Can the counsellor 'protect' the patient from these pressures? One has to bear in mind also that the standard of understanding required for consent to a medical investigation is different from that needed for participation in a research project. The higher level required in research should not be demanded in a simple clinical context as the provision of health care should be available to all. Otherwise, healthcare would be denied to many quite unnecessarily, on spurious grounds.

[50]Elwyn 2000.

15 Beyond Non-directiveness

Let us take stock of non-directiveness as it is now understood by practitioners. It is an interactional accomplishment involving judgement as to how firmly one can challenge the patient. It is reserved to decisions about reproduction, especially the prenatal setting and topics such as testing to determine carrier status, and to predictive testing for adult-onset disorders where there is no effective treatment. Making recommendations is a thoroughly accepted practice in many other areas covered in genetic consultations.

The preference of some practitioners for the term 'psychosocial genetic counselling' comes from a wish to cover the full ambit of genetic counselling practice without excluding those areas where non-directiveness is not appropriate, the wish to emphasise that genetic information can have a powerful impact on the patient and on those around them, and the wish not to be defined by a negative term. These are very reasonable considerations,[51] although non-directiveness still seems useful in emphasising the determination not to repeat serious errors from the past that affect such very sensitive areas of human life. It can serve as one key means to keep alive the memory of past mistakes that were widespread across many Western countries, neither being confined to Nazi Germany nor stopping in 1945.

Kessler distinguishes genetic counselling from psychotherapy in that the client cannot sensibly control the agenda, pacing or direction of genetic counselling in the way s/he can of psychotherapy. It is therefore very different from a thorough-going client centredness. However, the genetic counsellor can set a goal of improving the process of decision-making through enhancing the patient's autonomy. Kessler (1997) defines non-directiveness as, 'procedures aimed at promoting the autonomy and self-directedness of the client'.[52]

Kessler's insight that genetic counsellors can have the goal of enhancing the autonomy of their patients has been vital. This allows genetic counsellors to work with patients so as to improve their ability to make specific decisions. We also hope that this skill will generalise and will often lead to patients using better approaches to the making of important decisions in the future. Kessler, we should note, was firmly opposed to the concept of directiveness as one thing and non-directiveness as the opposite. He was always clear that there is a spectrum and practitioner interventions and behaviours could lie at any point along it: it would all depend upon on the details of what was said and how.

Further work in this area will depend upon the methodology used. Kessler's training as a therapist gave him great sensitivity to the use of language, but another approach that can help the scholar or practitioner to examine language use in an equivalent way is to be found within linguistics, pragmatics and discourse analysis. In fact, for the purposes of research, this approach may have benefits in the ease of establishing and communicating an argument. Practitioners have a lot to gain by

[51] Weil 2003.

[52] Kessler 1997.

inviting researchers from these areas to examine the discussions in the genetic counselling clinic using approaches such as theme-oriented discourse analysis and ethnography and its variants.[53]

Another important and helpful approach to non-directiveness was set out by Gerhard Wolff and Christine Jung (1995). They emphasise that genetic counsellors are going to influence their patients, but they must be alert to the influence they have and monitor how it works. While it may be acceptable (i.e. good practice) to enhance the decision-making capacity of one's patient, it would be wrong for a genetic counsellor to manipulate a patient so as to exert silent, unacknowledged control over them. They set out those aspects of the consultation that the genetic counsellor should lead and control and discuss the ideas of others on these questions.[54] Another psychotherapist with rich experience of the issues that arise in genetic counselling practice is Christine Evans, whose study of the field is full of insights into these complex processes.[55]

There are many aspects of genetic counselling that would need to be discussed in any assessment of the state of 'non-directiveness' in contemporary genetic counselling practice. This chapter is not an assessment of non-directiveness in genetic counselling today but, rather, an account of how we have reached the current state of affairs. However, it would be appropriate at least to list some of the issues that are live today.

First, how do we manage patients who are not yet quite 'mature'? How should we gauge maturity in young people who seek predictive genetic testing? How, and how firmly, should we challenge them in the counselling we provide before testing? How should we gauge 'maturity' without assessing ability or willingness to reflect?

