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Leeming, William

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Pre-Publication Draft

The Changing Face of Genetic Counselling in Canada

William Leeming, Ph.D.
Faculty of Liberal Arts & Sciences,
OCAD University

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Correspondence: 100 McCaul Street
Toronto, Ontario M5T 1W1
CANADA

E-mail: bleeming@faculty.ocadu.ca

Abstract: This article focuses on how occupational roles and work relationships have changed over time for genetic counselling activities in Canada. It begins with a review of the stages of consensus that were reached about a role for geneticists and genetic counselling in clinical settings and, second, the formation of medical genetics as a service specialism. Interviews conducted by the author and survey data collected by the Canadian Association of Genetic Counsellors/L'Association Canadienne des Conseillers en Genetique are then used to examine role divarication in genetic counselling and the boundary realignment in inter-professional relations between physician and non-physician personnel involved in counselling activities. This leads, in a final step, to summarize what the research shows about the changing face of genetic counselling in Canada and directions for future historical investigation.

Keywords. genetic counselling; health professions; professionalization; medical genetics

The history of medical genetics mostly consists of accounts of the clinical application of human genetics research and the growth of institution-based interest in medical genetics as a service specialism in Anglo-North American medicine after the Second World War.¹ A number of important milestones are indentified in these accounts: the emergence of heredity clinics and the utilization of Mendelian genetics for purposes of probability guesstimates and empiric risk figure calculations in heredity counselling, later called genetic counselling, in the 1940s; the start of non-physician consultants, i.e., “staff geneticists,” counselling in clinical settings in the 1950s; the introduction of new diagnostic tests and laboratory services in the 1960s for the investigation of chromosomal abnormalities and biochemical testing for metabolites in body fluids; the organization of newborn screening programs in the 1960s; the incorporation of techniques for culturing foetal cells from amniotic fluid and amniocentesis in the 1970s; the first concerted efforts to monitor the rate and direction of genetic counselling and laboratory services and the movement to establish medical genetics as a medical specialty through the 1970s and 1980s; and the “molecular revolution” in laboratory services in the 1980s leading to discussions of genomic medicine by the turn of the century. The key figures populating the historical accounts have mostly been the geneticists who were successful in creating career paths for themselves in medicine over a period of about four decades after the Second World War. Others people have included a variety of clinicians attracted to the prospect of applied human genetics in medicine; technicians who were recruited with backgrounds in cytology, biochemical, and other kinds of lab work; obstetricians recruited to perform amniocentesis; a range of individuals with nursing and social work backgrounds recruited to take up assistive roles to staff geneticists, later medical or clinical geneticists;² and genetic associates, later called genetic counsellors, who were ranked

lower than the clinical geneticists but higher than the nurses and social workers who increasingly took on counselling work.

The primary objective of this article is to help fill in gaps in the literature on the history of genetic counselling in Canada. In doing so, I recognize that certain aspects of the Canadian history of genetic counselling are integrally linked to the history of genetic counselling in the United States. Nevertheless, I maintain that it is important to broaden and extend the view that genetic counselling in North America represents a more or less coherent set of service arrangements that change over time in a more or less analogous manner. What look like coherent sets of service arrangements at the multinational level may also be viewed as loose networks of resource dependencies, personnel, and organizations which can be re-configured within the context of local health care delivery systems. In sum, I wish to contribute to the understanding of how, why, and to what effect local circumstances have affected particular courses of action or inaction.

The article begins with a review of the stages of consensus that were reached about a role for geneticists and genetic counselling in clinical settings and, second, the formation of medical genetics as a service specialism. This review mostly draws on the limited amount of secondary background materials available on the subject of medical genetics as well materials concerning a range of topics gathered from scientific and medical journals, textbooks, handbooks, and manuals. The main focus of the article, however, is on role divarication in genetic counselling and the boundary realignment in inter-professional relations between physician and non-physician personnel involved in counselling activities. This draws, first, on data collected for broader cross-national research I have been conducting which studies the development and growth of genetic health services in Canada and the United Kingdom. Specifically, the data used

