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On the relevance of the “genetics-based” approach to medicine for sociological perspectives on medical specialization

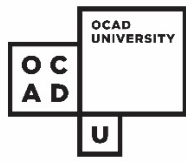
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On the Relevance of the “genetics-Based” Approach to Medicine for Sociological Perspectives on Medical Specialization

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Pre-Publication Draft

**On the Relevance of the “Genetics-Based” Approach to
Medicine for Sociological Perspectives on Medical Specialization**

Abstract: This paper draws on a study on the development of medical genetics as a medical specialism in the UK and Canada to reflect on how local and national contexts affect specialty formation. The paper begins by supporting earlier findings in the literature that stress, first, technological innovations as driving specialty formation, and, second, the domination of physicians in the division of medical labour. Beyond this, however, the paper explores the specific circumstances under which geneticists set about turning their work into a medical specialism based on a “genetics-based approach” to illness and how “medical genetics” as a specialism was assessed and configured to fit national and regional health service requirements.

In this paper, I draw on a work-in-progress summary of a cross-national study I am completing on the development and growth of medical genetics as a specialism in the UK and Canada to explore the circumstances under which the geneticists involved set about turning their work into a medical specialism based on a “genetics-based approach” to illness and how medical genetics as a service specialism was assessed and configured to fit national and regional health service requirements.¹ I have elsewhere (2001:457-65) provided a detailed survey of the relevant materials in the sociology of professions literature for studying national patterns of specialization in medicine and noted that at least four discrete lines of empirical research can be discerned, each one implicitly or explicitly representing a distinct way of understanding the specialization process and the collective character of the medical profession.

The first line of inquiry, building on earlier research that predicted incessant professionalization of occupations, stressed unfolding internal differentiation within professional organizations and the need to manage divergent beliefs, values and interests (e.g., Hughes, 1958, 1971; Janowitz, 1960; Glaser, 1964; Bucher and Stelling, 1964). A number of these writers indicated that increasing bureaucratization was limiting or reducing the range of activities and/or discretion of professionals (e.g., Wilensky, 1964; Hall, 1968; Johnson, 1972; Haug, 1973; Ritzer and Walczak, 1988). This was followed by a second line of inquiry, the so-called “power dominance perspective” on collective autonomy and professional control. With specific regard to medicine, Eliot Freidson (1970) argued that the organized autonomy of physicians had broadened into dominance over kindred occupations. Magali Sarfatti Larson (1977) emphasized the movement to standardization of practices and control over the dissemination of the knowledge base to dominate the market in knowledge-based services. Similarly, Andrew Abbott’s “system

of professions” (1988) put the accent on “cultural work” that would ensure that clients, competitors, the state and the public acknowledge the value of expertise and service offered by professionals. Relatedly, a third line of inquiry underscored mechanisms of social control and the dominance of medicine was said to have been extended through professionalization to areas of service that had previously come under other categories of everyday life such as giving birth, child rearing, and schooling (Zola, 1972; Foucault, 1973).

By contrast, one further line of inquiry, derived from a series of historical papers and books on medical specialties, was critical of the Anglo-American bias in sociological models of professionalization that focused on a strong degree of autonomy and self-governance among professionals and “analytical categories that appear to transcend the experience of particular professions” (Johnson, 1975: 186-7; cf. Shortt, 1983; Burnham, 1996). A different trajectory of professionalization, the historians pointed out, had occurred in Europe and elsewhere in which professional functioning had been largely affected by state control of resources and policies (Shortt, 1983; Geison, 1984: 2-3; Burrage, 1990; McClelland, 1991: 11-27). Authors consequently set about isolating for closer scrutiny the histories of significant institutions and groupings of practitioners who were self-identified as professionals at the national and/or local levels. Institutions and their members, it was argued, were not the by-products of professionalization but, rather, could be viewed as “an evolving set of solutions to changing problems” (Pernick, 1978: 1029).

Specifically with regard to the subject of the specialization process and the collective character of the medical profession, authors in the fourth line of inquiry have made a strong case for making a distinction between the medical specialization of the nineteenth century and the

specialization that takes place in the decades leading up to and following, roughly, 1950 (Geison, 1984; McClelland, 1991; Weisz, 2006 cf. Crosland, 1977; Burnham, 1998). The stages of development and periodizations that emerge shift focus away from the themes of encroaching bureaucratization and professional control. The emphasis is rather on the styles, function and consequences of medical research and teaching on innovation in clinical practice. Specialization in nineteenth-century medicine is associated with innovations in laboratory research and the training of general practitioners (Weisz, 2006: 3-63, cf. Shortt, 1983; Ramsey, 1984). And the major locus for empirical study here is the combination of medical school and teaching hospitals. Specialisms become the dominant form of medical practice and take away from earlier forms of “general” practice. More particularly, new approaches to professional training and certification emerge alongside new ways of framing the regulation and standardization of health care practices. As George Weisz (2006: xvi-xvii) observes, medical specialization “evolved from a largely local to a national phenomenon, producing new kinds of specialist associations, transforming the institutions of the larger medical profession, and, in some cases, becoming an issue for political authorities.” Additionally, he notes, since the 1960s, “many new specialties and subspecialties have grown out of innovations produced by the academic research sector (reproductive medicine, medical genetics), while others are the consequence of what are perceived as new social needs (family and adolescent medicine, geriatrics)” (2006: 231).