Finally, what factors must we look at outwith the genetic counselling clinic that are relevant to the delicate decisions people make? In the context of reproductive decisions, at least, there are some important factors that constrain patients' decisions. In addition to the 'medical' aspects of a disease, a disorder or disability/impairment are the social aspects and the lived experience of 'being affected by' the condition. Two aspects of this can make life much more difficult than the 'objective facts' might do alone. These aspects arise at the micro level of social interaction and at the macro level of politics and the social order.

At the micro level, stigma associated with a genetic disorder can be powerful. This can shape a person's life at least as much as the more 'biological' effects, and it can also shape the reproductive decisions that are made by them or other family members. While this was referred to in Erving Goffman's 1963 monograph,[56] it has become clear more recently that these effects do not only arise on the street in episodes with anonymous passers-by but are judgements made by close members of

[53]Roberts 2005; Scully 2007, using interpretive phenomenological analysis, which I provocatively regard as a variant of ethnography.

[54]Wolff 1995.

[55]Evans 2006.

[56]Goffman 1968.

the family and intimate partners. These decisions include decisions about reproduction, often informed by the witnessed stigmatisation of those affected and also the courtesy stigma experienced by those accompanying them.[57] How such processes can be managed so as to cause fewer difficulties for patients (especially those affected) is unclear, but awareness of the stigmatisation both on the street and even within the family must be a first step.

At the more macro level is the question of the social order and how society manages the questions of health and disease in the context of serious and increasing social inequality. Inequity in access to resources, especially to health care, creates the potential for wealth/poverty to influence a family's decisions about reproduction. The willingness to accept a child with a genetic disorder, perhaps familial or perhaps not, will be influenced by any factors including the family's confidence in the willingness of society to support the special health and social care needs and education of a child with serious problems of health and/or development. Those with limited financial resources may feel unable to continue with a pregnancy affected by a condition that would impose major demands on them unless they feel confident that society would support them and their child. Of course, there are many other factors involved in these decisions and no simple relationship between wealth and a family's willingness to welcome into the world a child with potentially serious problems. However, such decisions will be less stressful and perhaps cause less distress in societies where good health care, social support and education are available to all irrespective of family background and wealth.

When families have no confidence in society's support in hard times—or no trust that people's behaviour towards those who 'look different' on the street will generally be respectful—then non-directiveness on the part of genetic counsellors may play but a small role in their reproductive decisions. They will be swamped by other concerns.

16 Conclusion (A Challenge)

We value non-directiveness in genetic counselling, especially in the context of reproduction, because reproduction is such a private and intimate area of human life and so tightly bound up with identity. However, these sentiments may not seem so powerful or persuasive outside our little Western bubble of relative opulence. If you live in a country where childhood mortality has only recently fallen, and the importance of genetic disorders such as beta-thalassaemia has only recently been recognised, what options do you have for the treatment of such conditions?

Until recently, affected children would die of malnutrition and intercurrent illness and with the thalassaemia often not being recognised. At present, affected children are recognised and pilot programmes of blood transfusion have recently

[57] Boardman 2014; Clarke 2013; Clarke 2016.

been established. You cannot yet afford iron chelation therapy, to prevent the iron toxicity from regular transfusion, but that may soon become possible with aid channelled through WHO, the World Bank or other sources. However, in areas with transfusion services, the number of children requiring transfusion is increasing rapidly (as fewer children are dying). The only way you can introduce both transfusion and iron chelation across the country would be if the birth incidence of beta-thalassaemia was to fall drastically. The only way to achieve that would be to introduce beta-thalassaemia carrier screening for couples, before marriage, and prenatal diagnosis with the selective termination of affected pregnancies. Without such a programme of prevention, the steady increase in size of the group of affected but treated children would increase year-on-year, as the mortality has plummeted, so that the cost of the transfusion-plus-chelation programme would soon absorb more than the country's entire health care budget.

In such circumstances, is it not right for the government to promote carrier screening and the termination of affected pregnancies as obligations for the population? How can one do anything but support such a programme with great enthusiasm?

