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2010

## Tracing the Shifting Sands of 'Medical Genetics': What's in a Name?

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**Pre-Publication Draft**

**Tracing the Shifting Sands of ‘Medical Genetics’:  
What’s in a Name?**

*Abstract:* This paper focuses on the structural development of institution-based interest in genetics in Anglo-North American medicine after 1930 concomitantly with an analysis of the changes through which ideas about heredity and the hereditary transmission of diseases in families have passed. It maintains that the unfolding relationship between medicine and genetics can best be understood against the background of the shift in emphasis in conceptualisations of recurring patterns of disease in families from ‘biological relatedness’ to ‘related to chromosomes and genes.’ The paper begins with brief considerations of the historical confluences of, first, heredity and medicine and, second, genetics and medicine which, in a third section, leads to a discussion about a uniquely ‘genetics-based approach’ to medicine in the second half of the twentieth-century.

## 1. Introduction

The term ‘medical genetics’ first appears in Lancelot Hogben’s book ‘Genetic Principles in Medicine and Social Science’ (1931): ‘Whatever views one may entertain concerning the urgency of social policies based on genetic assumptions, the urgency of promptitude in developing the machinery of research in medical genetics should not be overlooked by any who have the advancement of pure science at heart.’<sup>1</sup> The term does not appear again in the book, but I feel confident in saying that ‘Genetic Principles in Medicine and Social Science’ was the source of inspiration for Madge Thurlow Macklin’s writings on the subject of medical genetics.<sup>2</sup> Whilst Macklin neither cites nor references Hogben in her work, she clearly shared a perceived need to, as Hogben put it, ‘infiltrate the curriculum of clinical studies’ with instruction in human genetics.<sup>3</sup> Moreover, Hogben’s enthusiasm for promoting genetics as ‘an exact science’<sup>4</sup> and the future potential of ‘chromosome maps’<sup>5</sup> both appear as themes in Macklin’s work.

It is fair to say that Macklin was much more tenacious and proactive than Hogben in her campaigns for human genetics instruction in medicine.<sup>6</sup> At the Third International Eugenics Congress held during August 1932, she declared:

I feel very much like Ulysses when he was steering between Scylla and Charybdis, for on the one hand there is the non-medical advocate of eugenics who may resent my emphasis upon the medical practitioner as the pivotal point in the eugenic programme, and on the other there is the medical practitioner who may object to my suggestion that he needs more education upon the subject of heredity as applied to medicine. My course, though difficult, is nevertheless clear.<sup>7</sup>

Genetics, she asserted, is ‘... a young science, and that genetics applied to medicine is a mere infant. But it is a very lusty one and will keep on crying until it is heard.’<sup>8</sup>

Macklin went on to write a series of articles on the subject which culminated in 1933 with a sample syllabus for a course in medical genetics. In addition, she produced a massive review article on the role heredity plays in clinical phenomena in which she provides an impressive list of 200 heritable diseases and discusses twin studies, consanguineous marriage, family pedigrees, and statistical techniques.<sup>9</sup> Macklin reasoned that attending to the hereditary components of diagnosis made possible early detection, diagnosis, and commencement of therapies, and was therefore important in terms of preventive medicine. Further to this, being a strong supporter of eugenics, she believed that ‘the triumphs of modern preventive medicine’ had served to ‘throw into stronger relief the problems of human inheritance;’

... persons spared from death by infection are kept alive to succumb to their constitutional disorders, so that we find the death rate from many of the degenerative disorders of the circulatory system, from cancer and from diabetes, rising.<sup>10</sup>

These ideas all provided grounds for supporting Hogben’s entreaty for scientific investigations of the physical basis of inheritance and for teamwork ‘on a very large scale’ involving the collaboration of geneticists, clinicians, and ethnologists to ‘assess the relative importance of nature and nurture in a specified range of conditions’ including ‘such physical characteristics as growth limits and resistance to disease.’<sup>11</sup> That being said, the contemporary historian Daniel Kevles has shown that fewer than two hundred people published any research in the early Anglo-North American contingent of human geneticists prior to the Second World War.<sup>12</sup> Of these, fewer than fifty published more than once. The situation changed noticeably after the Second World War.

Formal positions for human geneticists had been created in thirty-one centres in the United States (twenty-five), Canada (four), and England (two) by the end of the 1950s.<sup>13</sup> Comparatively speaking, five surveys, completed over a period of three decades, show that the proportion of North American medical schools with formal courses in genetics increased from 8.6 per cent in 1953 to 86.5 per cent in 1985.<sup>14</sup> Correspondingly, across the Atlantic, the membership lists of the Genetical Society of Great Britain show a sharp increase in members involved in medical research in the UK after 1959, rising steeply to 1969 when nearly 12 per cent of the 900 members of the Society were working in medicine.<sup>15</sup> The eventual growth and recognition of medical genetics as a service specialism in the UK and North America that occurred after 1970 came about largely as a result of technological innovations in the form of, first, the new laboratory technologies for identifying chromosomal anomalies and genetic metabolic disease, and, second, the advent of regional newborn screening programs and increased use of amniocentesis in prenatal diagnosis. In all of these countries, the intellectual and specialist movements that supported this growth were emergent phenomena, created, split, and reattached to different groups of actors, and reconfigured at least twice over the next four decades. In each instance, new kinds of working relationships appeared; sets of diverse actors in university-hospital settings coalesced into a new collectivity; and, as a collectivity, actors defined and/or redefined occupational roles and work rules. In the first instance, an elite of PhD- and MD-geneticists built career paths through their work in newly established clinical settings for heredity counselling. These individuals established specialised work patterns by combining hospital work and teaching posts. Furthermore, they drew a

clientele of patients on the basis of personal reputations for specialised expertise. In the second instance, counselling and laboratory services became standardised and specialised occupational roles and work rules for clinical and laboratory services were established. In the transitory movement from medical segment to medical specialty, the ideological direction of clinical practices conformed to a pattern widely adopted among contemporary medical specialties. As a result, a formal job classification – medical geneticist – became viable as a full-time occupation in medicine in the UK and North America.

This paper focuses on the structural development of institution-based interest in genetics in Anglo-North American medicine after 1930 concomitantly with an analysis of the changes through which ideas about heredity and the hereditary transmission of diseases in families have passed. Taking into consideration the lag between theoretical and therapeutic capability in the application of new scientific knowledge, I argue that the unfolding relationship between medicine and genetics can best be understood against the background of the shift in emphasis in conceptualisations of recurring patterns of disease in families from ‘biological relatedness’ to ‘related to chromosomes and genes.’ I understand the shift in emphasis to represent a bringing together of the organisational ideas of key innovators in science and medicine and I explore the corresponding characteristics of the institutions they built. I do not claim that the changes in conceptualisations represent average ‘medical thinking’ at the time; they did not. A minority built academic specialty was formed intending to train a new generation of medical specialists in order to reform clinical practice. Accordingly, a key aim of this

paper is intended to permit identification of what objects, questions, concepts, methods, and research are properly considered medical and the institutional steps through which a ‘genetics-based approach’ to medicine became distinguishable and duly recognised.

