

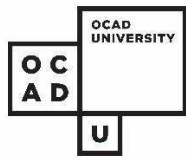
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The Early History of Medical Genetics in Canada

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Pre-Publication Draft

**The Early History of
Medical Genetics in Canada**

Abstract: This article shows that the intellectual and specialist movements that supported the growth of medical genetics in Canada between 1947 and 1990 were emergent phenomena, created, split, and reattached to different groups of actors, and reconfigured numerous times over the course of four decades. In each instance, new kinds of working relationships appeared; sets of diverse actors in local university-hospital settings coalesced into a new collectivity; and, as a collectivity, actors defined and/or redefined occupational roles and work rules. In its beginnings, medical genetics appears to be the object of a serious institutional manoeuvre: a movement in support of the creation of examining and teaching positions in human genetics in North American medical schools. With time, the institutionalization of ‘medical genetics’ took hold, spurred on by changes in the rate and direction of service delivery associated with genetic consultation and laboratory services in clinical settings. Medical genetics in Canada consequently gained a semblance of unanimity over its basic reference points and arrived at a meaning directly tributary to current acceptance of the term.

Keywords: Medical genetics, human genetics, medical specialism, hereditary disease, Canadian College of Medical Geneticists

The history of genetics in medicine has mainly been examined in relation to eugenics in the first half of the twentieth century and to molecular biology in the final third. With regard to the former, the history of eugenics in Europe and North America has been well traversed by historians, and they have rightfully been critical of eugenic science and physician involvement in campaigns to sterilize, euthanise and otherwise breed out what were perceived to be ‘unfit’ or ‘degenerate’ elements in populations.¹ In the latter case, by contrast, historians of molecular biology have produced excellent analyses of the discovery of DNA and the significance of subsequent efforts to map disease-causing genes.² But what has been left out of the picture is the enormous amount of work that has gone into organising ‘medical genetics’.³ Accordingly, the focus of this article is on ways genetic techniques have been taken up in mainstream medicine and the circumstances under which the clinicians and scientists involved set about turning their work into a medical specialism.

Specifically, this article provides an overview of the history of medical genetics in Canada from the opening of the first genetic counselling clinic in 1947 to the recognition by the Royal College of Physicians and Surgeons (Canada) of medical genetics as a free-standing medical specialty in 1989. I argue that the case of medical genetics in Canada lends insight into the beguiling complementarity of contemporary genetic medicine that promotes, simultaneously, the need for a collective adjustment of the intuitions and principles of all health care providers to incorporate a genetic approach to medicine in their clinical practices and a new medical specialism (i.e., medical

genetics). The particular circumstances of the Canadian situation, I submit, throw highlight on the convoluted and circuitous inroads of genetics into medicine, from the early days of Mendelian-based approaches to explaining patterns of hereditary and familial disease to the contemporary networks of genetic care which span multiple areas of service delivery. It is noteworthy that Canada was among the first nations to develop multidisciplinary regional genetics centres with genetic counsellors, laboratory scientists and academic colleagues. Likewise, 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. The College, established in 1976, preceded similar developments in the Netherlands, the United States, Finland, Sweden, Germany, France, and Denmark.

I begin the article by providing a brief background for understanding the relationship between genetics and North American medicine. This includes an examination of how Canadian and American geneticists created an expert role for themselves in the universities and medical schools. By circa 1930, I note, geneticists were increasingly finding work as researchers and science professors in departments of zoology, botany, or biology. Discussions followed in the 1940s and 1950s about a role for geneticists in clinical medicine performing teaching, research, and service functions. These discussions, I suggest, provided a framework for an understanding of medical genetics as applied human genetics. I then go on in the main section of the paper to show how Canadian geneticists, in a new role as experts, set out in the 1970s to establish standards of service delivery for multi-disciplinary centres providing genetic services. Services were assessed and configured to fit local health service requirements, most notably through the new policies and structures of the Canadian College of Medical Geneticists.

The Medico-Scientific Background of Medical Genetics in North America

As early as 1814, in his *Treatise on the Supposed Hereditary Properties of Diseases*, the English physician Joseph Adams had accurately – from a genetics perspective – distinguished between familial diseases ‘confined to a single generation, to brothers and sisters, the children of the same parent’ and hereditary diseases which are ‘traced from generation to generation’.⁴ In addition, he identified congenital illness as ‘disease appearing at birth’, noting that such conditions are more frequently familial rather than hereditary. Causal explanations for these phenomena, nevertheless, were not available until the next century. It was not until the early twentieth century that genetic theories, based on experimental and statistical examination of the reappearance of visible differences between generations of individuals, were sufficiently mature to provide such explanations.

The available evidence suggests that although the study of genetics gained 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 Britain and France.⁵ It was indeed the case that, although older life science programmes at American universities were dominated by traditional natural history foci, newer programmes were established after 1890 that were more likely to promote experimental research.⁶ New fields, such as genetics, which happened to be emerging in the midst of such growth, obtained the widest possible support by concentrating on the development of improved forms of pedigreed plants and animals.⁷ Little distinction was made in this context between what was then called ‘genetics’ and ‘practical breeding’.

