Foundation of the International Federation of Human Genetics Societies: The Catalyst

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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

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[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).

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[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

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[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]

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[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]

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[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*

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[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

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[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.

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[31] See EN 4.

[32] Aymé 2016; Personal correspondence to Marcus Pembrey, 26.07.2016.

[33] See EN 32.

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