# Introduction

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> 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.

## References

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