Experimental breeding initiatives started in the U.S. Department of Agriculture, state experimental stations, and under other public and private sponsorship.⁸ Certainly, before World War I, the agricultural connection provided geneticists with an institutional setting in which geneticists

could meet quasi-professionally and publish.⁹ Furthermore, the rapid expansion of agricultural research meant the supply of candidates in relevant applied sciences was insufficient to meet demand, and experimental station administrations began to hire individuals in the basic sciences – including genetics.¹⁰ After 1915, genetics in the United States ‘began to take the form of a sanctioned normative practice with its own well-defined methods and explanatory standards’.¹¹

Geneticists, nonetheless, worked predominantly in departments of zoology, botany, or biology, rather than departments of genetics or plant breeding.¹² And interest in human genetics mainly collected on the periphery of eugenics and ‘practical breeding’, limited to a very small group of enthusiasts. Daniel J. Kevles indicates 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.¹³ The situation changed noticeably after World War II. Formal positions for ‘human’ geneticists were set up and implemented in twenty-nine multidisciplinary centres in the United States (twenty-five) and Canada (four).¹⁴ Ohio State University in 1933 had been the first site to require a course in human genetics as part of the medical school curriculum. Other centres doing research in human genetics followed in the 1940s: Bowman Gray School of Medicine (1940), University of Michigan (1941), University of Minnesota (1943), and University of Utah (1945). In addition, the Universities of Chicago, California, Oklahoma, Texas, and Toronto became centres for instruction, as well as Johns Hopkins, Duke, and Tulane. The data collected for the present study support earlier findings that many of the individuals involved in human genetics instruction in these centres not only voiced support for eugenics policies but also drew on eugenics societies and associations for resources for attendant ‘heredity counselling’.¹⁵ Counselling in this context originally referred to a loosely conceived set of activities associated with marriage and reproductive

advice.

Very little is actually known about early attempts at human genetics instruction in North America. What we do know is that the Association of American Medical Colleges sponsored the first organised efforts to gather information on the extent of genetics instruction in American and Canadian medical schools. This began in 1946 with a survey that involved mailing questionnaires to eighty-four medical schools in the United States and Canada.¹⁶ It was found that seven schools provided formal courses in genetics, with an average of fifteen class hours of instruction. Lectures on genetics were given as part of other courses in twenty-five schools, with an average of five hours of classroom instruction. A second questionnaire survey, administered in 1953 by C. Nash Herndon, then director of the Department of Medical Genetics at Bowman-Gray School of Medicine, concluded that ‘there is no unanimity at present as to where genetic instruction really belongs in the medical curriculum’.¹⁷ By contrast, a 1954 report, based on a workshop of American and Canadian instructors, found unexpectedly high levels of support for genetics instruction in medical schools.¹⁸ The report represents the first attempt to produce a blueprint for the future of medical genetics. Moreover, discussion generated by the recommendations of the report appears in a number of later surveys and reviews on the subject.¹⁹ Its discussions of ‘integrated curriculum’ and a role for geneticists in medical schools were certainly the first of their kind, and merit close examination.

First of all, clinical 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’.²⁰ Second, and concurrently, ‘genetics ... serves as a useful tool in the prevention of disease and limitation of disability’.²¹ Turning to the question of what to teach, the report recommends five fundamental areas of study for consideration: the physical basis for heredity (i.e., chromosomes); 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'.

Furthermore, 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.²²

The medical geneticist 'could also have service and research functions'.²³ Thus, a multi-faceted role was envisioned. As a 'staff geneticist' in teaching hospital settings, s/he 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.

As noted above, the recommendations of the 1954 AAMC report enjoyed considerable support and further discussion in subsequent surveys and reviews. Comparatively speaking, five surveys 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 1985.²⁴ However, three surveys indicate that, rather than an integrated curriculum, genetics instruction increasingly became the preserve of paediatricians and geneticists teaching in independent genetics departments. This issue 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’.²⁵ From this emerged what I have called elsewhere a bifurcated ideological construct²⁶ that shaped and informed the means of organising a ‘genetics-based approach’ to medicine.²⁷ 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, a consultant working in the Neurofibromatosis Program at Baylor College of Medicine, professed:

... 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.²⁸ (Emphasis in the original)

Medical Genetics: The Canadian Case²⁹

The beginnings of medical genetics in Canada have been reported by others in relation to the coinage of the field’s name in 1932 by Madge Thurlow Macklin, then at the University of Western Ontario,³⁰ and in the formation of the first Canadian heredity counselling clinic at the (Toronto) Hospital for Sick Children in 1947 by Alan Brown and Norma Ford Walker.³¹ The institutionalisation of medical genetics as a medical specialism in Canada, however, has not been examined.

Fiona Miller’s study of early developments in Toronto has thrown highlight on Norma Ford Walker’s accomplishments in the field of dermatoglyphic pattern analysis.³² 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.³³ But what is problematic here is Miller's claim that Walker's research was 'marginal' and that she was involved in establishing some kind of a 'school' of medical genetics. In actuality, Norma Ford Walker had little interest in medicine per se.³⁴ 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.³⁵

Contrary to Miller's characterisation of this work as 'iconoclastic genetics' and 'marginal', the study of the quintuplets completed in 1937 was a significant contribution to contemporary studies of 'mental likeness' in twins and intelligence testing.³⁶ 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.³⁷ 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.³⁸

Norma Ford Walker's subsequent appointment as a genetics consultant in 1940 by Alan Brown, chief of paediatrics at the (Toronto) Hospital for Sick Children, entailed a kind of 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, Louis Siminovitch, appointed Geneticist-in-Chief to the Hospital for Sick Children in 1968, encountered a

fragmented set of service operations that had failed to keep pace with other multidisciplinary centres in North America.³⁹ In actuality, a fully integrated Division of Clinical Genetics, jointly responsible to the Department of Genetics and the Department of Paediatrics, did not appear until 1986.

Cumulative links between genetics and specialised areas of medical research and services can be studied more easily in developments at McGill University in Montreal. The interwar years saw well-known figures such as the English hereditarian H. B. Fantam as McGill's Strathcona 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 staff consisting of Fraser and an assistant. Fraser recalled the casual attitude shown toward counselling services in an interview:

When I started genetic counselling, it was essentially a physician calling you up and saying: 'I've got this mother who has a haemophiliac baby. What's the chance of recurrence?' And I would look it up and tell them. Then I began to see families in person, take family histories, and do the counselling myself.... I used to walk down the ward and look down the list of patients and see if anything genetically interesting was on the ward and then quiz the residents on recurrence risks. And

gradually, I got the reputation for being useful, now and then. And so genetics began gradually to be recognized around the hospital. But this was very new for Canada.⁴⁰

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 presently provide some combination of medical genetics training and services, 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 (four MDs, one PhD, one PhD/MD). 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'. 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 and Daniel Kevles 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'.⁴¹ 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.⁴² Canadian geneticists, by contrast, did not enjoy this kind of support during this period. As noted above, the

genetics department at McGill had received some start-up funding from the Rockefeller Foundation. But this was the exception.