If this is a fair assessment of the situation of countries that have recently gone through the demographic transition, is it equally fair to ask whether 'non-directiveness' is a luxury that can be afforded by prosperous, Western developed countries but that is out of reach for most other countries? How will these very arguments play out in wealthy Western countries as new, rational and effective but prohibitively expensive treatments are developed for increasing numbers of rare genetic diseases? Or is there a way to cut through this Gordian knot, by trusting to information and consent in rich and poor countries alike?

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A Comparative and Social History of Genetic Counselling?

Jean-Paul Gaudilliere

Abstract The existing historiography of genetic counselling focuses on the problematic relationship of medical genetics and eugenics with a strong focus on developments in the USA, France and Great Britain. This comment discusses the shared scenario for the advent of genetic counselling this rich collection of new national histories brings to the fore as well as the perspectives it opens for a comparative and social history of genetic counselling.

Keywords Eugenics • Genetic counselling • Molecularization • Pedigrees • Transnational history

Since this remarkable collection of assays on the history of genetic counselling and its developments in a wide range of national contexts has no chapter on the French case, it seems appropriate to begin my commentary with two quotes from papers the French paediatrician and first teacher of medical genetics in the country Maurice Lamy (1895–1975) published almost 20 years apart. Both papers consider the prevention of inherited diseases.

First:

> Once pathology has been identified as hereditary or once a given pathological pattern has been recognized as strongly influenced by hereditary factors, its mode of transmission must be specified. It so happens that many diseases are inherited in a rather simple way…. Reading pedigrees and statistical analyses thus provides us with highly valuable and immediately useful information…. The genetics of the future will therefore favour prophylaxis. Rational prevention can only be imagined if we learn how to identify from the mass the individuals affected with this hereditary fate…. Once the carriers of noxious

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> genes are identified, we shall know which marriages will possibly result in unfit progeny and should in consequence be advised against.[1]

Second:

> Progress in our knowledge of pathological heredity has or should have practical implications for prevention; I mean the need for organizing in France this genetic consultation, which is already operational in foreign countries where the least objectionable services are offered... Experience tells us that geneticists are asked for opinion and advice in four different situations. First when somebody, male or female, is affected with an abnormality and fears he or she might pass it on. More often, however, the request comes from a couple who has already experienced the birth of an unwelcome child. Occasionally, the parents of a would-be couple ask for advice. Finally, counselling is sought in relation to consanguinity issues.[2]

These two quotes aptly summarize the dramatic change of vocabulary and aims, which was at stake in the transition between what Lamy in 1951 called "eugenic counselling" and what he—and his colleagues at the French Academy of Medicine—labelled "genetic counselling" 20 years later. Eugenic counselling was about pedigrees, populations, hygiene, unfit progeny and advice against reproduction. Genetic counselling became a question of genes and metabolism, clinical encounter, patient's demands and hospital infrastructure. The replacement of the former by the latter was a matter of changing science, changing targets, changing organization and changing tools.

As we all know this transition from things deemed eugenic to practices called genetic is far from being a local phenomenon. It has actually become a repetitive motive of any paper historical or medical dealing with the coming of age of genetic counselling. Its specific features and generality in at least a few countries have been highlighted in the limited but significant historiography at hand.[3]

Historical studies of eugenics have thus insisted on national differences in the ways of problematizing the question of pathological inheritance and its social bearings and on the policies and targets of intervention based on this problematization. In contrast to the situation prevailing in Britain, eugenics in France was a medical issue.[4] One marker of this status is the fact that physicians, especially those specializing in reproductive medicine, i.e. obstetricians, gynaecologists and paediatricians, were very active in the local Eugenics Society (*Société française d'eugénique*). More importantly, the problems eugenics was to tackle were included in a broader hygiene nebula. Accordingly, the doctors participating in the French Eugenics Society were also engaged in societies fighting against tuberculosis, venereal diseases or population decline, leaving madness aside. They often considered, taking tuberculosis as a paradigmatic example, that

[1] Lamy 1951.

[2] Lamy 1970.

[3] Kevles 1985; Mazumdar 1992; Weindling 1989; Proctor 1988; Schneider 1990; Carol 1995; Gaudillière 2002.