## **2. The historical confluences of heredity and medicine**

Historians of medicine studying the topic of heredity normally posit an early or pre-modern period in which stories were collected about so-called monstrous births in the naturalist tradition of sixteenth-century Europe.<sup>16</sup> What is most noteworthy here, for the purposes of the present study, is the movement from the singularity of legends, anecdotes, and story-telling to the generality of systems of taxonomy supported by case studies that were published and archived.<sup>17</sup> Case studies of *morbid haereditarii* (heritable disease) recount a range and categories of physical/developmental forms as well as biographical aspects of illness episodes or narratives in time. As particular ‘clues’ and ‘symptoms’ took on special roles and significance (e.g., missing or supernumerary limbs, birth marks diminished stature) case studies took on emblematic status.<sup>18</sup> At the same time, the notion of the ‘familial taint’ lent typicality to the case at hand. The physician became ‘a chronicler of bodily events and systematic narrator of particular phenomena in a particular context.’<sup>19</sup> Case studies in turn supported natural history and the seeking out of nosological categories for classifications of disease.

Historians Laure Cartron and Carlos López-Beltrán suggest that physicians and physiologists in eighteenth-century France were the first to use the word heredity (*hérédité*) to refer to the phenomenology of peculiar character or trait transmission from parents to offspring.<sup>20</sup> Cartron specifically links the ideas of hereditary disposition to

disease, predisposing diathesis (i.e., an acquired susceptibility of the body to disease), and constitutional weakness (i.e., inherent weakness in the physical make-up of a person).<sup>21</sup> This supplements earlier work by Erwin Ackerknecht who showed that the notion of diathesis gained popularity in medical circles at the end of the eighteenth century and ‘constitution studies’ flourished in the United States, France, and Germany by the 1920s.<sup>22</sup> At the same time, Ackerknecht indicates interest in these topics was waning and being ‘outdistanced by bacteriology, endocrinology, serology, vitaminology, the neurology of the vegetative system, or genetics’ by the 1940s. Indeed, Antonio Ciocco, in his 1936 paper on the ‘modern study of constitution,’ complained about the apparent growing disregard of the ‘so-called genetic school of constitutionalists’ for the field constitutional somatology.<sup>23</sup> As late as 1943, George Draper, an associate professor of clinical medicine at Columbia University, sought support for constitutional medicine and recognition of the biological significance of the human constitution from the standpoint of a ‘school of biological thought known as “organicist” or “organical,” as opposed to the [school of] elementalist or particulate [thought of geneticists].’<sup>24</sup> Draper claimed that the modern physician ‘labours in the anthropological division of biology’ and that although the basic sciences of physics and chemistry had provided explanations of certain physiological phenomena found in the clinic, there still remained a number of vital phenomena in need of explanation ‘related to the whole organism’ including ‘the phenomena of heredity, growth, and development.’<sup>25</sup> Likewise, Julius Bauer, then professor of clinical medicine at College of Medical Evangelists, Los Angeles, observed:

It is taken for granted – usually without particular amazement – that the potential energy of the germ plasm accounts for the development of a full-fledged human

being out of a fertilized human ovum of microscopic proportions in the short period of nine months. It is not adequately realized, however, that this potential energy is at work throughout a whole lifetime; that the physiologic evolution and involution, the structure and function of the organs, their mutual relation, and the response to various stimuli depend on this potential energy; that deficiencies and abnormalities of this potential energy may be cooperative etiologic factors or the sole cause of diseases; that they account for the vast majority of congenital malformations in man. And this potential energy is just what we may call individual constitution. The great miracle of this potential energy is fully recognized as far as the fetal development is concerned, but the miracle is not over with the moment of birth.<sup>26</sup>

Bauer had emigrated via France to the United States after the German annexation of Austria in 1938.<sup>27</sup> Bauer hoped that constitutional medicine could be connected to current work being done in the United States with breeding and experimental genetics.<sup>28</sup>

At the same time, he acknowledged a certain weakness in such an idea:

One point ... should be stressed: constitution is not a physical entity but a panel and plan of the total set-up of the individual personality. Constitution comprises physical and mental traits, visible and invisible characteristics, which may or may not be detectable by all or any of the various methods of physical examination and laboratory work. Constitution is therefore not a measurable entity; but some of its components may be defined in exact figures.<sup>29</sup>

The weakness was not lost on critics of constitutional medicine.

A reviewer of the book, 'Human Constitution in Clinical Medicine' (1944), described what he called a 'subtle defeatism' in constitutional medicine that viewed disease as an 'inevitable product' in certain susceptible individuals' and an 'undercurrent of therapeutic pessimism.'<sup>30</sup> This, he argued, represented an 'anachronism' in modern medicine. While interest in the human constitution was subsequently taken up and advanced by some psychologists and physical anthropologists interested in constitutional somatotyping, genetics came to occupy an increasingly important position in the clinical study of heredity as a result of a scientific line of inquiry that confined investigations to a

few specific problems associated with the transmission of physical characteristics between generations.<sup>31</sup> More specifically, Mendelian research on aspects of factorial transmission, sexual reproduction, and the production of physical variation in organisms served to limit the scope of biological inquiry and departed significantly from earlier programmes of research to explain the links between heredity, embryonic development and evolution.<sup>32</sup>

### **3. The historical confluences of genetics and medicine**

Archibald Garrod's work on 'inborn errors of metabolism' is a likely place to look for early connections between genetics and medicine.<sup>33</sup> Garrod had developed an interest in chemical pathology while working as a physician on the staff of several hospitals in London in the late nineteenth- and early twentieth-centuries.<sup>34</sup> After being appointed as an assistant physician to the outpatients department of the Hospital for Sick Children in Great Ormond Street, he began to write about cases of rare disorders such as alkaptonuria and porphyrinuria which produce different colours in urine. Garrod's initial observations led him to focus on chemical errors in the metabolism which appear as congenital defects that persist throughout life.<sup>35</sup> He went on to interpret the recurrence patterns in families as following Mendel's law of recessive inheritance.<sup>36</sup> For this he received advice and the collegial approbation of William Bateson, an early proponent of Mendelian genetics and founder of the Genetical Society of Great Britain, and Frederick Gowland Hopkins, a chemist who, in 1894, graduated in medicine and taught for four years physiology and toxicology at Guy's Hospital before moving on to Cambridge University.<sup>37</sup> Still, although the links between defective enzymes, disruptions in metabolic pathways, and disease

were evident in the contemporary biochemical literature, biochemists were generally unfamiliar with the tenets of Mendel's laws. Equally, geneticists and physicians were unable to link the idea of a genetically-based enzyme deficiency to any concept of genes or of gene action.<sup>38</sup>