Specifically, and with a view to the ideas of evolving stages of development and periodizations, I examine in this paper how a range of medical interests were brought together around: (1) Mendelian genetics and the adoption of probability and statistics as scientific methods of quantifying the risk of heritable disease; (2) the acceptance by medical academia of

probabilistic and new multifactorial models of disease aetiology; and (3) the negotiated protocols and standards of medical practice worked out by bodies such as the relevant royal colleges, the linked associations and societies for medical professionals, affected training and research authorities, and government.

*The Medico-scientific Background of Medical Genetics
in North America and the UK*

The available histories suggest that although the study of genetics gained scientific acceptance between 1915 and 1930 in Norway, Sweden, Denmark, the United States and the Soviet Union, it was less well received in Germany, and quite poorly received in the United Kingdom and France (Allen, 1978: 278-283; Harwood, 1993: 143-56). By and large, European scientists were more interested in studying the grander Darwinian theories of evolution and trained in descriptive and qualitative methods of research (Sapp, 1983; Rushton, 2000). Those supporting the new probabilistic reasoning of Mendelian theories of genetics tended to have a background in experimentalism, and were more likely to be found in the agricultural and social sciences, and in disciplines such as demography, vital statistics, and agricultural science.

The term “medical genetics” first appeared in articles written by Madge Thurlow Macklin (1932, 1933), then teaching histology and embryology at the University of Western Ontario. The term also appears in books written by John A. Fraser Roberts (1940), then Principal Investigator at the Burden Mental Research Department, Stoke Park Colony, Bristol, and Laurence H. Snyder (1941), then professor of medical genetics at Ohio State University. These publications were ostensibly introductions to applied human genetics for medical students and physicians. At no time, however, do the early authors on medical genetics *qua geneticists* claim to be specialists in

the field of medicine. They are, rather, appreciative reporters of innovations in science. Common concerns, nonetheless, are evident and all have to do with the perceived need, first, to close the gap between theoretical and clinical capability in the application of genetics-based knowledge in clinical practice, and, second, to educate individuals about the practical uses of genetic theory in medicine.

Daniel Kevles (1985:205) has indicated that fewer than two hundred people published any research in the early Anglo-North American contingent of human geneticists. 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 (Leeming, 2004: 483-4). But little is actually known about the contents of early human genetics research and teaching in Anglo-North American universities and colleges. What we do know is that the Association of American Medical Colleges (AAMC) in North America sponsored the first organized efforts to gather information on the extent of genetics instruction in medical schools as early as 1946 (Leeming, 2004: 484-6). An AAMC report on of a 1954 workshop on “Objectives of Teaching” is of particular interest for the unexpectedly high levels of demand indicated for genetics instruction in medical schools (Subcommittee on Objectives of Teaching, 1955). Moreover, the discussion generated by the recommendations of the Report appears in a number of later surveys and reviews on the subject.²

Clinical techniques 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” (1955: 19). Turning to the question of who should teach genetics, participants in the

workshop generally agreed upon “a trained medical geneticist on the staff” who “could also have service and research functions” (1955: 21-22). Thus, a multi-faceted role is envisioned for medical geneticists working in North American teaching hospitals. As a “staff geneticist,” the medical geneticist would provide clinical consultation and physical examination with respect to cases of: birth defects, known inherited disorders in families, multiple miscarriages and stillbirths, mental retardation (where cause is unknown) or developmental delay, growth disorders, dysmorphological features, and ambiguous genitalia or abnormal sexual development.

Comparatively speaking, five subsequent surveys, completed over a period of three decades, show that the proportion of medical schools with formal courses in genetics increased from 8.6 per cent in 1953 to 86.5 per cent in 1985.³ At the same time, three of the surveys indicated that, rather than an integrated curriculum, genetics instruction increasingly became the preserve of paediatricians and geneticists working in independent genetics departments. This matter went on to be described in reviews and surveys as an *interdisciplinary problem*; a problem frustrating the larger goal “that ‘genetically thinking’ becomes an integral part of [all] medical practice” (Motulsky, 1979; Hickman, 1981: 246). From this emerged what I (2001: 468-69; cf. Yoxen, 1982) have elsewhere called a bifurcated ideological construct that shaped and informed the means of organizing what was being called a “genetics-based approach” to disease.⁴ 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* health disciplines. On the other hand, it indicated that when and where service providers were unable to deliver the new procedures, a class of specialists (i.e., medical geneticists) would be available for consultation.

A useful observation to be made at this point about “medical genetics” is its peculiar

character after 1950; that of an elementary albeit synthetic development in medical training and academic research. Interviewees in the cross-national study I am completing on medical genetics in the UK and Canada have described a kind of Mertonian science community, and the type of specialized work associated with the geneticist in clinical practice recalls what Victor Thompson (1964:25-7) described as “personal specialization.”