As Angus McLaren has shown, the establishment of eugenics institutions like the Eugenics Society of Canada came about relatively late in comparison to developments in the United States.⁴³ Eugenics policies were successfully enacted in the provinces of British Columbia and Ontario in favour of segregating ‘mentally defective’ school children.⁴⁴ Eugenics sterilization laws were enacted in Alberta and British Columbia.⁴⁵ But the physicians who were active in the eugenics programmes came from the field of public health – not genetics research or clinical medicine.⁴⁶ Early financial support for genetic counselling services came through hospital budgets or was divested from funds earmarked for research.⁴⁷

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. The early meetings were tiny and, initially, it was difficult to attract articles to publish in its journal, the *American Journal of Human Genetics*. But within five years the membership had grown to 565 individuals with 316 institutional subscribers to the journal.⁴⁸ 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.

From the perspective of the present study, the American Society of Human Genetics is

especially important for the ideological stance it took. The first president, Hermann J. Muller, a Nobel prize laureate at Indiana University well known for his discovery of artificially induced mutations in genes, observed:

Included in the ranks of our present group there are many persons of genetic competence who are primarily medical men, of varied specialties, there are some genetically qualified anthropologists, psychologists and students of social sciences, and there is a good share of persons whose main field is genetics itself but who have acquired a considerable interest in and knowledge of one or more of the specifically human subjects. Our board of directors and our editorial staff have been chosen so as to represent all these groups. We hope to therefore be able to avoid that dilettantism which has in the past characterized so many attempts to study human hereditary. . . .

. . . It happens that the prolonged delay in setting up the present working association has one very fortunate aspect. This derives from the fact that, until very recent years, the subject of human heredity was buffeted about by pressure groups from the extreme political right and left, who sought to impose their social preconceptions in the form of a spurious ‘nature-nurture controversy’, in which the methods of objective science were largely forgotten. The development of a more scientifically minded group of students of the subject has required the influence of basic genetics, working over many years. And in recent years, this sounder attitude has been reinforced by the lessons of the terrible mistakes made by the political protagonists of fascism and communism, respectively, when they gained the power to translate their biological prejudices into action.⁴⁹

These observations reflect turning points in the development of both human genetics and medical genetics. The geneticists, who now had the sense of security that comes with permanent university posts, became increasingly vocal in their disdain for the pseudo-science of amateurs – what Muller was calling ‘that dilettantism.’⁵⁰

A wholesale shift in the meaning of ‘eugenics’ followed along the lines of certain irreconcilable differences 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 counselling’; 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’.⁵¹

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), would emphasize a ‘scientific basis for the concepts of individuality and variation within the species.’⁵² ‘Pathologic genes’ were themselves to be viewed as ‘etiologic agents of disease’; agents that, with time, would account for an increasing proportion of the clinician’s workload. Hence, James V. Neel, director of the Heredity Clinic at the University of Michigan, predicted:

In the year 1900, pneumonia, influenza, tuberculosis, diarrhea, diphtheria and typhoid fever accounted for 610.9 deaths among each 100,000 inhabitants of the United States. In the year 1947 ... these same diseases were responsible for only 78.0 deaths per 100,000 population. It may confidently be anticipated that in another ten years, when the full impact of the recent rain of antibiotics has been realized, those diseases will account for no more than 30 to 40 deaths per 100,000. ...

... The result of this rapid disappearance of the infectious and contagious diseases from the medical scene has, of course, been, and to an increasing extent will be, the direction of medical attention towards conditions variously and overlappingly termed constitutional, endocrine, metabolic, or congenital. It is in the etiology of just these conditions that the role of genetic factors is most evident. A working knowledge of the elementary principles of genetics is thus in many instances indispensable to the formulation of a well rounded picture of a given disease.⁵³

Additionally, non-directive genetic counselling would conform to evolving medico-legal norms that served to increase patient autonomy over physician beneficence. There were ongoing debates about physicians’ freedoms of decision-making in the years following the revelations of Nazi atrocities at the Nuremberg Trials.⁵⁴ These freedoms, many argued, should be governed by a fiduciary duty of respect for patient autonomy that stressed non-interference. A principle emerged that ‘every human being of adult years and sound mind has the right to determine what shall be done with his own body’. Genetic counselling was, consequently, 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 standardise genetic counselling services was subsequently 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 and 1970s. 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.⁵⁵ 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 by Jérôme Lejeune, Marthe Gauthier and Raymond Turpin in 1959.⁵⁶ This was quickly followed by reports from cytological laboratories in England.⁵⁷ The publication of these findings in quick succession during 1959 caused a sensation among scientists and clinicians alike. The development of human cytological genetics or 'cytogenetics' provided clinical tools to uncover the genetical make-up of relatively common disorders. And it is important here to stress the novelty of cytogenetics during this period. Chromosomal abnormalities were mostly of unknown aetiology and this was a time of uncertainty concerning the precise relationship of congenital and hereditary disease. What mattered was something new: the chromosomal bases of diseases.

While the Europeans were working out techniques to count human chromosomes, Murray Barr and Canadian colleagues at the University of Western Ontario were studying sexual dimorphism at the cellular level by observing and describing the sex chromatin body in the neuronal nuclei of female cats and its absence in male cats.⁵⁸ The clinical possibilities of Barr's cytogenetic laboratory

were pursued by Howard Valentine, a professor in the Department of Paediatrics in the late 1950s.