It is important to note that while Garrod was intrigued by the discovery of the metabolic disease phenylketonuria in the 1930s, the association between the disease and genetics was made by others.<sup>39</sup> Moreover, there is evidence that Garrod was more interested in the holistic line of inquiry found in constitutional studies. Pauline Mazumdar, for example, has found that his reconfiguration of the theory of diathesis under the notion of 'chemical individuality' was 'constructed within the guidelines set by the German clinicians who formed a group round Friedrich Martius, professor in Rostock, Wilhelm His, Friedrich Kraus, and Otto Lubarsch in Berlin, and Julius Bauer in Vienna.'<sup>40</sup> That being said, J. B. S. Haldane's research on the genetics of flower coloration of the 1920s and 1930s at the John Innes Institutions and Sewall Wright's studies of melanin in animal coats of the early 1940s were important to the geneticisation of Garrod's theories.<sup>41</sup> And, in the mid-1930s, Beadle and Ephrussi took Garrod's ideas further and, using grafting experiments, sketched the steps and genetics that lead to the eye color of *Drosophila melanogaster* (i.e., the fruit fly). This demonstrated the sequential chain of events in a normal metabolic chain and contributed significantly to Beadle and Tatum's 'one gene, one enzyme' hypothesis which stated that the function of a gene is to direct the formation of a particular enzyme. Garrod's term 'inborn errors of metabolism' was subsequently changed in the 1950s to 'metabolic disorders' and came to refer to what affects the ability

of a cell to perform biochemical reactions that involve the processing or transport of proteins (amino acids), carbohydrates (sugars and starches), or lipids (fatty acids). In sum, Haldane, Beadle, Ephrussi, and Tatum reconfigured Garrod's ideas in a manner that gradually informed a picture of how genetic information is converted to specified sequences of proteins with enzymatic function. Each of the steps involved in the problem-shift surrounding metabolic disorders was made within the context of the search for specific causes to chemical disturbances, and each additional step expanded the original clinical research program. This line of thinking, in turn, laid the conceptual groundwork for devising testing procedures and technologies for the detection and quantification of soluble metabolites in urine and blood after 1930.

A second connection between genetics and medicine can be seen to develop as increasing numbers of researchers working in the fields of morphology, embryology, and cytology set about looking for abnormalities in chromosomal structures in order to establish correspondences between structure and hereditary effect. As early as 1901, the pathologist J. George Adami had proposed a physical model for inheritance based on the 'fortuitous comingling' of chromatin during sexual reproduction.<sup>42</sup> Various particulate theories of heredity followed based on ideas about the relations between Mendelian 'factors' (later called genes) and characteristics and traits in living organisms. Hugo de Vries and William Bateson each had ideas about 'unit characters' that connected with 'factors.'<sup>43</sup> An attempt to use genetic theory to explain heredity in terms of chromosomal events also appears in W. F. R. Weldon's lectures on heredity at University College London as early as 1904.<sup>44</sup> But it was the work of Thomas Hunt Morgan and colleagues

at Columbia University that consolidated the chromosomal theory of heredity in Anglo-North American science after 1915 and encouraged ongoing research which sought to localise hereditary materials within the chromosome.<sup>45</sup>

The association of sex-determination with a chromosomal element had been suggested in the early 1890s with Hermann Henking and colleagues in Germany reporting an unpaired 'chromatin-element' that did not pair with the others during meiosis in the wasp *Pyrrhocoris*. Henking noted that the male wasps possess twenty-three chromosomes. Twenty-two of these were observed as active in eleven pairs during spermatogenesis; the twenty-third remained unpaired.<sup>46</sup> Moreover the behaviour of the 'odd' element was different with respect to movement during cell divisions. But Henking did not go on to identify this element as a chromosome. It was the American cytologist and palaeontologist Clarence E. McClung and his student, Walter S. Sutton, who observed similar uneven pairings in the grasshopper *Xiphidium* in 1899 and named the element the 'accessory chromosome.'<sup>47</sup> Sutton, following the work of the German cytologist Theodor Heinrich Boveri, went on to develop definitive arguments concerning the association and orderly behaviour of paternal and maternal chromosomes as constituting the physical basis of the Mendel's laws of inheritance.<sup>48</sup> The chromosome theory as expounded by Boveri and Sutton provided a framework for others who sought to localise hereditary elements in the nucleus of the cell. Briefly, Boveri and Sutton proposed that, first, chromosomes retain their individuality from one cell division to another; second, the developing egg obtains corresponding chromosome sets from egg and sperm nuclei; and, third, each of these parental sets, separately, is sufficient for

normal development of the ‘germ,’ i.e., hereditary essence. Given that these three hypotheses identify the chromosomes as carriers of hereditary substance, two divergent possibilities presented themselves. Each chromosome might contain the total hereditary substance or that each chromosome may be the carrier of different portions of the hereditary elements. In the latter case, each chromosome would be of different genetic value.

In 1911, the American embryologist Thomas Hunt Morgan, studying the determination of sex and the hereditary transmission of mutations in *Drosophila melanogaster*, coined the phrase ‘sex-limited inheritance’ and associated it with a specific chromosome.<sup>49</sup> This work led, in 1915, to the publication of *The Mechanism of Mendelian Heredity*, which Morgan coauthored with Alfred Henry Sturtevant, Hermann Joseph Muller, and Calvin Bridges. Importantly, when these authors turned to the chromosome as the epistemic object of study they did not attempt to subsume embryological concerns about the physiological capabilities of the egg or embryo under a particulate model. Rather, they segregated questions of development and the role of the cytoplasmic-organisational context in realizing the acquisition of hereditary characters.<sup>50</sup> This instigated a dichotomous view of the organism and what Ron Amundson has called ‘the cleavage between heredity and development.’<sup>51</sup> The concept of heredity consequently ‘narrowed’ and the interpretation of phenotypic events such as hereditary disease could then be pursued in relation to the study of a hypothetical entity called a gene. Morgan, in this context, managed to lay the conceptual foundation for ongoing research on the physical location of genes on chromosomes in which heredity appeared as

a background assumption of a formal system of genetic operations. He dubbed this research ‘transmission genetics’ which he distinguished as different from ‘developmental genetics’ which was to be pursued in relation to experimental embryological studies.<sup>52</sup> Alongside this conceptual foundation, the material object of the chromosome was embedded within associated laboratory practices in the fields of cytology and microscopy which would prove invaluable to laboratory medicine.<sup>53</sup>