In Merton’s science community, the reader will recall, the exchange of expert knowledge is straightforward and non-repetitive across a market interface that requires little in the way of social infrastructure (Merton, 1973; cf. Hagstrom, 1965; Storer, 1966). Scientific labour entails broad-based, undirected research that is usually conducted in universities under the volition of the individual researcher, passed on to society through peer-reviewed journals and other forms of knowledge transference, which leads in turn to goods and services. The production of knowledge is driven by a self-contained reward system: the researcher gains prestige in the scientific community and his/her commitment is sustained through intradisciplinary control of status and entitlements more than monetary rewards (e.g., academic appointments, formal titles, weighted indices of publications, authorial citations, etc.). At the same time, the rate and direction of knowledge production remains unpredictable, and involves the play and capriciousness of scientists’ personal interests. Specialist behaviour thus arises from the *person*, and not the *task* (Thompson, 1964:27).⁵ Using Thompson’s nomenclature, there was high personal specialization in the medical genetics of the 1950s, but only one operative role, i.e., the geneticist in the teaching hospital setting.

Individuals interviewed for my study reported that the circumstances of medical geneticists changing significantly after the 1950s. According to interviewees, new kinds of

working relationships appeared, and occupational roles and work rules changed. All interviewees pointed to technological innovations in the form of new laboratory technologies for identifying chromosomal anomalies and genetic metabolic disease as driving the rate and direction of changes in the workplace. More particularly, local associative strategies surrounding specialty formation and the institutionalization of medical genetics in North America and the UK would go on to emphasize multidisciplinary task specialization among genetic health care providers and more or less continually unfolding internal differentiation among academic health centres, community clinics, consumer and voluntary groups, government agencies, and public health departments. That said, in contrast to the UK and Canada, a broad cohesive national plan for building a genetic health service delivery infrastructure in the United States never appeared (cf. Lin-Fu and Lloyd-Puryear, 2000; Centres for Disease Control and Prevention, 1997).

Medical Genetics: The Canadian Case

Fiona Miller's study (2002) of early developments in Toronto throws highlight on Norma Ford Walker's accomplishments in the field of dermatoglyphic pattern analysis. Indeed, following the discovery of the chromosomal basis of Down's syndrome in 1959, scientific interest in dermal patterns intensified, and pattern analysis was recognized as a useful method for diagnosing patients for chromosome analysis (Thompson and Thompson, 1966: 245-6). But what is problematic here is Miller's claim that Walker's research was "marginal" and that she was involved in establishing a "school" of medical genetics. In actuality, Norma Ford Walker had little interest in medicine *per se*.⁶ Trained as an invertebrate zoologist at the University of Toronto; she gained considerable notoriety in science circles for her work on the dermatoglyphics of the Dionne quintuplets, the first quintuplets known to survive infancy.⁷ Contrary to Miller's characterization of this work as "iconoclastic

genetics,” the study of the quintuplets completed in 1937 was a significant contribution to contemporary studies of “mental likeness” in twins and intelligence testing. The importance of twin studies, especially monozygotic twins (i.e., from “one egg,” identical) and reared together, was widely viewed as proof positive of the perdurability of human nature (Gould, 1981: 234-320). More particularly, the Dionne study lent credible support for Gesell’s proposition that even the most identical twins had inbred differences that could become more or less profound according to motor training and mental stimulation (Gesell and Thompson, 1929).

Norma Ford Walker’s subsequent appointment as a genetics consultant in 1940 at the (Toronto) Hospital for Sick Children, entailed a kind of resource exchange relationship: Hospital clinicians would send patients and families to her. She would calculate patterns of hereditary transmission and counsel the families on issues relevant to family planning and the recurrence of familial traits. In return, she was permitted to freely pursue her own research interests, along with those of her students, in the hospital. It is important to note, however, that the hospital’s arrangements for Walker’s services never included laboratory supports. Thus, Ford Walker’s successors encountered a fragmented set of service operations that had failed to keep pace with other genetic services in North America. In actuality, a fully integrated Division of Clinical Genetics, jointly responsible to the Department of Genetics and the Department of Paediatrics, did not appear in Toronto until 1986.

Cumulative links between genetics and specialized areas of medical research and services can be recognized more easily in developments at McGill University in Montreal (Leeming, 2004: 488). The interwar years saw well-known figures such as the English hereditarian H. B. Fantam as McGill’s Strathcoma Professor of Zoology, and a brief stint of teaching by Lancelot Hogben, who

went on to become professor of social biology at the London School of Economics. In 1934 an independent department of genetics was formed. There had been disagreement at the university level between the departments of zoology and botany about who should teach undergraduate genetics. The dispute was resolved by way of receiving a Rockefeller Foundation grant to start up an independent genetics department. The first chair was C. Leonard Huskins, a plant geneticist who also consulted with local physicians on Mendelian disorders. In 1949, F. Clarke Fraser, a PhD-geneticist in his final year of medical school, was asked to set up a genetic counselling service at the Montreal Children's Hospital. This came as a result of negotiations between J. Wallace Boyes, Huskins's successor at McGill, and Alton Goldbloom, the hospital's chief of paediatrics. A department of medical genetics was formally approved at the hospital in 1951, with a modest staff consisting of Fraser and an assistant.