He recalled in an interview:

When the sex chromatin business broke, I kind of heard about it, but that didn't particularly interest me because the sex chromosomes weren't really all that connected with paediatric disorders. At least they weren't recognized as such at that time. Turner syndrome and the XO sex chromosome complement was kind of recognized as a clinical entity among the paediatricians ... but in general it wasn't until Down's syndrome was recognized as being a chromosomal abnormality that I really became interested in cytogenetics. And then I read Murray Barr's work and I got interested in further developments of cytogenetics because of Down's syndrome, which was an interest of mine even before cytogenetics was invented. I had been puzzled and interested in Down's syndrome. And then when Down's syndrome broke, and it was recognized that this was a cytogenetic abnormality, then that fired up my interest. Then I teamed up with Fred Sergovich, who ... was a graduate student of Murray Barr's, who went to work on the cytogenetics of Down syndrome.⁵⁹

Meanwhile, interest in cytogenetics in Quebec was stimulated by the discoveries of Jérôme Lejeune and colleagues in France. Lejeune announced the discovery of trisomy 21 and Down's syndrome at a seminar in the McGill University genetics department in 1958, following 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 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).

The problem of transporting diagnostic test samples from disparate locations in the Canadian provinces presented a huge challenge for geneticists running laboratories in hospitals that were largely situated close to the American border.⁶⁰ Plant cytologists and other technical staff were recruited to perform a service function in chromosome laboratories,⁶¹ and a new occupational category was created, 'cytogeneticists'.⁶¹ A laboratory testing protocol was devised whereby physicians communicated by telephone suspected cases of chromosomal abnormality to either staff geneticists or cytogeneticists in regional centres. If it appeared that a chromosomal analysis was in order, a test kit was dispatched, usually by bus, to the consulting physician. The physician drew blood from the patient, mixed test samples with the culture media supplied, and then shipped everything back to the laboratory. A cytogeneticist's report was then prepared for the consulting physician.

Biochemical testing, a parallel development to chromosome analysis, involves biochemical demonstration of abnormal metabolites in body fluids. Biochemical analysis is useful in cases where the products (i.e., end results) of an abnormal gene are present. Monogenetic gene mutations may either block the synthesis of certain enzymes or lead to the production of enzymes with abnormal structures. In either situation, metabolic processes are disrupted. 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'.⁶² Physicians would look for tell-tale signs and symptoms (e.g., failure to thrive, developmental delay, ocular abnormalities) that might be

indicative of a 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 chemical 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.⁶³ 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.⁶⁴ Between 1963 and 1969, newborn screening programs were subsequently set up in nine provinces largely administered through provincial public health programmes. The first was established in 1963 in Prince Edward Island. Programs followed in British Columbia in 1964; Saskatchewan, Manitoba, and Ontario in 1965; New Brunswick and Nova Scotia in 1966; and Alberta and Quebec in 1969. Only in Quebec did geneticists take a proactive role in the organization of a province-wide program.

Le Réseau de la Médecine Génétique du Québec was formed in October 1969 on the recommendation of four heads of the paediatrics departments at the Centre Hospitalier Universitaire de Laval, Le Centre Hospitalier Universitaire de Sherbrooke, the Montreal Children's Hospital and l'Hôpital Ste-Justine. The network began receiving provincial funding that year, and incorporated itself as Le Réseau Provincial de la Médecine Génétique du Québec in 1971. The mandate of the Réseau, firstly, was to develop a centralized program for the early

detection of metabolic diseases in newborns. It did so by coordinating resources in the four hospitals. Additionally, it held a mandate to do research and development in the area of genetic services. By the mid-1970s, the Réseau provided diagnostic services, counselling and treatment for approximately thirty metabolic disorders.

Screening programs in the other provinces emerged as independent provincial ministry of health programs.⁶⁵ But, despite the fact that screening came under the purview of public health branches and departments of health and community services, the genetics centres provided the laboratory and counselling services. A service protocol was devised in which a staff member of either a public health unit or a genetics clinic would review the hospital lists of newborns and ensure that blood samples were taken. An attending physician receiving a positive test report had the option of either counselling the family him- or herself, or utilizing the services of the regional genetics centre.

Further to the new diagnostic test regimes of the 1960s, geneticists experienced increases in workload after 1970 with the work associated with prenatal diagnosis. Refined techniques for culturing foetal cells from amniotic fluid were developed and amniocentesis became useful as an outpatient procedure for obtaining test samples for chromosomal and biochemical analysis. Obstetricians performed all the amniocentesis procedures. This involved removing amniotic fluid during early pregnancy by puncturing the amniotic sac with an aspiration needle – a procedure with risks to the mother, including bleeding and infections, and to the foetus, including needle puncture, premature labour, and, potentially, spontaneous abortion. If the amniocentesis was performed in a genetics centre, 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. Moreover, 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 services was mounted in the early 1970s when geneticists realised 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 accreditation of individuals offering genetic counselling. 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.⁶⁶ 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, 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 services.⁶⁷ 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 restrictions surrounding who and who could not provide patient care. What emerged from all this was a coalition to form a corporation to be known as the Canadian College of Medical Geneticists.⁶⁸

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. The proposal was accepted, and a steering committee was elected to make an application for incorporation and draw up a constitution and by-laws for the new College. A statement from the Steering Committee 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’.⁶⁹ 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.⁷⁰

The first meeting of the newly-formed Board of Directors took place at the Club Saint-Denis

in Montreal on November 3, 1975. 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.⁷¹ 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.

Before turning to the substance of the negotiations surrounding specialty formation, some background about the Royal College is necessary. Incorporated by Act of Parliament and receiving Royal Assent in June of 1929, the Royal College has had no medical licensing powers per se.⁷² An official invitation for the Royal College to establish the certification of specialists was received from the Canadian Medical Association in 1937, and a joint committee of the Association and Royal College was formed. Subsequent relations between the Royal College and would-be specialists would focus on, first, determining requirements and standards of training, and, second, conducting examinations for specialist certification. A committee structure was created comprised of sub-committees made up of individuals representing independent specialties or sub-specialties.