in the article comes from open-ended qualitative interviews that were conducted in Canada with service providers involved in genetic counselling in eighteen regional genetics centres over three periods: 1. April 1997 to June 1999 (n=44), 2. September 2001 to May 2003 (n=20), and 3. September 2008 to February 2010 (n=22). Interviewees included twenty-five PhD geneticists (PhDG), thirty MD geneticists (MDG), five nurse genetic counsellors (NGC), and twenty-six MSc-trained genetic counsellors (MScGC). I have made a conscious effort to include among my interviewees individuals in each service catchment area in Canada. These include centres in St. John's, Halifax, Montréal, Québec City, Toronto, North York, Hamilton, London, Oshawa, Ottawa, Winnipeg, Saskatoon, Calgary, and Vancouver. There are absentees, but the absentees are generally smaller centres with links to larger centres elsewhere. Further to this, the number of individuals interviewed from each province reflects, in part, the unequal size and number of regional genetics centres. So, for example, the North West Territories, Yukon, and Nunivut draw on the services of southern provinces; New Brunswick and Prince Edward Island utilize the centre in Halifax, Nova Scotia. Generally speaking the regional centres are designed to serve a catchment area of 1 ½ to 3 million peoples. The original rationale, dating back to the early 1980s, was based on empiric calculations of the proportion of, first, congenital malformations and, second, chronic disease with a major genetic component.

All interviews, where permission was granted, were audio-taped and transcribed into electronic versions. All potentially relevant text was marked for analysis using a computer-aided qualitative data analysis software package (ATLAS.ti). The terms and “codes” used to describe the text have been continually reviewed, categorized, and cross-categorized to uncover prominent themes, patterns and substantial clusters of concepts. Furthermore, with a view to setting the stage for long term analyses of the implications of inter-professional relations in

genetic health services, several key methodological issues have been taken into consideration. On the one hand, data collection has intentionally aimed at capturing complexities rather than aiming at simplification of local circumstances encountered while doing fieldwork. On the other, data analysis has sought to elucidate processes of change in local situations as well as they elucidate national patterns and stabilities. Relatedly, data has been analysed with a view to unravelling the significance of occupational re-stratification sufficiently to make contradictions, ambivalences, and perceived irrelevances clear.

As a final point, I group my interview findings around themes and compare the themes with the findings of surveys conducted every five years by the Professional Issues Committee (PIC) of the Canadian Association of Genetic Counsellors/L' Association Canadienne des Conseillers en Genetique (CAGC/ACCG) (beginning in 1991) in order to track the changing work environment in Canada and new occupational self-awareness among non-physician genetic counsellors.³ In conclusion, I return to what the research shows about the changing face of genetic counselling in Canada and directions for future historical study.

Background: Heredity Counselling, Genetic Counselling and Inter-Professional Relations

In the 1940s, scientists such as Herluf H. Strandkov (University of Chicago) and Laurence H. Snyder (University of Ohio) made reputations for themselves providing heredity-related family counselling, i.e., “heredity counselling.”⁴ The Heredity Clinic of the University of Michigan opened its doors in 1941, funded with a research grant from the Board of Governors of the Horace H. Rackham School of Graduate Studies. The Charles Fremont Dight Institute of the University of Minnesota was established the same year. Likewise, at Winston-Salem, North Carolina, the Out-Patient Department of the North Carolina Baptist Hospital (the teaching hospital of the Bowman Gray School of Medicine) provided heredity counselling under the

direction of C. Nash Herndon. Other heredity counselling services were set up through the 1940s at the Laboratory of Human Genetics, University of Utah; University of Texas; and, in Canada, the Hospital for Sick Children in cooperation with the Department of Zoology, University of Toronto.