Developments of the sort described above at Toronto and Montreal can reasonably be viewed as starting points for later developments in Canada. Of the fifteen sites in Canada that provided some combination of medical genetics training and services by the end of the century, eight (Edmonton, Vancouver, Winnipeg, London, Montreal [Ste-Justine], Quebec City, Hamilton, Kingston) were set up by individuals who had studied in Toronto and Montreal. The remaining five sites (Saskatoon, Ottawa, Halifax, Calgary, St. John's) were set up by individuals who originally trained in the UK. Only two of these received training in genetics in the UK, however. One trained in Canada, another in the United States, and a third "picked it up on the job" (Leeming, 2004: 488). Thus, Toronto and Montreal figure into the story of medical genetics in Canada as key sites of information exchange and genetics training. But, then again, this was a period when there were few referrals of patients for genetic counselling. The exchange of information about genetics was pursued largely in terms of

teaching and research interests by individuals who only provided counselling when called upon to do so. What's more, one has to consider how important links with the United States were for collegial support and approbation.

Diane B. Paul (1988: 137) and Daniel Kevles (1985: 199, 208-9, 252-4) have indicated that in the two decades following the Second World War “virtually all institutional patrons [in the United States] of work in medical genetics and genetic counseling also had eugenic motivations.” Specifically, attention is drawn to support from the Rockefeller, Carnegie, Wenner-Grenn, McGregor, and Rackham foundations, the Commonwealth and Pioneer Funds, and the American Eugenics Society. Canadian geneticists, by contrast, did not enjoy this kind of support during this period (McLaren, 1990: 90-1, 99-106, 108, 112-115). As noted above, the genetics department at McGill had received some start-up funding from the Rockefeller Foundation. But this was the exception.

It was the American Society of Human Genetics, established in 1949, which provided Canadian geneticists with opportunities to meet and exchange information with like-minded fellows on an intra-continental level. The idea for the Society was proposed at an informal gathering in December 1947 held during the meetings of the American Association for the Advancement of Science at Chicago. Within five years the membership had grown to 565 individuals with 316 institutional subscribers to the journal.⁸ Americans made up 84 per cent of the membership at the time. Of the remaining 16 per cent, 5 per cent were Canadians. That said, of the 27 Canadian members, 25 were from Ontario and Quebec. And 14 of these individuals were affiliated with the University of Toronto (9) and McGill (5). Moreover, from the start, Canadians served as directors of the Society and editors of the journal.

A wholesale shift in the importance of “eugenics” for medical professionals interested in genetics followed certain irreconcilable differences at the American Society of Human Genetics between the ameliorism of the eugenic practices and what was being presented as the value-free science of human genetics. This shift found 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” (Reed, 1955: 14-15). Non-directive genetic counselling, in the context of a medical service provided by staff geneticists (*à la* the 1954 American Association of Medical College report), was to be viewed in terms of a contract for services and the essence of the physician-patient relationship would be transformed from one of status to one of contract.

Importantly, the movement to formally standardize genetic counselling services was linked to changes in the clinical division of labour, changes that were integrally linked to the development of new diagnostic tests and laboratory services in the 1960s. The first development involved the investigation of chromosomal abnormalities. Human chromosomal analysis in the 1950s and 1960s involved techniques largely developed in cytological studies of animal and plant species carried out in the 1920s and 1930s. Improved methods during this period made it easier to count human chromosomes and to study their morphology. This, in turn, permitted some types of chromosomal abnormalities, including missing or extra copies of a chromosome or gross breaks and rejoinings (translocations), to be detected by microscopic examination. The presence of an additional small acrocentric chromosome in typical cases of Down’s syndrome was first reported in France in 1959 and quickly followed by reports from cytological laboratories in England (Lejeune *et al.*, 1959; cf.

Ford, Jones *et al.*, 1959; Ford, Polani *et al.*, 1959). The publication of these findings in quick succession during 1959 caused a sensation among scientists and clinicians alike (e.g., Stevenson, 1961). The development of human cytological genetics or “cytogenetics” provided, for the first time, clinical tools to uncover the genetical make-up of relatively common disorders associated with mental illness.

Lejeune’s announcement of the discovery of trisomy 21 and Down’s syndrome took place at a seminar in the McGill University genetics department in 1958, following the meetings of the International Genetics Congress at Montreal that summer. Jacques Gagnon, a pathologist at Université de Montréal, went to study with Lejeune in France and then brought cytogenetics back to Montreal at l’Hôpital Ste-Justine in 1959. Cytogenetics was established at McGill the following year when Louis Dallaire set up a laboratory to study chromosomal translocations for his PhD research under the supervision of Clarke Fraser. Fraser, who was also the geneticist on staff at the Montreal Children’s Hospital, subsequently asked Dallaire to develop a service laboratory for the hospital in 1964. A succession of regional cytogenetic services quickly followed in other provincial teaching centres: University of Alberta Hospital at Edmonton (1962); Department of Paediatrics, University of Saskatchewan at Saskatoon (1964); Ottawa Civic Hospital (1965); Department of Pathology, Queen’s University at Kingston (1968); Children’s Hospital of Manitoba at Winnipeg (1969); Foothills Hospital at Calgary (1969); and Izaak Walton Killam Hospital at Halifax (1970).

A parallel development to chromosome analysis, biochemical testing, provided new ways of identifying genetic events by revealing abnormal metabolites in body fluids. After 1960, the basic division of labour involved in biochemical testing followed a pattern similar to that of

chromosome analysis: individuals with backgrounds in chemistry were recruited to perform a service function in “biochemical laboratories,” and a new occupational category appeared, “biochemical geneticists.” Physicians would look for tell-tale signs and symptoms (e.g., failure to thrive, developmental delay, ocular abnormalities) that might be indicative of metabolic disease. A geneticist would be consulted regarding the family history and, if a laboratory evaluation was in order, blood or urine was obtained and shipped to the laboratory where it would undergo testing. A laboratory report would be returned to the consulting physician with information about a geneticist who was available for consultation.