Research on human chromosomes did not progress as quickly as work on relatively simple organisms like *Drosophila melanogaster* for reasons of serious technical problems in the preparation and examination of tissues under microscopic examination.<sup>54</sup> But by the mid-1930s, chromosomes could be recognised individually through their shapes and bands and, by the late-1930s, examination materials prepared with colchicines allowed researchers to spread out and tease the individual chromosomes apart.<sup>55</sup> The process was refined in the 1950s and the precise number of the human ‘karyotype’ was established as 46 chromosomes. Advances in karyotype analysis in the 1950s permitted some types of major chromosomal abnormalities, including missing or extra copies of a chromosome or gross breaks and rejoinings (i.e., translocations), to be detected by microscopic examination. And, by the late 1950s, the syndromes of Down, Turner, and Klinefelter were correctly karyotyped and women with triple-X identified, establishing a basis for what later became known as ‘cytogenetics’ in laboratory medicine. A standard system of nomenclature for human mitotic chromosomes in medicine followed in 1960.<sup>56</sup> The classificatory system, in turn, facilitated greater cooperation between laboratory

scientists in France, Sweden, Japan, North America, and the UK, as well as enhancing the diagnostic potential of cytogenetics in laboratory medicine.

It is important here to stress the novelty of human cytogenetics and biochemical analysis during the postwar period. Chromosomal and biochemical analyses provided the first clinical tools to uncover the genetical element of certain rare and common heritable disorders. At the same time, it is important to understand that the impact of these innovations on clinical practice evolved slowly, with disparity and divisions in how the individuals involved thought about themselves and the ways they characterised their activities.

#### **4. A genetics-based approach to medicine**

A 1954 Association of American Medical Colleges report represents the first concerted effort to produce a blueprint for the future of genetics in medicine.<sup>57</sup> On a high level of generality, its discussions of an ‘integrated curriculum’ and a role for geneticists in medical schools remain faithful to what Lancelot Hogben and Madge Thurlow Macklin had originally proposed in the 1930s. First of all, methods involving the identification of hereditary factors in disease are described in the report as supplementing the practices of ‘any [medical] specialty that can be named’.<sup>58</sup> Second, and concurrently, ‘genetics ... serves as a useful tool in the prevention of disease and limitation of disability’.<sup>59</sup> Turning to the question of *what to teach*, the report recommends five fundamental areas of study for consideration: the physical basis for heredity; the basic single gene mechanisms; interaction between heredity and environment; mutation and its evolutionary significance; knowledge of population genetics as well as instruction in the practice of

genetic counselling. The authors suggest that these fundamentals can be covered adequately in twelve to fourteen hours of lectures, ‘if supplemented by a moderate amount of reading in available texts’ – less than half what Macklin had envisioned as a thirty-six hour lecture course.<sup>60</sup>

Turning to the question of *who should teach*, participants in the workshop generally agreed upon ‘a trained medical geneticist on the staff.’

He might be either an M.D. with special training in genetics, or a Ph.D. in human genetics with added training in the special applications of his subject to medical problems. Such a person could be attached to any department in accord with administrative convenience. He could function in several ways: he might teach the course or give the lectures on general principles in the second year. He could be used for integrated teaching in many areas, as for example in the problems of maternal-fetal immunologic incompatibilities, in connection with the teaching of metabolic diseases, anaemia, bone and eye diseases and in other areas.<sup>61</sup>

The medical geneticist ‘could also have service and research functions.’<sup>62</sup> Thus, a multi-faceted role was envisioned. As ‘staff geneticist’ in a teaching hospital setting would provide advisory services, on the one hand, in family counselling directly with consultants, and, on the other, to practitioners and researchers requiring consultation in cases involving complex genetic problems.

Importantly, there was a wholesale shift in the way genetic counselling was to be conducted and certain irreconcilable differences identified between the ameliorism of the eugenic ‘heredity consciousness’ and ‘preventive ideas of medicine’ advocated by Hogben and Macklin and what was being presented as value-free science in the postwar period.<sup>63</sup> This shift would find its expression most forcefully in what Sheldon C. Reed, director of the Dight Institute of Human Genetics, called ‘non-directive genetic counseling;’ a procedure intended to explain to patients ‘what the genetic situation is ...

but the decision must be a personal one between the husband and wife, and theirs alone.<sup>64</sup> Reed explained:

Counseling in medical genetics is a most important practical application of the findings of the science of human genetics. It could help almost every family if available to them. We are still in the beginning stages of the development of sound practices in genetic counselling. As long as we do not take ourselves too seriously but instead approach the problems in a light-hearted manner, there will be no danger of the gory excesses committed in the name of eugenics in the past.

...

... It is the physician who is likely to have a hand in shaping the future evolution of mankind because our reproduction is no longer completely capricious. The desire for a happy family of normal children is one of the strongest human motivations. For the first time in history the physician is able to be of major assistance in achieving the highest of life's goals. If a couple finds that the family is increasing beyond their fondest expectations, he can slow down the flood. In civilized countries responsible parents no longer leave reproduction to the vagaries of chance. The physician officiates at the birth of the child and is the authoritative source of scientific information about reproduction.<sup>65</sup>

In this context, non-directive genetic counselling, in effect, was to become a contract for services and the essence of the physician-patient relationship would be transformed from one of status to one of contract.<sup>66</sup>

The recommendations of the 1954 AAMC report enjoyed considerable support and further discussion in subsequent surveys and reviews. Three surveys indicate that, rather than an integrated curriculum, genetics instruction increasingly became the preserve of paediatricians and geneticists teaching in independent genetics departments.<sup>67</sup> This issue went on to be described in the surveys and reviews as an interdisciplinary 'problem;' a problem frustrating the larger goal 'that "genetically thinking" becomes an integral part of [all] medical practice.'<sup>68</sup> From this emerged what I have called elsewhere a bifurcated ideological construct<sup>69</sup> that shaped and informed the means of organising a 'genetics-based approach' to medicine.<sup>70</sup> The construct stipulated, on the one hand, that

the mandate of medical genetics was to add a new set of medical procedures to the clinical repertoire of *all* individuals trained as physicians. On the other hand, it indicated that when and where physicians were unable to provide the new procedures, a class of *specialists* (i.e., medical geneticists) would be available for consultation. Accordingly, Vincent M. Riccardi, Baylor College of Medicine, stressed:

... it is incumbent on clinicians in all health care disciplines to recognize when health impairment is due, in part or in whole, to a genetic cause. All clinicians must be able to determine whether a given disorder is genetic, possibly genetic, or not genetic, and be able to share that information with the patient or family and refer them, if necessary, to specialists for further assistance. ...