[4] Carol 1995.

hereditary transmission of diseases was above all a question of constitution and predisposition, the influence of which on an individual's life could be controlled through changes in the familial and social environment. The concept of "*hérédo-contagion*" was widely used in articles and pamphlets, while "*sélection"* remained peripheral when present, which was rare. One major difference with the British or German eugenics complex was however the stamp, on the French movement, of a natalist perspective and natalist policies. After World War I, fighting depopulation and the declining birth rate became a dominant motto in social policies and social medicine, leading to a strong medical support of the 1920 law banning contraception and abortion. In contrast to the British movement in favour of birth control, the goal of the moment was to induce mothers to have more babies rather than to organize the selection of the lives to come by prohibiting the reproduction of the less fit. Quantity rather than quality of the population was what mattered.

The first decade after World War II was a time of reconstruction in all the sectors of the country's life. The health care system was significantly changed with the institution of a national health insurance system providing payment of medical care for all workers and their relatives. State intervention was increasingly seen as a normal mechanism to foster economic growth, as well as social reform when it was deemed necessary. As part of the trend towards state-based health planning, science became a target of national public investments with the establishment (or the reinforcement) of agencies such as the *Centre National de la Recherche Scientifique or the Institut National d'Hygiène*, which not only supported research in a few universities but, more importantly, set up their own laboratories.[5] At the crossroads of these two tendencies, science was increasingly viewed as a critical asset on the path towards medical progress. Medical research was however not necessarily synonymous of clinical, human and hospital-based investigations. Within a general framework that favoured national agencies and their laboratories, emphasis was placed on the need for "basic" knowledge. Scientific reconstruction privileged biomedicine and, to borrow the term coined by the virus specialists working at the Pasteur Institute, the "de-medicalization" of domains once considered as disciplines subordinated to clinical knowledge, such as biochemistry, bacteriology, virology, immunology, physiology, embryology and genetics.

One additional and important feature of the period was that from the late 1940s on, the new articulation between experimental biology and clinical practice was rooted in a process of "molecularization". Molecules were increasingly considered by biomedical researchers as the most important analysis units and intervention targets.[6] Escalating interest in macromolecules such as viruses, and later DNA, was typical of the molecularization of research objects. Correlatively, the first-order priority placed on the chemical designing of drugs and chemotherapy in general was typical of the molecularization of medical targets. This post-war "molecularization" impacted the study of familial and hereditary diseases in ways

[5]Picard 1992.

[6]Gaudillière 2002; Quirke 2008.

that can be discussed from two angles: (1) that of the new visibility acquired by inherited biochemical disorders and (2) that of the transformation in uses of pedigrees, their normalization along Mendelian lines and their integration into the new practice of genetic counselling.

As we know from the work of historians among whom D. Paul deserves a special mention, by the late 1960s, in the UK and in the USA, genetic counselling was firmly established as a medical procedure organizing the diagnosis and control of genetic disorders.[7] Its specificity was due to four features: (1) the idea that diagnosis was not necessarily the identification of an existing disease but the determination of a range of possibilities of various orders of importance that could be defined in quantitative terms with probabilities; (2) the use of family trees as a means to collect and circulate information about both the diagnosis and the prognosis; (3) the fact that genetic counselling was not a consultation focused on a single patient but on the medical handling of reproductive units, i.e. a couple or even entire families; and (4) in contrast with traditional clinical encounters, emphasis was placed on the separation between the objective analysis of the situation and the choice between existing options regarding reproductive life and medical intervention (sterilization, abortion or possibly a therapeutic course).

Medical genetics in post-war France developed along similar lines. At the Children's Hospital in Paris, the service of our founding character—Maurice Lamy—focused on the identification of "truly hereditary" diseases, meaning Mendelian genetic disorders. Differential diagnosis, mode of transmission and incidence were the main targets of investigations based on what can be called Mendelian pedigrees. The latter showed three or four generations of affected or non-affected individuals and provided the basis for aggregation, statistical calculus and a probabilistic interpretation of the transmission ratio. This is well illustrated by the local work on muscular dystrophy.[8] Children affected with this peculiar form of paralysis were encountered in local consultations for poliomyelitis. The final analysis published by the group was based on 77 pedigrees, the comparison of which had made it possible to disentangle "myopathy" into two different diseases defined on the basis of their mode of transmission. Muscular dystrophy was accordingly mingling a rare recessive autosomal disorder and a more frequent form of the disease linked to the X chromosome.[9] Pleading against the vagueness of all notions of hereditary transmission, not only did Lamy and his colleagues mobilize classical genetics concepts to fight the culture of transmission, they also adopted "inborn errors of metabolism" as a conceptual means to associate, just as Penrose had, Mendelian pedigrees and biochemical testing. At the Paris Children's Hospital, counselling did not exist as such in the 1950s, when hereditary disorder cases were seen and handled within different consultations. There were nonetheless some common features in the way clinical cases were managed. Lamy, for instance,