The movement to introduce newborn screening programs in the provinces began when a simple and inexpensive metabolite inhibition assay was developed in the United States to detect a treatable metabolic disease. The test, combined with a treatment (dietary phenylalanine restriction), had led to a highly successful therapy for phenylketonuria. Therapeutic and/or curative interventions followed for galactosaemia, congenital hypothyroidism, aminoacidopathies, fructose intolerance, tyrosinaemia, and other metabolic conditions. Newborn screening programs for these conditions were subsequently set up in nine provinces, between 1963 and 1969, largely administered through provincial public health programmes (Leeming, 2004: 492-3). Only in Quebec did geneticists take a proactive role in the organization of a province-wide program (Leeming, 2004: 493).

Despite the fact that Canadian newborn screening services came mostly under the purview of public health branches and departments of health and community services, laboratory and counselling services were provided by the genetics departments in teaching hospitals. In addition, geneticists experienced increases in workload as techniques for culturing foetal cells

from amniotic fluid were refined and amniocentesis became useful as an outpatient procedure for obtaining test samples for chromosomal and biochemical analysis from pregnant women.

Obstetricians performed all the amniocentesis procedures. Geneticists provided pre-procedure counselling to inform the patient of the risks associated with the procedure. The geneticists would also schedule when and where the procedure was offered. Furthermore, the geneticists would review test results and provide counselling in the event of a positive result.

The first concerted effort to monitor the rate and direction of genetic counseling and laboratory services was mounted in the early 1970s when Canadian geneticists realized that they were spending proportionally less time in the classrooms and labs and more time in the clinics. Members of the Genetics Society of Canada established a committee in 1971, the Committee on Genetics as it Relates to Social Problems, with a mandate to examine standards of care, payment for services rendered, and the training and accreditation of service providers. A survey by questionnaire of twenty-two individuals known to be providing genetic services in thirteen cities (Toronto, Montreal, Quebec City, Vancouver, Victoria, Ottawa, Kingston, London, Hamilton, Winnipeg, Edmonton, Saskatoon, Halifax) confirmed that an increase in demand for genetic counseling had occurred and attributed the increase to innovations in laboratory and obstetric services (Miller, 1972). The survey showed that the costs of laboratory services were not directly reimbursed through provincial health care insurance schemes. Costs were largely paid for with research funds, or were being absorbed through hospital global budgets.

The problems identified in the survey were made the focus of an informal meeting two years later that was organized as part of the 1973 annual conference of the Genetics Society of Canada in North York, Ontario at York University. It was here that the idea of creating a formal mechanism to

maintain standards of genetic services first surfaced. In a statement prepared by the “Committee on Genetics as it Relates to Social Problems” (1973), and approved by the Genetics Society of Canada Executive, a recommendation was made that the Society lobby the appropriate federal and provincial government authorities, first, to acknowledge genetic counselling was an important health service, and, second, to develop a means of accrediting the centres offering laboratory and counseling services. Further to this, multi-disciplinary centres were described that would integrate PhD-geneticists (i.e., non-physicians) into clinical practice. Interviewees for the present study who participated in the meeting said that they were deliberately trying to topple Royal College restrictions surrounding who and who could not provide patient care.⁹ What emerged from all this was a coalition to form a corporation to be known as the Canadian College of Medical Geneticists.

A proposal to create the Canadian College of Medical Geneticists was presented to an assembly of thirty-three individuals who were invited to attend a three-day meeting at the Guild Inn in Scarborough, Ontario in November of 1974. All attendees were providers of genetic services and nine provinces were represented. A statement from the Steering Committee (1975) followed in the April 1975 issue of *The Genetics Society of Canada Bulletin* declaring the proposed College was “not a scientific society such as the Genetics Society of Canada, but an organization concerned with the establishment and enforcement of professional standards on health care delivery in the field of Medical Genetics.” Later statements asserted that the College would consist of and represent “those properly qualified PhDs and MDs” and affirmed “that individuals with a PhD have a role to play in delivering genetic services.” The delivery of services would be associated with medical centres where physicians trained in medical genetics would provide patient consultation. The centre would assume responsibility for monitoring the quality of the laboratory services and accreditation

evaluations would be formulated by the Canadian College of Medical Geneticists.

The application for the incorporation of the College was recorded by the Ministry of Consumer and Corporate Affairs on January 13, 1976. The initial focus was on recruiting as members a fair representation of individuals in active clinical practice. A special dispensation was devised to grandparent individuals deemed to already possess sufficient knowledge and skills to be considered Fellows of the College. Additionally, in 1976, the Board courted controversy by submitting a proposal to the Royal College of Physicians and Surgeons (Canada) requesting the formation of a sub-specialty in medical genetics. Despite the concern of many members that PhD-geneticists might get left behind if such an application were accepted, the College of Medical Geneticists and the Royal College pursued a series of negotiations over the next decade that would ultimately change the course of delivering genetic services in Canada.