*Ensuring that a family with a genetic disorder receives genetic counselling is a primary care responsibility, even though the actual genetic counselling may be carried out by a specialist in a secondary or tertiary care facility.*<sup>71</sup> (*Emphasis in the original*)

It is certainly the case that local associative strategies among medical professionals surrounding specialty formation and the institutionalisation of genetics-based diagnostic and laboratory services initially emphasised, on the one hand, multidisciplinary task specialisation among service providers working in regional centres and, on the other, more or less continually unfolding internal differentiation among academic health centres, community clinics, consumer and voluntary groups, government agencies, and public health departments. The patterned and the temporal organisation of contemporary genetic health services in the UK and North America nevertheless reflects different levels of interplay between autonomous action and constraining structure in local settings. On the one hand, geneticists entering clinical settings learned to conduct themselves in accordance with the same proscriptions that stem from the diverse histories of service relationships at the particular institutions employing them. Still, geneticists

also maintain that, *as medical specialists*, they provided services in a manner that is similar, if not the same, as others *in their specialism* around the world. Accordingly, it is important, from an historical standpoint, to distinguish between what are here idealtypically the features of medical genetics *in toto* and factors affecting the way that specialty services have been delivered locally.

The addition of the new laboratory services for identifying chromosomal anomalies and genetic metabolic disease described in the previous section of this paper contributed significantly to the growth of counselling clinics which, during the 1950s, had developed in an *ad hoc* and piecemeal fashion.<sup>72</sup> These technological innovations combined with the organisation of regional newborn screening programs in the 1960s and increased use of amniocentesis in prenatal diagnosis in the 1970s meant that genetics consultants experienced an increase in demand for services and, consequently, an increase in workload. As a result, concerted measures were taken to not only secure more resources and expansion for existing services, but to elevate the level and quality of services across, on the one hand, provincial and state genetics centres in Canada and the United States and, on the other, National Health Service regions in the UK.

With the increase in demand for genetic services, one can see the sequence of associative movements, segmentalisation, and other aspects inherent in the process of specialty formation typically found in sociological accounts of medical specialties.<sup>73</sup> In all cases, geneticists offering counselling and laboratory services found themselves spending proportionally less time in academia and research, and more time worrying about what was going on in the clinics. More pointedly, geneticists who provided

counselling and laboratory services in the late 1960s became more self-conscious about their patterns of work in professional medicine. Increasingly, two key questions were raised in professional circles: How does medical genetics fit into the delivery of patient care? How do geneticists *as medical specialists* incorporate *other* specialists and *non-medical* personnel into their own schemes of work and aspiration? Both of these questions, in time, found expression in an assortment of concrete efforts to establish some kind of formal mechanism to create and maintain standards for the delivery of genetic services at the national level.

Canada was among the first nations to develop standards of service and training overseen and administered by a body of medical professionals: The Canadian College of Medical Geneticists.<sup>74</sup> The College, established in 1976, preceded similar developments in the Netherlands, the United States, Finland, Sweden, Germany, France, and Denmark. The Canadian case shows how occupational specialisation in the broader field of genetics and medicine underwent remarkable divarication in a relatively short period of time. Moreover, it shows that the role of the geneticist in medicine evolved to a point where interchangeability between clinical- and laboratory-based functions abated.

Medical genetics in the UK took a more circuitous route. Broadly speaking, the formal structure for the National Health Service 'integrated genetics service' fit well within the regimes of service delivery instituted between 1974 and 1982 when the NHS was reorganised to integrate hospital, community health care and family practitioner services under a unified management structure. By 1982, genetic laboratory services and counselling clinics were to be found in nineteen NHS regions across the UK.<sup>75</sup> The

regional genetic centre became the hallmark of British medical genetics, holding regular clinics in the centre and also satellite clinics to which clinicians would be dispatched to see patients in District General Hospitals. A decade later, responsibility for genetic services would devolve from regional administration to conurbations of districts.

The notion of the regional genetics centre that emerged from the 1970s and 1980s combined ideas about the health needs of populations with an omnibus 'genetic approach' to health and illness. In its simplest geographical aspect, regional services consisted of two generalised unit parts: the centre and the adjoining catchment area. The two developed together, each presupposing the other. But while the centre was compact and readily visible, the catchment area was diffuse and difficult of precise observation. The boundaries of regional genetic services in fact appeared in varying degrees of distinctness at the local level according to the repertoire of policy instruments available, the preferences of the dominant technocratic elites, and the position and power of local bureaucracies to control funding and other kinds of resources. Genetic services, in this context, represent a series of concentric zones around service centres which differ in the degree of attachment of their occupants to the centres, of the frequency of movement of patients or patient information to and from the centres, and in the extent to which contacts with the centres are, on the one hand, direct, involving the movement of individuals, or, on the other, indirect, involving a circulation of information and specimens rather than people.

With regard to the division of labour in the delivery of services in regional genetics centres in both the UK and North America, two broad sets of activities can be

discerned that involved the geneticist in the capacity of consultant. The first set falls under a general category of prenatal care in pregnancy and childbirth, and overlaps with the jurisdictional claims of obstetrics and gynaecology. This set of activities can be distinguished from ‘general genetics,’ which is a catch-all category for clinical activities involving infants, children and adults. As a set of activities unto itself, it can be further divided into three sub-sets. Activities in the first sub-set overlap with the jurisdictional claims of neonatology. This involves the diagnosis and management of congenital anomalies and diseases in newborns. The second sub-set takes up broader paediatric concerns and focuses on the diagnosis and management of genetic conditions in children. Finally, the third sub-set deals with, on the one hand, the diagnosis of adult-onset diseases and, on the other, screening for carriers of heritable conditions. In this regard, the character of the jurisdictional interface with *other* specialists (i.e., *non-geneticists*) shifts paradigmatically depending on whether the patient is a pregnant woman, an infant, a child, or an adult.