Between 1930 and 1947, the number of specialties that the Royal College recognized increased from two to twenty-one.⁷³ There then followed a fifteen-year period in which further recognition was not granted. A second wave of specialty formation occurred between 1964 and 1974,

with a total of thirteen new specialist sub-committees being admitted to the Royal College's Division of Medicine. Concerned about the rapid proliferation of specialties, the Council of the Royal College acted in 1974 on a recommendation to declare a moratorium on the recognition of new specialties – a course of action that had been taken earlier by the American Board of Medical Specialties under similar circumstances. New policies followed with, on the one hand, a set of guidelines for defining specialty areas, and, on the other, a new Certificate of Special Competence to signify recognition of a sub-specialty subordinate to certification in a primary specialty. With regard to the latter, it was suggested that the Certificate might obviate the need to recognize new specialties and alleviate concerns about the fragmentation of the medical profession.

The moratorium was in effect lifted in January 1975 when Council approved the recognition of paediatric general surgery as a specialty by means of a Certificate of Special Competence. Primary certification soon followed in geriatric medicine, nephrology, thoracic surgery, and perinatal medicine. Partial recognition (i.e., a certificate of special competence in medical genetics) was granted in 1979 to medical genetics, but owing to differences with the Canadian College of Medical Geneticists, the Royal College suspended the implementation of the recognition.

As noted above, the Canadian College of Medical Geneticists had made an application for recognition as a specialty in 1976. Correspondence, initiated by the Board of Directors, was referred to the Royal College's Committee on Specialty Development, and informal contacts were made. Meetings followed between the Committee and members of the Canadian College of Medical Geneticists over a three-year period to explore the possibility of a certificate of special competence or, alternatively, a free-standing specialty in medical genetics. Based on what interviewees recalled about the tone of the negotiations, it appears that the Royal College's Committee on Specialty

Development followed closely the 1975 guidelines on specialty recognition. In May of 1978 the geneticists provided to the Committee on Specialty Development a brief consisting of reasons why medical genetics constituted a separate body of medical knowledge; a proposal for specialty training in medical genetics; and up-dated lists of the names, credentials and places of work of College fellows, Board directors, and members of the six standing committees of the College. In addition, the brief specified that medical genetics had already been recognized as a medical specialty in the Province of Saskatchewan and that the majority of all medical geneticists currently practicing and teaching in Canada were members of the College. Further to this, it indicated that the American College of Medical Geneticists currently in formation was modelled on the Canadian College of Medical Geneticists.

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. These groups were generally unresponsive, but at another level, the College was successful. By the early 1980s, interviewees for the present study recalled a substantial portion of postings for medical genetics positions in Canada requiring prospective candidates to have either passed or be eligible to take the College's written and oral examinations.

It was 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. 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.

Of the sixteen Canadian universities with faculties of medicine, seven offered Royal College accredited residency training programs in medical genetics, and seven offered training programs accredited by the Canadian College of Medical Geneticists. Within the overall range of programs, those designated 'medical genetics' with Royal College accreditation were separable from programs in 'clinical genetics', 'cytogenetics', 'biochemical genetics', and 'molecular genetics' accredited by the Canadian College of Medical Geneticists. Additionally, the Canadian College of Medical Geneticists issued certificates in four other categories to successful applicants with MD or PhD degrees: medical genetics, cytogenetics, biochemical genetics, and molecular genetics. In practical terms, this meant that holders of MD degrees could either apply for certification as medical genetics specialists after completing a defined period of specialty residency in a program recognized by the Royal College, or, after obtaining certification as specialists in another area of practice recognized by the Royal College, apply for certification as clinical geneticists recognized by the Canadian College of Medical Geneticists. Holders of PhD degrees could only apply for Canadian College of Medical Geneticists certification. Ten years after specialty recognition, only two clinical geneticists

were employed in provincial genetics centres with just Royal College training in medical genetics. At the same time, a healthy percentage (forty four per cent) of the MD-geneticists was certified by both bodies, indicating a relatively high level of acceptance in the field for the RCPSC medical geneticist category. The other fifty three per cent of clinicians providing counselling and consultation in genetics centres, by contrast, were made up of individuals who had entered the field prior to 1989.

As a final point, it is worth noting that the role of MD-geneticists evolved in relation to the roles of other medical specialists. In brief, two broad sets of activities can be identified. The first set falls under a general category of prenatal care in pregnancy and childbirth, and overlaps with the services of obstetrics and gynaecology. Activities in this set continue to be referred to by their function: ‘prenatal diagnosis’. This can be distinguished from what is called ‘general genetics’. General genetics is a catch-all category for activities involving infants, children and adults. As a set of activities unto itself, it can be further divided into three subsets. Activities in the first subset overlap with the area of neonatology. This involves the diagnosis and management of congenital anomalies and diseases in newborns. The second subset takes up broader paediatric concerns and focuses on the diagnosis and management of disorders in children. Finally, the third subset deals with adult-onset diseases and screening for carriers of heritable conditions. In this regard, the nature of the interface with other specialists shifts paradigmatically depending on whether the patient is a pregnant woman, an infant, a child, or an adult.

Concluding Observations

The history of Canadian medical geneticists outlined in this article describes, first, early efforts in collaboration with American geneticists to create examining and teaching positions in medical schools for human genetics instruction and, secondly, independent efforts to create a new medical

specialty. With regard to the former, early discussions of an ‘integrated curriculum’ and a role for geneticists in medicine envisioned teaching, research, and service functions. In the 1940s and 1950s, the role of the medical geneticist emphasized teaching and research tasks in medical academia. Referral for genetic consultation was based largely on personal reputations for specialized knowledge and scientific expertise. And medical faculties in universities served as centres where new skills could be developed and knowledge and information exchanged.

