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Tracing the shifting sands of 'medical genetics': what's in a name?

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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'.[1] 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.[2] 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.[3] Moreover, Hogben's enthusiasm for promoting genetics as 'an exact science'[4] and the future potential of 'chromosome maps'[5] 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.[6] 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

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[1] 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.

[2] 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. 127–145. 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' (Hogben, 1933).

[3] Hogben (1931), p. 202.

[4] Ibid., p. 214; cf., Macklin (1931), p. 614; (1933a), p. 1335.

[5] Hogben (1931), p. 215; cf. Macklin (1933a), p. 1335.

[6] Macklin (1932), 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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> 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.[7]

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'.[8]

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 two hundred heritable diseases and discusses twin studies, consanguineous marriage, family pedigrees, and statistical techniques.[9] 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.[10]

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'.[11] 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.[12] 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.[13] 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.[14] 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.[15] 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 Ph.D.- and M.D.-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 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 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.[16] 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.[17] Case studies of *morbid haereditarii* (heritable disease)

[7] Macklin (1933b), p. 20.
[8] Macklin (1932), pp. 485–486.
[9] Macklin (1933a), p. 1335.
[10] Ibid.
[11] Hogben (1931), pp. 216–217.
[12] Kevles (1985), p. 205.
[13] Leeming (2004), pp. 483–484.
[14] Levine et al. (1977); Robertson & Haley (1946); Herndon (1954); Childs et al. (1981); Riccardi & Schmickel (1987).
[15] Lewis (1969), pp. 5–6.
[16] Daston & Park (2001), p. 149; cf. Findlen (1994), Pomian (1990).
[17] See López-Beltrán (2006).