It is especially noteworthy that open competition between clinical geneticists and other specialists for jurisdiction over particular kinds of patients and types of disease has gone virtually unheard of.<sup>76</sup> By the turn of the century, clinical interest in applied human genetics expanded to embrace a wider set of service relations as progressively more physicians and allied health personnel from specialty areas *other than medical genetics* either took over the diagnosis and management of patients in independent clinics or worked jointly with genetic service providers in hybridised clinical units for specific illness groups characterised as ‘complex,’ ‘multifactorial’ or ‘polygenic.’ This would

normally be seen from historical and sociological viewpoints as evidence of competing specialist segments and jurisdictional disputes in medicine, i.e., as an encroachment on the medical geneticists' claim to professional jurisdiction over a genetics-based approach to medicine.<sup>77</sup> Correspondingly, a late twentieth-century analysis of primary medical specialties might expect an ideological shift on the part of medical genetics in the direction of *de-specialisation* in order to accommodate the expanding remit of the genetics-based approach in medicine and changes in the administration of health services marked by deregulation and a transition away from centralised styles of governance.<sup>78</sup> But service providers in genetic health services *themselves* have not seen the situation in this way. Generally speaking, they see *other* specialists as coming around to their way of thinking.<sup>79</sup>

Interjurisdictional relationships between specialists have been negotiated in a manner reminiscent of what the American sociologist Anselm Strauss called 'silent bargains' and 'implicit negotiation.'<sup>80</sup> Strauss made a distinction here between 'transactions openly carried out between parties who recognise their own negotiating' and negotiations that are 'implicit, their products being tacit agreements or understandings;'

... the main issue is ... that actions are being taken with respect to non-negotiated limits imposed or signalled by one side and agreed to directly by the other. These kinds of silent bargains, then, would seem to pertain to agreements that are not much brought into explicit discussion and that represent limits *within* which negotiation can go on. Sometimes .... they go on in *support* of the limits, or they temporarily *stretch* the limits.<sup>81</sup> (Emphasis in original)

An argument follows that goes something like this: In the first place, the jurisdictional interface between medical genetics and other specialties has been

reinforced and strengthened by innovations and routines in applied human genetics. Networked innovation and the circulation of standards of laboratory and clinical practice (i.e., the genetics-based approach to medicine) can be seen to have diffused horizontally across a large number of specialty areas, as its advantages apply in different clinical settings and service activities. As awareness of the genetics-based approach to medicine increases, collegial recognition of the expert role of the geneticist among medical specialties is reinforced and fortified. Medical genetics is therefore inclusive as opposed to exclusive in the occupational hub culture of medicine.

In both the UK and North America, diagnostic tests and counselling for numerous illness groups became universally available in the 1980s through networks of regional genetic health services, although the availability of the full range of tests varied from centre-to-centre. All centres had access to laboratories for chromosome analysis (i.e., cytogenetics) and biochemical analysis for metabolic disorders. The delivery of laboratory services in genetics centres fell under the budget of a laboratory services department administered by pathology departments or divisions of medical genetics. In the case of infrequent testing for rare diseases, the costs would be absorbed into the budgets of researchers studying the specific conditions in question populations. Payment for the counselling services, however, was different owing to the vagaries of the provincial fee schedules in Canada and health market costings in the United States. Under the NHS, steps were taken to centralise six service components – genetic assessment, clinical diagnosis, cytogenetic analysis, biochemical diagnosis, prenatal genetic diagnosis, and case management involving genetic counselling – to be funded with global budgets

with the understanding that a portion of funding be allocated to satellite clinic programming.

It was the introduction of DNA testing regimes in the mid-1980s that sets into relief major differences in the UK and North America with respect to the position and power of local bureaucracies to influence growth in medical genetics. Whilst I have already commented in the previous section of this paper on the importance of advancements in biochemical and chromosomal analyses for specialty formation, I have not thus far said anything about the so-called ‘molecular revolution.’ Use of the term ‘new genetics’ in the medico-scientific literature appeared sometime around 1979 with reports and comments in medical journals concerning applications of recombinant DNA techniques, and new approaches to mapping and determining the fine structure of the human genome.<sup>82</sup> The terms ‘molecular genetics’ and ‘molecular geneticist’ appeared soon thereafter.<sup>83</sup>

Early acknowledgement of the clinical potential for DNA testing techniques by British geneticists is evident in two reports issued by two working parties of the Clinical Genetics Society in the early 1980s<sup>84</sup> and in a joint Department of Health and MRC initiative started in 1984 to assess recombinant DNA technology in relation to existing genetic services at Manchester, London, and Cardiff.<sup>85</sup> The evaluation included recommendations from the Health Service Research Unit at St Thomas’s Hospital, London for the establishment of a strong centralised bureaucracy at the national level to oversee the existing network of regional DNA laboratories.<sup>86</sup> None of the regional genetics centres provided the full range of tests available. Moreover, there was no

strategic overview to ensure the availability of effective testing services and to avoid unnecessary duplication of service provision. The differences between testing facilities were made up for largely by informal inter-centre service arrangements largely sustained by local interests. DNA testing had not become routine by this time, automation and kit-based technologies had yet to make a major impact on laboratory work. Most of the raw materials needed to do testing (e.g., DNA probes, restriction enzymes and gels for electrophoresis) could all be made in the laboratory, and with no expensive instrumentation, set-up costs for laboratories were low. There was, nonetheless, constant change and shifting ground within the technology of molecular diagnostics which meant that laboratory staff had to remain on top of advancements in basic research.<sup>87</sup>

British policymakers went on to outline steps to regulate the administration of genetic diagnostic tests and formalise inter-centre service arrangements on a national level for laboratories throughout the UK. The regulation of genetic testing began with the establishment of the Advisory Committee on Genetic Testing in 1996. The mandate of the Committee was to establish efficacy and product information requirements to be met by manufacturers and suppliers of genetic tests. The Committee was subsumed into the Human Genetics Commission in 1999 which began a public consultation on the future of testing regulation in 2002.<sup>88</sup> A 2003 White Paper, *Our Inheritance, Our Future*, subsequently outlined a commitment on the part of government of more than £18 million to upgrade NHS genetics laboratory facilities and £3.5 million to increase the workforce in the laboratories.<sup>89</sup> Consequently, two National Genetics Reference Laboratories were set up at Manchester and Salisbury to research and evaluate new technologies and ways

of testing, offer training, disseminate information on best practice, and expand existing quality assurance programs.

## **6. Conclusions**

When we consider ‘medical genetics’ as a historical subject, we find that its distinctiveness from other medical specialisms is a matter for debate and we do not seem to be dealing with a single set of techniques and methods. The story of medical genetics is therefore not a linear history of ideas, although ideas are at its centre. One can note a reductionist manoeuvre to unify and subsume (i.e., *reduce*), on the one hand, all that has been observed in clinical accounts of episodes of familial illness and, on the other, all that has been observed about cells, chromosomes, genes, proteins, and enzymes in genetic science, in order to explain processes and structures talked about in complex higher-order theories concerning the aetiology of hereditary diseases. The conceptualisation of ‘hereditary disease’ as a unique entity, like the common tendency in medicine to segment ailments of the body according to organ sites, has opened up new vistas of clinical research and practice, which, in turn, have justified the study of human genetics as a matter of serious concern for clinicians.

This paper shows that there were stages of consensus reached about a particular approach claimed as fundamental for knowledge about hereditary disease. Implicit in this discussion is an assumption that successful monitoring of patterns and rates of disease is the result of technological advances in science and medicine. Chromosomal and biochemical analyses provided the first clinical tools to uncover and determine the genetical element of certain heritable diseases. Correspondingly, the essence of the

physician-patient relationship in genetic counselling would be transformed from one of status to one of contract. New kinds of working relationships appeared with the growth and development of medical genetics as a service specialism. Sets of diverse actors in university-hospital settings coalesced into a new collectivity; and, as a collectivity, actors defined and/or redefined occupational roles and work rules.