Circumstances changed in the 1960s and 1970s. The clinical possibilities of technological advances in the form of new laboratory technologies for identifying chromosomal anomalies and genetic metabolic disease were quickly recognized. Demand for genetic advisory services increased, and the geneticists, in turn, moved to establish standards of practice and lay the groundwork for the division of labour we see in Canadian genetics centres today. This last aspect is especially important as regards what it now means to be a medical geneticist. The principal selective mechanism to maintain the collective identity of geneticists as medical specialists is the division of labour, which at once differentiates them from other specialized workers in medicine (e.g., laboratory workers, scientists, other medical specialists).

However, in addition, significant questions can be raised for future consideration and historical inquiry. The material presented in this article shows how occupational specialization 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. So, how will medical genetics as a free-standing medical specialty maintain itself as a specialty area? Are there sufficient resources and infrastructure to encourage an ongoing sense of collective identity of medical

geneticists as medical specialists? And, should there occur a lack of supports internal to medical genetics to encourage stability and foster growth, will there be external factors to do so, e.g., monetary gains, collegial approbation, patient demand, public policies?

Additionally, we are seeing more and more diagnostic tests as a result of the ‘genomics revolution’ that are adding to the clinician’s ‘toolbox’. Increasingly it is the case that clinicians who hold specialist qualifications in a specialty outside of medical genetics are presenting themselves as experts on the genetics of a particular illness or illness group. Might the activities of these other medical specialists, then, reduce demand for the services of medical geneticists? Can we say that the relationship between genetics and medicine is evolving and unfolding over time in a variety of clinical settings? If so, there then follows yet another question: How will specialists – medical geneticists and otherwise – keep (or be kept) abreast of scientific developments in genetics? Moreover, can any specialist afford not to keep abreast of these developments?

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critical comments on an earlier draft of the manuscript. Finally, I warmly thank John V. Pickstone for his input on a later draft of the manuscript.

Notes

¹ See, in particular, D. B. Paul, *The Politics of Heredity: Essays on Eugenics, Biomedicine, and the Nature-Nurture Debate* (New York, 1998); A. McLaren, *Our Own Master Race: Eugenics in Canada 1885-1945* (Toronto, 1990); L. Farrall, *The Origins and Growth of the English Eugenics Movement 1865-1925* (New York, 1985); D. J. Kevles, *In the Name of Eugenics: Genetics and Uses of Human Heredity*, (Berkeley, 1985).

² See, for example, R. Cook-Degan, *Gene Quest: Science, Politics, and the Human Genome Project* (New York, 1993); D. J. Kevles and L. Hood (eds), *The Code of Codes* (Cambridge, 1992); T. F. Lee, *The Human Genome Project: Cracking the Genetic Code of Life* (New York, 1991); N. A. Holtzman, *Proceed with Caution: Predicting Genetic Risks in the Recombinant DNA Era* (Baltimore, 1989).

³ R. G. Resta and D. Paul, 'Introduction: Historical Aspects of Medical Genetics', *American Journal of the Medical Genetics*, 115 (2002), 73-4.

⁴ J. Adams, *A Treatise on the Supposed Hereditary Properties of Diseases, Containing Remarks on the Unfounded Terrors and Ill-Judged Cautions Consequent on Such Erroneous Opinions; with Notes, Illustrative of the Subject, Particularly in Madness and Scrofula* (London, 1814).

⁵ See, for example, G. Allen, *Thomas Hunt Morgan: The Man and His Science* (Princeton, 1978), pp. 278-83.

⁶ J. Harwood, *Styles of Scientific Thought: The German Genetics Community, 1900-1933* (Chicago, 1993), pp. 143-56.

⁷ For an extensive discussion of the subject, see H. Cravens, *The Triumph of Evolution: American Scientists and the Heredity-Environment Controversy* (Philadelphia, 1978).

⁸ Cravens, *The Triumph of Evolution*, pp. 27-37; A. H. Hunter, *Science in the Federal Government: A History of Policies and Activities to 1940* (Cambridge, 1957).

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¹⁰ M. Rossiter, 'The Organization of the Agricultural Sciences', in A. Oleson and J. Voss (eds.), *The Organization of Knowledge in Modern America, 1860-1920* (Baltimore, 1979), pp. 211-48.

¹¹ J. Sapp, 'The Struggle for Authority in the Field of Heredity, 1900-1932: New Perspectives on the Rise of Genetics', *Journal of the History of Biology*, 16 (1983), 311-42.

¹² Harwood, *Styles of Scientific Thought*, p.156.

¹³ Kevles, *In the Name of Eugenics*, p. 205.

¹⁴ By comparison, it is noteworthy that there was only one centre in England involved in similar activities during this period.

¹⁵ See, in particular, I. R. Dowbiggin, *Keeping America Sane: Psychiatry and Eugenics in the United States and Canada, 1880-1940* (Ithaca, 1997); Paul, *The Politics of Heredity*, pp. 133-56; Kevles, *In the Name of Eugenics*, pp. 193-211.

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¹⁷ C. Herndon, 'Human Genetics and Medical Education', *Journal of Medical Education*, 29 (1954), 13-19.

¹⁸ Subcommittee on Objectives of Teaching, Association of American Medical Colleges, 'The Teaching of Pathology, Microbiology, Immunology, Genetics', *Journal of Medical Education*, 30 (Suppl., Sept. 1955), 1-23.

¹⁹ J. V. Neel, 'The Functions of a Department of Genetics in a Medical School', *Journal of Medical Education*, 3 (1957), 493-500; F. Hickman, 'Comprehensive Planning of Human Genetics Education: A Position Statement', *American Journal of Medical Genetics*, 8 (1981), 243-7; B. Childs, C. A. Huether and E. A. Murphy, 'Human Genetics Teaching in U.S. Medical Schools', *American Journal of Human Genetics*, 33 (1981), 1-10; B. Childs, 'Genetics in the Medical Curriculum', *American Journal of Medical Genetics*, 13 (1982), 319-24; R. G. Davidson and B. Childs, 'Perspectives in the Teaching of Human Genetics', *Advances in Human Genetics*, 16 (1987), 79-119; V. M. Riccardi and R. D. Schmickel, 'Human Genetics as a Component of Medical School Curricula: A Report to The American Society of Human Genetics', *American Journal of Human Genetics*, 42 (1987), 639-43; J. M. Graham, et. al., 'Report of the Task Force on Teaching Human Genetics in North American Medical Schools', *American Journal of Human Genetics*, 44 (1989), 161-5.