In 1981, the College formally declined an offer made by the Royal College to grant a certificate of special competence in medical genetics to Fellows of the Royal College certified in either paediatrics or internal medicine as their primary certification. However, members continued to talk informally to individuals on the Royal College's Committee on Specialty Development while, simultaneously, making inquiries with the provincial licensing bodies about the Canadian College of Medical Geneticists becoming some kind of national accrediting body. It was in fact not until 1988 that the Specialty and Manpower Committee of the Royal College supported the creation of a free-standing specialty with a five-year training program. The recommendation was approved by the Credentials Committee and Royal College's Council so that, in 1989, medical genetics attained status as one of fourteen clinical specialties recognized by the Royal College's Division of Medicine. By the time Canadian medical geneticists had gained specialty recognition from the Royal College

there were eighteen centres in eight provinces that provided counselling and laboratory services. All belonged to university-hospital affiliated programs, with the exception of three centres in Ontario (Oshawa, Credit Valley, North York). In addition, nine provinces had established outreach programs whereby staff from genetics centres was dispatched on a regular basis to hold clinics sites in outlying areas. As a final point, it is worth noting that the role of PhD-geneticists was overturned in terms of a clinical role in medical genetics during the process of specialty formation. PhD-geneticists continued on in the field with respect to teaching human genetics in university medical faculties and with respect to the delivery of laboratory services independent of the counselling services. *As a rule*, however, they ceased to counsel patients or administer regional genetics centres in Canada.

Medical Genetics: The UK Case

By all accounts, the average physician in the UK was not terribly interested in genetical explanations about the relationship between heredity and disease causality until well after 1960. Indeed, David Lewis's survey of the membership lists of the Genetical Society of Great Britain shows a sharp increase in members involved in medical research after 1959, rising steeply to 1969 when nearly 12 per cent of the 900 members of the Society were working in medicine (Lewis 1969: 5-6). Lewis attributed this increase to technological advances in human cytological research and work being done in the area of chromosomal abnormalities in humans. He also cited the research surrounding the Rh blood group and haemoglobin variants being done at the National Institute for Medical Research, Lister Institute, Chester Beatty Research Institute; and the Medical Research Council units in the UK.

By 1964 there was sufficient medical interest in a genetical understanding to disease to sustain both a specialist journal, *Journal of Medical Genetics*, and a new society, the Clinical

Genetics Society. Initially, the Clinical Genetics Society represented a kind of divided nucleus within the emerging field of medical genetics; a polycentric structure with multiple bases of interest (Leeming, 2005: 547-51). At the most basic level, the Society was divided along lines of primary specialization, although the area of paediatrics came into the foreground with the Society's early recommendations on the training of clinical geneticists in the early 1970s. The Paediatric Specialist Advisory Committee of the Royal College of Physicians supported a resolution that the Member of the Royal College of Physicians (MRCP) would be considered by them to be an essential qualification for genetic service providers, as it was then for consultants in every other branch of clinical medicine. The Advisory Committee's recommendations followed a particular line of logic: parents with a family history of hereditary disease or congenital malformations were traditionally counselled by paediatric specialists with expertise in the care and treatment of diseases and disorders of the newborn. Furthermore, the new testing regimes of the 1960s for identifying chromosomal anomalies and metabolic diseases had widened the remit of genetic consultation within paediatrics. And geneticists were clearly experiencing increases in workload with the work associated with new programs in newborn screening, mid-trimester amniocentesis and antenatal diagnosis.

At the same time, the membership of the Clinical Genetics Society had a varied background including specialists in ophthalmology, psychiatry, obstetrics and general practice – fields that were not branches of clinical medicine under the Royal College. Having the MRCP as a training requirement, many believed, would be too restrictive and potentially hold back the development of a genetics-based approach to health care in clinical specialty areas other than paediatrics. Further to this, concerns were raised about the evolving relations between clinical

and laboratory personnel. Some members went so far as to suggest that training for medical genetics should require the MRCPATH. In other words, geneticists should run laboratories under the control of departments of pathology *and* also see patients. Alternatively, others suggested separate roles for the clinician and scientist, advocating a formal association be made between clinical genetics, cytogenetics and biochemical genetics. In the end, the arguments and debates resulted in a breakup within the Clinical Genetics Society. The cytogeneticists, fearing a paediatrics-dominated Society would exclude non-physicians, split off and formed their own body, the Association of Clinical Cytogeneticists. The Association became an interstitial organization that arbitrated in matters concerning the research and service functions of cytogenetic laboratories. This set the example, much later, in 1988, for a third body, the Clinical Molecular Genetics Society, representing scientists interested in diagnostic applications of molecular genetics.¹⁰

In 1978, Alan Johnston, reporting on the findings of the Clinical Genetics Society's *ad-hoc* working party on the training of medical geneticists, remarked that although the chance convergence of different disciplines into the field of human genetics had resulted in a number of spectacular contributions "which have had repercussions through the whole science of genetics," and medical genetics had progressed to the point where highly specialized training and professional standards of practice were now necessary (Johnston, 1978). Citing the example of the recently incorporated Canadian College of Medical Geneticists, the UK working party set out to recommend standards of health service delivery. The medical geneticist of the future would have four main functions;

... to contribute to diagnosis (including antenatal); to counsel patients and their relatives; to

maintain genetic registers; and to act as consultant to cytogenetic, biochemical, and other relevant laboratories. In short, he or she would be responsible for organizing a comprehensive genetic advisory service. Additional functions would be involvement in teaching and research, which would be particularly important since he would be working mainly in major centres, usually teaching hospitals. Thus a joint NHS/university appointment at consultant level to a Genetic Advisory Centre is preferable (Johnston, 1978: 260).