Initially, an elite of geneticists built career paths through their work in newly established clinical settings for genetic health services. These individuals established specialised work patterns by combining hospital work and teaching posts. They drew a clientele of patients on the basis of personal reputations for specialised expertise in a manner that recalls what Victor Thompson described as ‘personal specialisation.’<sup>90</sup> Specialist status arose from the *person*, and not the *task*. Using Thompson’s nomenclature, there was high personal specialization in the medical genetics prior to specialty formation, but only one operative role, i.e., the geneticist in the teaching hospital setting. In the translatory movement from medical segment to medical specialty, the ideological direction of clinical practices conformed to a pattern widely adopted among contemporary medical specialties. As a result, a formal job classification – medical or clinical geneticist – became viable as a full-time occupation in medicine. ‘Task specialisation’ followed with counselling and laboratory services becoming standardised and specialised occupational roles and work rules for clinical and laboratory services being institutionalised across networks of regional genetic service centres in the 1970s.

As a final point, the paper indicates that, of late, networked innovation and the circulation of standards of laboratory and clinical practice are diffusing horizontally across a wide variety of specialty areas as the advantages the genetics-based approach are applied in different clinical settings and specialist activities *other than medical genetics*. This, I would like to suggest, represents a new development in what I have described as a bifurcated ideological construct that has shaped and informed the means of organising a genetics-based approach to medicine. The construct stipulates, on the one hand, that the mandate of medical genetics is to add a new set of medical procedures to the clinical repertoire of *all* health service providers. On the other hand, the mandate also provides for a class of *specialists* (i.e., medical geneticists) who will be available for consultation and assistance to *other* health service providers in pursuit of offering a genetics-based approach to medicine. The new development reinforces the idea that medical genetics is inclusive as opposed to exclusive in the occupational hub culture of medicine.

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Footnotes:

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<sup>1</sup> Hogben (1931), p. 214. Lancelot Hogben was appointed Chair of Social Biology and introduced genetics as a subject at the London School of Economics in 1930.

<sup>2</sup> The origins of the term ‘medical genetics’ are frequently attributed to Madge Thurlow Macklin, an American trained physician teaching histology and embryology at the University of Western Ontario. Biographies for Madge Thurlow Maklin are available in Soltan (1992), pp. 11-26; McLaren (1990), pp. 127-45. See, also, Comfort (2006), Kevles (1985). Hogben later uses the term ‘clinical genetics’ in his William Withering Memorial Lectures to the Faculty of Medicine of the University of Birmingham, published in his ‘Nature and Nurture’ (1933).

<sup>3</sup> Hogben (1931), p. 202.

<sup>4</sup> Hogben (1931), p. 214; cf., Macklin (1931), p. 614, (1933*a*), p. 1335.

<sup>5</sup> Hogben (1931), p. 215; cf. Macklin (1933*a*), p. 1335.

<sup>6</sup> Macklin (1932*a*), p. 485. According to C. Nash Herndon, Bowman Gray School of Medicine in North Carolina, Macklin was likely the first to introduce genetics into a medical curriculum ‘as a “bootleg” addition to another course’ in the early 1930s.’ See Herndon (1956), p. 2. Then again, Macklin remained employed only part-time as a sessional lecturer all the time she was in Canada and did not have an opportunity to teach a full course on human genetics until 1946 when she was appointed cancer research associate at Ohio State University by the National Research Council. It is noteworthy that Ohio State University was the site of the first required course in human genetics formally recognized as part of the curriculum of a medical school in North America in 1933.

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<sup>7</sup> Macklin (1933c), p. 20.

<sup>8</sup> Macklin (1932a), pp. 485-486.

<sup>9</sup> Macklin (1933a), (1935).

<sup>10</sup> Macklin (1933a), p. 1335.

<sup>11</sup> Hogben (1931), pp. 216-217.

<sup>12</sup> Kevles (1985), p. 205.

<sup>13</sup> Leeming (2004), pp. 483-484.

<sup>14</sup> Levine, Gursky and Rimoin (1977); Robertson and Haley (1946); Herndon (1954); Childs, Huether and Murphy (1981); Riccardi and Schmickel (1987).

<sup>15</sup> Lewis (1969), pp. 5-6.

<sup>16</sup> Daston and Park (2001), p. 149; cf. Findlen (1994), Pomian (1990).

<sup>17</sup> See López-Beltrán (2006).

<sup>18</sup> Bruner (1991).

<sup>19</sup> Epstein (1995), p. 25.

<sup>20</sup> López-Beltrán (2006), p. 47, Cartron (2007).

<sup>21</sup> In the *Traité de Médecine*, Fodérér stressed ‘constitutional weakness’ as a special condition that could produce a variety of illnesses or disabilities, and was inherited at the moment of conception, at birth, or during weaning. Given that the transmission of external physical characteristics and traits seemed constant among members of the same family, ‘there could be no doubt that the same transmission process was also at work in determining the internal constitution of the body.’ Cartron (2007), pp. 160-161.

<sup>22</sup> Ackerknecht (1982), p. 325.

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<sup>23</sup> Ciocco (1936), pp. 34-35.

<sup>24</sup> Draper (1944), p. 297.

<sup>25</sup> *Ibid.*

<sup>26</sup> Bauer (1945), p. *vi*.

<sup>27</sup> Timmermann (2001), p. 738.

<sup>28</sup> Bauer (1945), pp. 3-5.

<sup>29</sup> Bauer (1945), pp. 9-10.

<sup>30</sup> Paul (1945).

<sup>31</sup> A comprehensive survey of biological interest in the human constitution is available in Tucker and Lessa (1940*a*, 1940*b*). As regards constitutional psychology, see Sheldon, Steven and Tucker (1940). Further to this, the human constitution was subsequently taken up and advanced by physical anthropologists interested in the anthropometrical aspect of constitutional somatotyping. See, for example, Montagu (1947), chap. 8; Comas (1960), chaps. 4,5.

<sup>32</sup> Richmond (2007), pp. 169-70; Amundson (2005).

<sup>33</sup> Many of the writers in the field of medical genetics cite Garrod as a pioneer and founder. See, for example, Harper (2004), Dronamraju (1992), Scriver and Childs (1989), Emery (1968).

<sup>34</sup> Bearn (1993).

<sup>35</sup> Dronamraju, K. (1992), p. 217.

<sup>36</sup> Garrod (1902); cf. Sapp (1983), p. 318.

<sup>37</sup> Dronamraju (1992), pp. 216-217.