²⁰ Subcommittee on Objectives of Teaching, 'The Teaching of Pathology, Microbiology, Immunology, Genetics', p. 19.

²¹ *Ibid.*, p. 19.

²² *Ibid.*, p. 21.

²³ *Ibid.*, p. 21-2.

²⁴ M. D. Levine, J. M. Gursky and D. L. Rimoin, 'Results of a Survey on the Teaching of Medical Genetics to Medical Students, House Officers, and Fellows', in H. A. Lubs and F. de la

Cruz (eds), *Genetic Counseling* (New York, 1977), pp. 349-68; Robertson and Haley 'Genetics in the Medical Curriculum', pp. 351-3; Herndon, 'Human Genetics and Medical Education', pp. 13-19; Childs, Huether and Murphy, 'Human Genetics Teaching in U.S. Medical Schools', pp. 1-10; Riccardi and Schmickel, 'Human Genetics as a Component of Medical School Curricula', pp. 639-43.

²⁵ A. G. Motulsky, 'Postdoctoral Education and Training in Medical Genetics', in I. H. Porter and E. B. Hook (eds), *Service and Education in Medical Genetics*, (New York, San Francisco, London, 1979), 409-18; Hickman, 'Comprehensive Planning of Human Genetics Education: A Position Statement', p. 246.

²⁶ Following Robbins and Johnston, 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. See D. Robbins and R. Johnston, 'The Role of Cognitive and Occupational Differentiation in Scientific Controversies', *Social Studies of Science* 6 (1976), 349-68.

²⁷ Leeming, 'Professionalization Theory, Medical Specialists and the Concept of "National Patterns of Specialization"', *Social Science Information* 40 (2001), 455-85.

²⁸ V. M. Riccardi, *The Genetic Approach to Human Disease* (New York, 1977), pp. 2-3.

²⁹ Two kinds of data are used in the study of Canadian medical geneticists that follows. Preparatory research was based on the analysis of primary and secondary source material drawn from a range of libraries. In addition to providing technical details and facts, this material furnished a source of general information about genetics and medicine against which to compare the responses forthcoming in interviews – the second source of data for this study. Open-ended

qualitative interviews were conducted between April 1997 and June 1999. This involved a sample of sixty-seven respondents. A conscious effort was made to include in the sample individuals delivering genetic services in each Canadian province.

³⁰ M. Thurlow Macklin, 'Medical Genetics: A Necessity to the Up-to-date Medical Curriculum', *Journal of Heredity* 23 (1932), 485-6. See, also, H. C. Soltan, 'Madge Macklin', in H. C. Soltan (ed.), *Medical Genetics in Canada: Evolution of a Hybrid Discipline* (London, 1992), p. 11; McLaren, *Our Own Master Race*, p. 137; Kevles, *In the Name of Eugenics*, p. 176.

³¹ F. Miller, 'The Importance of Being Marginal: Norma Ford Walker and a Canadian School of Medical Genetics', *American Journal of Medical Genetics*, 115 (2002), 102-10.

³² Miller, 'The Importance of Being Marginal', p. 102. Dermatoglyphics studies and systematically classifies the patterns of the ridged skin of the palm, fingers, soles and toes for purposes of investigating dysmorphological conditions.

³³ J. S. Thompson and M. W. Thompson, *Genetics in Medicine* (Philadelphia, 1966), pp. 245-6.

³⁴ Norma Ford Walker passed away in 1968. Much of the background information concerning Walker was provided to me in a personal interview (26 January 1999) and subsequent communications with her student Margaret W. Thompson.

³⁵ Born in Corbeil, Ontario in 1934 the sisters went on to become a sensation of the depression-era. See E. Teshler, *The Dionnes* (Toronto, 2000).

³⁶ W. E. Blatz, N. Chant, M. W. Charles, M. I. Fletcher, N. H. C. Ford, A. L. Harris, J. W. MacArthur, M. Mason and D. A. Millichamp, *Collected Studies on the Dionne Quintuplets* (Toronto, 1937).

³⁷ S. Jay Gould, *The Mismeasure of Man* (New York, 1981), pp. 234-320.

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- ³⁸ A. Gesell and H. Thompson, 'Learning and Growth in Identical Infant Twins: An Experimental Study by the Method of Co-Twin Control', *Genetic Psychology Monographs* 6 (1929), pp. 1-124.
- ³⁹ Personal interview (28 January 1999) with Louis Siminovitch.
- ⁴⁰ Telephone interview (19 January 1999) with F. Clarke Fraser.
- ⁴¹ Paul, *The Politics of Heredity*, p. 137; Kevles, *In the Name of Eugenics*, pp. 199, 208-9, 252-4.
- ⁴² On the Pioneer Fund, see B. Mehler, 'The New Eugenics: Academic Racism in the U.S. Today', *Science for the People*, 15 (1983), 18-23.
- ⁴³ McLaren, *Our Own Master Race*, pp. 112-15.
- ⁴⁴ McLaren, *Our Own Master Race*, pp. 91, 108.
- ⁴⁵ McLaren, *Our Own Master Race*, pp. 90-1, 99-106.
- ⁴⁶ McLaren, *Our Own Master Race*, p. 114.
- ⁴⁷ Details concerning the backgrounds of the six early centres offering genetic services at Toronto, Montreal, Edmonton, Vancouver, Saskatoon, and Halifax are available in my unpublished dissertation, *Medical Specialization and Medical Genetics in Canada*, pp. 69-102.
- ⁴⁸ The September 1954 membership list is appended to the secretary's report and published in *American Journal of Human Genetics*, 7 (1955), 466-95.
- ⁴⁹ H. J. Muller, 'Progress and Prospects in Human Genetics', *American Journal of Human Genetics*, 1 (1949), pp. 1-2.
- ⁵⁰ Eugenics historians have frequently drawn attention to the individual geneticists who began to separate themselves from what Kevles has called 'mainline eugenics' after the First World War. (See Kevles, *In the Name of Eugenics*, p. 122.) My interest here is in the collective adjustment of

intuitions and principles that followed the Second World War.