[7]Paul 1995 and 1998; Kevles 1985; Lindee 2005.

[8]Gaudillière 2002.

[9]de Grouchy 1953 and 1954.

supplemented the diagnosis of Mendelian diseases with advice to parents coming to the polio consultation, which provided the service with the large majority of muscular dystrophy patients included in the study on hereditary transmission.[10] At the same time, other members of the unit organized the diagnosis of PKU in newborns.

The nature and status of genetic counselling changed rapidly in the 1970s to become a routine, not to say mass practice. Sociological and historical studies focusing on the USA and UK have anchored this change in two series of events.[11] The first was the development of chromosomal analysis as a diagnostic tool to supplement and eventually replace clinical examination and biochemical analysis. Historians have taken the feminist movement of the 1960s and the successful mobilizations to abolish the legal ban on abortion in many developed countries as the second and most important root of the reinvention of genetic counselling. Before prenatal diagnosis, medical abortion had a problematic but significant role in the routine management of pregnancies. In France, the pro-natalist commitments of the interwar period had left an opening for abortion, i.e. when the life of the mother was at stake. In the 1950s, indications for a "therapeutic abortion" were gradually extended to conditions bearing the certainty of major abnormalities in the newborn. Cases of pregnant women contracting rubella provided the first publicly discussed condition, as rubella dramatically increased the risk of deafness and blindness. However, it is very likely that from the late 1950s on, hospital services like those of the Paris Children's Hospital informally performed therapeutic abortion for other conditions, including haemophilia and muscular dystrophy.[12]

My aim in reminding this story is not to add "one more case study" to the rich collection assembled in this volume, which add Sweden, Czechoslovakia, Austria, Belgium, the German Democratic Republic, the German Federal Republic, Greece or Mexico to the classical three (USA, UK and France). It is rather to make more visible the tension underlying most papers in this series, namely the tension between the shared scenario originating in the historiography of post-war medical genetics in the UK and USA and the temptation of reading variations from this scenario as national differences of social, political or cultural origins.

In a very crude and succinct form, the shared scenario approaches the path to genetic counselling as a consequence of scientific and technical innovation, which in turn led to new medical practices and representations, to finally meet changing social demands, thus resulting in institutionalization of a new medical specialty and—eventually—specific public health policies. The reference path thus consists in three periods: (1) the 1940s–1960s with the transition from eugenics to medical genetics, from population management to clinical handling; (2) the 1970s–1980s associated with the development of new technologies (routine karyotyping, amniocentesis, ultrasound imaging) and a process of professionalization and

[10] J. Frézal, interview with the author.

[11] Kolker 1994; Rapp 2000; Schwartz Cowan 2008.

[12] J. Boué, interview with the author.

institutionalization of counselling; and (3) the 1990s and after associated with the development of molecular techniques for studying genes and a process of "biomedicalization" focusing less on the relationship between the laboratory and the clinic than on new patients' roles, including critique and counter-expertise, risk assessment and management, mounting economic pressure on social expenditures in general and medical costs in peculiar.

The first feature to be highlighted is that this collection provides a significant number of confirmations and thus reinforces the shared scenario in spite of significant national variations. One common feature to the chapters focusing on national configurations is accordingly to reassess the way out of eugenics. This was not an abrupt and radical departure but a matter of two decades paved with significant continuities in terms of personnel, tools (for instance the use of pedigrees), issues and vocabulary. Placing the interwar eugenics initiatives at arms' length of course took different forms according to the type of eugenics movements that had emerged in each country and the specific policies they sought to implement. One may for instance oppose the German uneasy confrontation to national socialist laws with the rather smooth transition that took place in France where "pro-natalist" policies and concerns for the familial transmission of major infectious disorders remained high after 1945.