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<sup>38</sup> Scriver and Childs (1989), pp. 3-16.

<sup>39</sup> Prasad and Galbraith (2005), p. 201; Centerwall and Centerwall (1961); Mazumdar (1992), pp. 235-6. A preliminary test for phenylketonuria was developed in 1934 by a Norwegian physician, Asbjorn Følling, who identified abnormal levels of phenylpyruvic acid in the urine of two individuals with severe intellectual impairment and motor problems. The study of 'Følling's disease' was subsequently taken up from a genetics perspective in the mid-1930s by Lionel Penrose, a science graduate of Cambridge and physician, then employed by the Medical Research Council to conduct a detailed study of the origins of 'mental deficiency' amongst patients at the Royal Eastern Counties' Institution at Colchester. Biochemical studies by an American physician and biochemist, George Jervis, indicated that the disorder originated in the liver during infancy and was caused by an inherited inability to convert (metabolise) phenylalanine into tyrosine. A promising treatment was subsequently developed based on a low phenylalanine diet by a team of biochemists and paediatricians at the Birmingham Children's Hospital in the late-1940s.

<sup>40</sup> Mazumdar (1990), p. 182. Cf. Prasad and Galbraith (2005), p. 201; Scriver and Childs (1989), p. xi.

<sup>41</sup> Dronamraju (1992), p. 217; Scriver and Childs (1989), p. 11. Importantly, Dronamraju notes that Haldane incorrectly attributed the ideas to Lucien Cuénot, who, in 1902, published a demonstration of Mendelian ratios for the inheritance of coat colour characters in mice. See, also, Hickman and Cairns (2003), p. 839.

<sup>42</sup> Adami (1901*a*), (1901*b*).

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- <sup>43</sup> Moss (2003), pp. 25-28.
- <sup>44</sup> Weldon (1905*a*), (1905*b*).
- <sup>45</sup> Admundson (2005), pp. 148-158; Depew and Weber (1997), pp. 234- 235.
- <sup>46</sup> Gilbert (1978), p. 325.
- <sup>47</sup> McClung (1902).
- <sup>48</sup> Sutton (1903).
- <sup>49</sup> Maienschein (1984).
- <sup>50</sup> Moss (2003), p. 36; Gilbert (1978), p. 310.
- <sup>51</sup> Amundson (2005), p. 148.
- <sup>52</sup> Dunn (1965), pp. 190-191.
- <sup>53</sup> Wallace (1992), pp. 22-31.
- <sup>54</sup> Kevles (1985), pp. 238-248.
- <sup>55</sup> Harris (1995), p. 63.
- <sup>56</sup> Anonymous (1960).
- <sup>57</sup> Association of American Medical Colleges (1955).
- <sup>58</sup> Association of American Medical Colleges (1955), p. 19.
- <sup>59</sup> *Ibid.*, p. 19.
- <sup>60</sup> Macklin (1933*a*), pp. 1341-1343.
- <sup>61</sup> Association of American Medical Colleges (1955), p. 21.
- <sup>62</sup> *Ibid.*, p. 21-2.

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<sup>63</sup> Heredity clinics and heredity counselling on the basis of Mendelian genetics and statistical studies of family pedigrees can be found as early as 1940s in the UK and North America. See Leeming (2004), (2005).

<sup>64</sup> Reed (1963), pp. 14-15.

<sup>65</sup> Reed (1963), pp. 5, 228-229.

<sup>66</sup> Harris (1998), p. 336; Wertz and Fletcher (1988); Weitz (1981). The ethical issues surrounding genetic counselling and non-directiveness and the purposes of genetic counselling remain highly contentious and are beyond the scope of the present study. See, for example, Clarke (1990, 1991, 1993), Chadwick (1993), Koch and Svendsen (2005).

<sup>67</sup> Levine, Gursky and Rimoin (1977); Childs, Huether and Murphy (1981); Riccardi and Schmickel (1987).

<sup>68</sup> Motulsky (1979); Hickman (1981), p. 246.

<sup>69</sup> Following Robbins and Johnston (1976), I use the term 'ideology' in a restricted sense. It refers only to those systems of closely related beliefs, ideas and attitudes that exist among the groupings of medical professionals and scientists studied in this article.

<sup>70</sup> Self-reference removed for purposes of referee review.

<sup>71</sup> Riccardi (1977), pp. 2-3.

<sup>72</sup> For a detailed history of the origins of medical genetics in the UK, see Leeming (2005). For details on early genetic health centres in Canada, see Leeming (2004). For details on early medical genetics in the United States, see Comfort (2006).

<sup>73</sup> See, for example, Leeming (2001), Halpern (1988), Gritzer and Arluke (1985), Bucher (1972).

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<sup>74</sup> Leeming (2004).

<sup>75</sup> By comparison, there were eighteen service centres in Canada. It is more difficult to apply the notion of regional service centres in the United States owing to the fact that a broad cohesive national plan for building a genetic health service delivery infrastructure never materialised. On issues pertaining to infrastructure and genetic health service delivery in the United States, see Lin-Fu and Lloyd-Puryear (2000) and Centres for Disease Control and Prevention (1997). A listing compiled by the National Clearinghouse for Human Genetic Diseases (1980) provided the addresses of two hundred and thirty groups and organisations providing an array of counselling and laboratory services associated with genetic counselling, prenatal diagnosis, biochemical genetics, and cytogenetics. These include departments of paediatrics, neurology, ophthalmology, pathology, obstetrics/gynaecology, psychiatry, and dentistry in hospitals. Departments of medical genetics and genetics units in medical centres are also listed, as are various public health offices.

<sup>76</sup> Leeming (2007).

<sup>77</sup> See, for example, Weisz (2006), Leeming (2001), Döhler (1993), Abbot (1988).

<sup>78</sup> See, for example, Twaddle (2002), Giarelli (2004).

<sup>79</sup> Early expressions of this point of view appear in Riccardi (1977), Hickman (1981). See also Childs (1999), Leeming (2001), McKusick (2002), Harper (2008).

<sup>80</sup> Strauss (1978), pp. 224-233.

<sup>81</sup> *Ibid.*, pp. 227-228.

<sup>82</sup> Weatherall (1985), Bell (1998).

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<sup>83</sup> Elles (1996).

<sup>84</sup> Fitzsimmons *et al.* (1982); Harris *et al.* (1983), pp. 26-27.

<sup>85</sup> DHSS (1987). See, also, Harris *et al.* (1989).

<sup>86</sup> Rona and Beech (1993), p. 190.

<sup>87</sup> Elles (1996), pp. 16-17.

<sup>88</sup> Human Genetics Commission (2003).

<sup>89</sup> Department of Health (2003), pp. 28-29.

<sup>90</sup> Thompson (1964), pp. 25-27.