⁵¹ S. C. Reed, *Counseling in Medical Genetics* (Philadelphia, 1955), pp. 14-15.

⁵² Subcommittee on Objectives of Teaching, Association of American Medical Colleges, 'The Teaching of Pathology, Microbiology, Immunology, Genetics', p. 18.

⁵³ J. V. Neel, 'Some Applications of the Principles of Genetics to the Practice of Medicine', *Medical Clinics of North America* 35 (1951), p.519.

⁵⁴ J. G. Haber, 'Patients, Agents, and Informed Consent', *Journal of Health and Law* 1 (1985-86), 43-59; c.f. T. Szasz and J. Hollander, 'A Contribution to the Philosophy of Medicine – The Basic Models of the Doctor-Patient Relationship', *Archives of Internal Medicine* (1956), 585-7.

⁵⁵ M. J. D. White, *Animal Cytology and Evolution*, 2nd ed. (Cambridge, 1954); cf. C. P. Swanson, *Cytology and Cytogenetics* (New York, 1957).

⁵⁶ J. Lejeune, M. Gauthier and R. Turpin, 'Études des Chromosomes Somatiques de Neuf Enfants Mongoliens', *Comptes Rendus de l'Académie des Sciences* 248 (1959), 1721-2.

⁵⁷ C. E. Ford, K. W. Jones, O. J. Miller, U. Mittwoch, L. S. Penrose, M. Ridler and A Shapiro, 'The Chromosomes In A Patient Showing Both Mongolism And The Klinefelter Syndrome', *Lancet* 1 (1959), 709-10; C. E. Ford, P. E. Polani, J. H. Briggs and P. M. F. Bishop 'A Sex-Chromosome Anomaly In A Case Of Gonadal Dysgenesis (Turner's Syndrome)', *Lancet* 1 (1959), 711-13.

⁵⁸ M. L. Barr, 'Human Cytogenetics: Some Reminiscences', *Bio Essays* 9 (1988), 79-82.

⁵⁹ Telephone interview (8 February 1999) with Howard Valentine.

⁶⁰ K. L. Ying and J. W. Gerrard, 'A Service for Chromosome Studies in Saskatchewan', *Canadian Medical Association Journal* 94 (1966), 646-8.

⁶¹ I. A. Uchida and M. Ray, 'Mail-Order Chromosome Analysis', *Canadian Medical*

Association Journal 94 (1966), 649-50.

⁶² A. G. Bearn, 'The Contribution of Clinical Medicine to Biochemical Genetics', in H. K. Beecher (ed.), *Disease and the Advancement of Basic Science* (Cambridge, 1960), pp. 8-26.

⁶³ R. Guthrie and A. Susi, 'A simple phenylalanine method for detecting phenylketonuria in large populations of newborn infants', *Pediatrics* 32 (1963), 338-43; W. B. Hanley, J. T. R. Clarke and W. Schoonheydt, 'Maternal phenylketonuria (PKU): a review', *Clinical Biochemistry* 20 (1987), 149-56.

⁶⁴ A. M. O. Veale, 'Screening for Hereditary Metabolic Disorders', in H. Bickel, R. Guthrie, G. Hammersen (eds.), *Neonatal Screening for Inborn Errors of Metabolism* (Berlin, 1980), pp. 7-18.

⁶⁵ See J. C. Haworth, J. R. Miller, and C. R. Scriver, 'Screening, counselling and treatment of hereditary metabolic disease; a survey of resources in Canada', *Canadian Medical Association Journal* 111 (1974), 1147-53.

⁶⁶ J. R. Miller, 'Report of Genetic Counselling Services in Canada', *Genetics Society of Canada Bulletin*, 2 (1972), 21-3.

⁶⁷ Committee on Genetics as it Relates to Social Problems, 'Report of the Committee on Genetics as it Relates to Social Problems', *Genetics Society of Canada Bulletin* 5 (1973), 5-6.

⁶⁸ 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 Hubert C. Soltan for his review of what I have written 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.

⁶⁹ Steering Committee, 'The Canadian College of Medical Genetics', *Genetics Society of Canada Bulletin*, 6 (1975), p. 32.

⁷⁰ The originality and novelty of the Canadian initiative should not be overlooked here. Dr. Park Gerald of Boston's Children's Hospital Medical Center, for example, responded to the announcement of the Canadian College of Medical Genetics in 1976 by saying: 'I really hope ... that you will be interested in the comments of those south of the border before it is finalized. I must say I speak a little defensively. You may be setting a pattern for all of us without our having a chance to participate'. Cited in H. A. Lubs and F. de la Cruz, *Genetic Counseling. A Monograph of the National Institute of Child Health and Human Development* (New York, 1977), pp. 553-4.

⁷¹ The first entrance examinations were held in 1977. And over the next decade, approximately ten fellows were admitted to the College each year after passing written and oral examinations.

⁷² Still, the Royal College imprimatur has been recognized by most provincial licensing authorities as proof that the holder has successfully pursued an approved course of postgraduate education in medicine. See D. S. Lewis, *The Royal College of Physicians and Surgeons of Canada 1920-1960* (Montreal, 1962), p. 25.

⁷³ D. A. E. Shephard, *The Royal College of Physicians and Surgeons of Canada 1960-1980* (Ottawa, 1985), pp. 336-7.