Medicalization however seems to provide for a common denominator. The term should not be understood in its classical meaning, i.e. the medical framing and medical handling of social problems, but as a mere realignment of pathological heredity management with the clinic, which consisted in (1) an increasing involvement of medical (first of all paediatricians) and paramedical (the emerging group of counsellors) personnel; (2) the opening of a new clinical space (the counselling session); and (3) new links with biomedical laboratories (first with the growing interest in inborn error of metabolism, later with chromosomal disorders).

However powerful, this notion of medicalization does not provide all responses to the transition problem. One useful category—although not directly discussed in the papers—may be "governmentality", which was introduced in Michel Foucault's late work to single out forms of biopolitics, which focus less on disciplinary control and bodily coercion and more on the normalization of conducts. Most chapters thus document the mounting importance the proponents of genetic counselling have placed on the dialogue with couples, on the alliance of personal choice and autonomy and on the idea of risk evaluation and risk taking. These features all fit into a liberal framework, which balances the collective burden of diseases (public health aims) with individual benefits and freedom through the acceptance of norms, including that of responsibility towards future generations and "lives to be".

A second feature emerging out of the collection and worth discussing relates to the second stage in the scenario that is the 1970s turning point and its technological roots, i.e. the package of biochemical testing, karyotyping, amniocentesis and ultrasound imaging that proved central to the extension of post- and prenatal diagnosis of genetic disorders. Among these, karyotyping occupies a central place, maybe because it has been thoroughly investigated and provides "the" obvious link to the "new" post-war genetics meaning a laboratory-centred practice

inscribing disease within the macromolecular entities (DNA and chromosomes) of molecular biology. The diffusion of karyotyping thus exemplifies the advent of true molecular "lesions" beyond the biochemical "errors" that made the visibility of PKU in the previous decades. The mechanism of its diffusion however remains poorly understood beyond the obvious facts that procedures somehow circulated and became the province of growing collectives of geneticists/heredity experts who campaigned for policies changes and backed genetic counselling as form of care and new setting.

The most visible lesson, which can be taken from the set of chapters dealing with the 1970s, is however one of technological under-determination since the social context of institutionalization appears paramount in both the similarities and variations across national borders. Unsurprisingly changes in the status of abortion come to the fore, thus linking the history of genetic counselling with the women mobilizations of the 1970s and their differentiated relationship to health and medicine. Here, an important distinction needs to be made. It is not that the legal changes associated with the passage of laws de facto recognizing women's right to "free" abortion were mandatory to the spread of genetic counselling. Even in places where such change did not occur (as in most countries of Latin America), medical geneticists managed to further the linkage between prenatal diagnosis with special clause turning abortion into a restricted but effective clinical intervention, be it in the name of mother's health, clinical exception or (more rarely) prevention and public health. One should therefore invert the classical chronology of abortion debates and genetic diagnosis and consider that in some instances at least (France, Germany and maybe Sweden), the biomedical legitimacy of such "therapeutic" abortions actually paved the way to the acceptance of "free" abortion, at least in medical circles.

One last common thread regarding the shared scenario is the rise of debates on diseases and identities, which emerged in countries like Germany or the USA in the 1980s–1990s. These debates brought to the fore the fundamental tension underlying the liberal vision of genetic counselling, namely the fact that in spite of its individual, choice-oriented ethos the practice has collective effects and may foster powerful normative choices regarding the boundary between the normal and the pathological, acceptable and non-acceptable disabilities. Down syndrome is in this respect probably the most visible and documented case. In spite of the deep variability of prenatal testing practices across countries, the fact is that terminations of pregnancy following a positive diagnosis have become "normal" if not consensual outcomes. Interestingly, although critical views often link this situation with concerns regarding the costs of care and the sustainability of health system financing, the case of France where more than 90% of prenatal diagnosis end this way shows that the norm has emerged without any connection to public policies and financial reforms. The issue thus arising is therefore not a question of power in the classical meaning of the term but a question of norms and (bio)sociality. In other words, professional or administrative regulation of genetic counselling does not "impose" abortion as most favoured solution, but it emerges as the evident choice

when care options are rare and poor, barely accessible and little known, thus placing most of the burden on families.