The role of the medical geneticist and the creation of regional genetic services were topics pursued in detail by the Clinical Genetics Society in two subsequent working parties (Fitzsimmons, *et al.*, 1982; Harris *et al.*, 1983). The Royal College of Physicians became involved, initially through the work of the Medical Genetics Sub-Committee of the Paediatric Specialty Advisory Committee and, in 1984, with the formation of a standing Committee on Clinical Genetics. Both the Medical Genetics Sub-Committee and the standing Committee were largely made up of members of the Clinical Genetics Society.

As in the Canadian case, the initial inquiries into the status of genetic diagnostic and laboratory services in the UK showed that there was considerable variation in the range of services offered from region to region and much of what made up service arrangements was largely dependent on university and research funding. Geneticists offering these services, on the other hand, were clearly willing to cooperate with government programs for integrated services so long as sufficient resources were made available.¹¹ The Clinical Genetics Society consequently set up working parties in the 1980s who in turn endorsed an “integrated regional genetics service” that would act as the “focus to which the primary health care team, hospital consultants and other specialists should direct particular problems relating to inheritance, including the diagnosis and prevention of birth defects” (Fitzsimmons *et al.*, 1982: 2). Moreover, each service centre would set up computer registration of genetic diagnostic information (genetic

registers) for the express purpose of tracing, following-up, and counselling individuals at risk of having or transmitting a serious genetic disorder. These centres would, in accordance with the organizational structure of NHS Regional Health Authorities, serve a population of 1 ½ to 3 million and would, on this basis, provide for several District General Hospitals. The rationale for the allocation of catchment areas was based on the calculation of the proportion of, first, congenital malformations and, second, chronic disease with a major genetic component. In addition, the 1983 Working Party envisaged programmes to extend counselling to district general hospitals through regular visits by centrally based whole-time clinical geneticists (Harris *et al.*, 1983: 22). Local clinicians would prepare information on patients in advance of the “satellite” clinics and visiting clinical geneticists would share after-care responsibilities with family practitioners.

At a high level of generality, the formal structure for the integrated genetics service proposed by the Clinical Genetics Society fit well within the regimes of service delivery instituted between 1974 and 1982 when the National Health Service was reorganized 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. The “regional genetic service” 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. The organization of the genetics centres themselves would follow along the lines of the multidisciplinary centres established earlier in Canada – but with noticeable differences. Consultants (i.e., clinical geneticists) were responsible for genetic counselling and syndrome identification for all referred

individuals. Correspondingly, there was close collaboration between the clinical geneticists and the heads of laboratories in the areas of, initially, cytogenetics and biochemical analysis and, later, molecular analysis. Both clinical geneticists and scientists participated in the administration of services as well as in the forward planning of genetics services at the regional level.

*Discussion: Professionalized Human Geneticists,
Specialization, and Networked Innovation*

“One of the main difficulties in dealing with the new genetic technologies,” Lassen and Jamison have perceptively noted in their study of public opinion on the subject, “is that they are solutions in search of problems” (2006: 8). Genetic technologies have not been developed to cure diseases, they observe, “even though such claims are often made on their behalf” (*ibid.*). Nonetheless, we tend to see geneticists as major players in the future of science and medicine. And we take very seriously the implications of their work for *our* futures. This last point is well-illustrated in the collection of papers brought together for this special issue of the CRSA, *Genes and Society: Looking Back on the Future*.

In this paper, I began by describing how a range of medical interests came together around the adoption of probability and statistics as scientific methods of quantifying the risk of heritable disease and the acceptance by medical academia of probabilistic and new multifactorial models of disease aetiology and genetics. Until the 1950s, medical interest in human genetics was confined to personal specialization (*à la* Thompson) and the capriciousness of individual scientists and clinicians. With time, formal recognition of the term “medical genetics” took hold, spurred on by a growing number of supporters, a narrow base of original conceptual unity, and the emergence of new laboratory technologies, around 1960, for studying chromosomal

anomalies and tests for genetic metabolic disease. These general-purpose technologies fueled the vigorous and sustained growth of genetic health services and were clearly implicated in significant changes in health care delivery systems. This included driving the rate and direction of changes in the workplace. An intrinsic technical logic and language evolved in the 1960s that provided a kind of *lingua franca* spoken by providers of “genetic health services.” After 1970, local associative strategies surrounding specialty formation and the institutionalization of medical genetics in Canada and the UK would go on to emphasize, first, multidisciplinary task specialization among genetic health care providers and, second, more or less continually unfolding internal differentiation among academic health centres, community clinics, consumer and voluntary groups, government agencies, and public health departments. Medical genetics *as a specialism* consequently gained a semblance of unanimity over its basic reference points and arrived at a meaning directly tributary to current acceptance of the term.