Comparing this wide range of national histories of genetic counselling does not only provide for a reinforced periodization of its advent or a common analytical framework stressing the processes of biomedicalization. The collection as well points to new direction of inquiry among which the question of knowledge is especially challenging.

What emerges is less the need for an exploration of the changing corpus of theories and concepts mobilized in genetic counselling, be it the classical and problematic relationship between genotype and phenotype, or the more recent boundary categories introduced by genomic research like the mutations of unknown function but statistically associated with constitutional disorders. It is rather to approach the "science" of genetic counselling as practical and social construction, as collective culture emerging out of the shared and differentiated scenario mentioned above. The history of genetic counselling for instance appears as a history of multidisciplinary knowledge with geneticists, biochemists, epidemiologists, anthropologists, public health specialists and clinicians of heterogeneous specialties (from paediatrics to gynaecology) involved and interacting in various ways according to time and place. These complex networks, their institutional counterparts and their effects on the conduct of genetic counselling deserve further inquiries.

A second and even more challenging dimension is the changing fabric of evidence, what L. Fleck once called the making of (test as) scientific fact. Just like bacteriological diagnosis, genetic counselling is a laboratory practice translated into clinical and social medicine. The development of a new boundary tool like karyotyping thus entailed a difficult problem of regress. In order to stabilize the notion that Down syndrome is a trisomy 21 that can be diagnosed with karyotypes, one needed to stabilize *at the same time* the technique and the correlation between the clinical diagnoses on the one hand and the presence of three exemplars of chromosome 21 on the other. In the late 1950s and early 1960s, reports of clinical cases with normal chromosomal number or reports of trisomy 21 without clinical signs reveal how arduous such stabilization was. As Fleck told us when examining the making of the Wassermann serological diagnosis of syphilis, such processes require a specific community of experts, a period of tinkering and technical adaptation and strong cultural commitments. Such analysis of the collective and practical making of genetic counselling's basic facts, for instance, of the many critical correlations between congenital/constitutional disorders and the signs observed on ultrasounds images, remains to be done.

At a more general level, and to bring this comment to a conclusion, Fleck's insistence on the role of representations and ideograms may be instrumental in following the changing arrangements of ideas, tools and social links underlying the historical trajectory of genetic counselling. One may—for instance—focus on pedigrees, on their permanent but diverse uses all along this trajectory. One may accordingly suggest that genetic counselling (in a broad sense in order to include

Table 1 Four types of pedigrees

	Eugenic pedigrees	Clinical pedigrees	Genetic pedigrees	Molecular pedigrees
Idea	Degeneration	Family illness	Transmission of Mendelian factors	Molecular mutation
Form of scientific Work	Social surveys, reports	Anatomic-pathological exams, nosologic classification	Clinical diagnosis, statistics, etiologic classification	Diagnosis, DNA analysis, computation (models)
Ideogram	Group family tree	Case family tree	Collection of transmission trees	Family tree with markers
Esoteric/ Exoteric circles	Eugenics societies, journals, exhibitions, political assemblies	Medical specialities, family, hospital consultation	Geneticists' societies, counsellors' associations, family–patient collectives, genetics consultation	Idem. + Biology laboratory, support group, organization
Articulation procedures and type of expertise	Administrative expertise, education and legislation	Professional expertise, biographic recording, care	Professional expertise, counselling and managing reproduction	Distributed but top-down expertise, risk management

the counselling activities backed by eugenics movements) successively mobilized four different types of pedigrees (Table 1):

The interest of such typology is less to propose a chronological order than to point to specific types of arrangements and thus to pave the way for a social history of genetic counselling, which would not tear apart the epistemic, the technical and the sociopolitical. The molecular pedigrees of the recent decades may thus be approached as ingredients of a putative new regime in medical genetics, a regime, which—as several papers in this series allude to—focuses on risk objectification and management, patients' (collective) participation and economic regulation through cost-effectiveness measurement.

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