But such a bald description actually tells us little about the heterogeneity of genetics-based health services in Canada, the UK, and elsewhere. Medical genetics is, of course, itself an occupational category and thus predicated on “insider-outsider” relations. Generally speaking, the medical geneticists studied in this paper claim the genetics-based approach to illness as their area of expertise (i.e., a “jurisdiction” *B la* Abbott). But, at the same time, the “genetics-based approach” to illness operates as a bifurcated ideological construct that shapes and informs a wider range of practices. The construct stipulates, on the one hand, that the mandate of medical genetics is to add a new set of biomedical procedures to the clinical repertoire of *all* health disciplines. On the other hand, it points out that when and where service providers are unable to deliver the new procedures, a class of specialists (i.e., medical geneticists) are available.

Indeed, specialists in *other* areas of medical practice have increasingly come to see to the treatment and medical management of patients with numerous genetics-based components, often providing information to patients concerning “the genetics” of the condition at hand (Hedgecoe, 2003; Kerr, 2004; Cox and Starzomski, 2004). This may be viewed as evidence of competing segments and jurisdictional disputes; as an encroachment on the medical geneticists’ claim to jurisdiction. But medical geneticists *themselves* do not view the situation this way. Generally speaking, they see *other* specialists coming around to their way of thinking; as adopting a genetics-based approach. The argument goes something like this: In the first place, the jurisdictional interface with 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., a genetics-based approach to medicine) can be seen to diffuse horizontally across a large number of specialty areas, as its advantages apply to different activities. As awareness of the genetics-based approach to illness increases in medicine globally (i.e., *in general*), collegial recognition of the expert role of the medical geneticist in medicine is fortified. Medical genetics is therefore inclusive as opposed to exclusive in the occupational hub culture of medical professionals.¹²

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Notes

¹ The material that follows draws on findings reported in two earlier papers (Leeming, 2004; 2005).

Two kinds of data are used in the comparative case study. Preparatory research was based on the analysis of primary and secondary source materials drawn from a range of libraries and archives. In addition to providing technical details and facts, this material furnished a source of general information about practical genetics applications against which to compare the responses forthcoming in interviews – the second source of data for this study. Open-ended qualitative interviews began in April 1997 in Canada and the UK. To date this has involved a sample of one hundred and sixty-four interviewees.

² See, Neel (1957), Hickman (1981), Childs, Huether and Murphy (1981), Childs (1982), Davidson and Childs (1987), Riccardi and Schmickel (1987), Graham, *et. al.* (1989).

³ See, Levine, Gursky and Rimoin (1977); Robertson and Haley (1946); Herndon (1954); Childs, Huether and Murphy (1981); Riccardi and Schmickel (1987).

⁴ Following the example of Robbins and Johnston (1976: 353), 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. It is not here used in its broader sense, i.e., as a *Weltanschauung*. I am interested only in how geneticists make sense of medical genetics *and* seek to further their collective professional aims. This can be contrasted with, for example, what the historian Susan Lindee has called “genetic essentialism” and “genetic folklore.” Lindee’s interest is in how notions from popular culture influence technical conclusions and the establishment of facts in the culture of medical science (2006: 7).

⁵ In Thompson’s schema, specialization of tasks refers to work specificity, i.e., “making activities

more specific” (1961:25). By contrast the specialization of people refers to the adaptation of the individual to environmental circumstances.

⁶ Norma Ford Walker passed away in 1968. Much of the background information concerning Walker was provided to me in personal interviews and subsequent communications with her students.

⁷ Born in Corbeil, Ontario in 1934 the sisters went on to become a sensation of the Canadian depression-era.

⁸ The September 1954 membership list is appended to the secretary’s report and published in *American Journal of Human Genetics*, 7 (1955), pp. 466-95.

⁹ A significant proportion of the material that follows in this section comes from interviews with people who were involved in the process of establishing the Canadian College of Medical Geneticists. I am indebted to Dr. Hubert C. Soltan for his review of what I have written elsewhere on this subject. I am also indebted to Peggy Souter and Jean McQuilliam of the Membership Section of the Royal College of Physicians and Surgeons (Canada) for their assistance.

¹⁰ A final organization, the Genetic Nurses and Social Worker’s Association, was also created that year for allied health personnel working in the field. The four organizations eventually came together under an umbrella organization known as the British Society for Human Genetics, formed in 1996.

¹¹ See, for example, Medical Research Council/Department of Health and Social Security Joint Working Group on Genetics (1980), British Paediatric Association (1979), Medical Research

Council Sub-Committee on Genetics (1978), Department of Health and Social Security (1977:22-7; 1976a, 1976b, 1976c).

¹² The reader can make a comparison here with Peter Keating and Alberto Cambrosio's model of "biomedical platforms" which suggests "a distinctive place for technology and its development other than that of a simple tool for the furtherance of intellectual goals" (2003: 2). Keating and Alberto Cambrosio have described biomedical platforms as "material and discursive arrangements that act as the bench upon which conventions concerning the biological or normal are connected with conventions concerning the medical or pathological" (2003:4). What emerges from their model is a useful and flexible way to conceptualize medical specialization and the ability to transform the organization and structures of contemporary biomedical institutions and practices at both the national and local levels. Specialisms appear in this model in terms of overlapping organizing principles. They are not so much the result of turf wars among competing segments of service providers but, rather, products of mutual dependency and networked innovation (2003: 19-20; cf. Powell, Koput, and Smith-Doerr, and 1996; Scarbrough and Swan, 2005; Shinn, 2005).