In essence, heredity counselling consisted of providing answers to questions about hereditary traits. Questions mostly concerned families in which one or more cases of a hereditary abnormality had already occurred focusing, first, on whether an individual known to have a given trait could pass on the trait to offspring, and, secondly, on the probability of recurrence of a trait in families where one or more affected children had already appeared in a sibship.⁵ Given that the explanation of hereditary processes remained highly theoretical and the aetiological mechanisms underlying hereditary processes were unclear, answers to these questions were delivered in terms of probability guesstimates and empiric risk figure calculations.⁶ Empiric risk figures were calculated for the likelihood of the recurrence of traits in a family or, using existing statistics, to calculate the chance that the future offspring would exhibit such a trait when a parent exhibiting the trait marries into a normal family. Additionally, research in heredity clinics concentrated on the collection and analysis of family records and other kinds of information that was either directly sought after by the resident researchers or came to the clinics through individuals interested in the subject of human inheritance. Data collection in the field started with the identification of the expression of a trait (i.e., physical characteristics, disease, disorder) and status (i.e., kinship, sibship, birthdates, marital, living/dead) surrounding the family of which the *propositus* (i.e., the member who brings the family to the attention of the investigator) is a member. The *propositus* was the central figure of the family or kinship network.⁷ Information would be recorded on a pedigree chart and/or organized on a trait schedule.

The approach to heredity counselling that came to best exemplify the sense of mission of heredity counsellors in postwar North America was that of Sheldon C. Reed. Reed argued that it was never the policy of the Dight Institute to give advice to “clients” *per se*. Rather, counselling was provided in order to enable clients to make clear-cut decisions.

It has never been the policy of the Dight Institute to advise clients that they should, or should not, have subsequent children. Once the couple understands what the genetic situation is, the decision will be a personal matter between the husband and wife.⁸

Heredity counselling was here based on a doctrine of what Reed called “non-directiveness” which presumed that the clients of what Reed would go on to call “genetic counseling” were all mentally sound and rational actors motivated by “a common desire for normal children and healthy families.” “In civilized countries,” he professed, “responsible parents no longer leave reproduction to the vagaries of chance.”⁹ The standpoint taken here was one of methodological individualism whereby the clients are counselled within specific, given constraints and on the basis of the conditions they are living with.

In Reed’s view, physicians’ training in the complex field of human heredity was inadequate if not wholly lacking.¹⁰ At the same time, he believed that physicians would largely be unwilling to make the type of commitment necessary for genetic counselling.

If advice about heredity is to be given, it would seem that the specialist in the field, the geneticist, would be a sounder position to do so than even one’s best friend. The first requirement of the counselor, then, is a reasonable knowledge of human genetics. The second requirement is that he have a feeling for the sensitivities, attitudes, and reactions of the client. An additional advantage would be a medical degree with the training it signifies. This advantage can be somewhat negated by the very strong pressure of the superior income and social prestige which the practice of medicine offers the physician in comparison with the certainty of a none too lavish salary paid by the academic institution where the counseling should be centered.¹¹

Nonetheless, the patient had a right to “expect the answers he gets regarding heredity in his family to be of the same high quality as the surgical or other medical counseling which he

receives.”¹² In years to come, Reed would promote genetic counselling as a part of social work and a valuable public service to be aligned with preventive medicine.¹³

In contrast, many North American geneticists involved in heredity counselling held a different perspective to that of Sheldon Reed, maintaining that they wanted a more active role for the human geneticist *as scientist* working alongside clinicians in medicine. James V. Neel, director of the Heredity Clinic at the University of Michigan, used the metaphor of “teamwork” to explain:

... the geneticist has perhaps been pictured as one interested solely in more or less abstract statistics and probabilities. While such considerations are one important aspect of his field of endeavour, this is by no means the entire story. Increasingly, the geneticist should be regarded as a member of a research team, bringing the problem of the etiology of a particular disease a set of analytical methods which are of greatest value when combined with the approach of the physiologist, the biochemist, and the clinician. The problem of cataloguing diseases with respect to the degree that heredity enters the picture, while an important first step, is in the long run from the standpoint of potential therapeutics of secondary value to specifying the ultimate nature of the genetically controlled departure from the norm.¹⁴

More forcefully, Laurence H. Snyder, then dean of the medical school at the University of Oklahoma, affirmed that the “decisive roles played by the bacteriologist in facilitating the control of infectious disease, by the biochemist in outlining the regulation of nutritional disorders, by the psychologist and psychiatrist in aiding in overcoming the harmful effects of emotional stresses, by the physician in combating the ravages of physiological aberrancies, and by the surgeon in repairing damage due to trauma and irritation, are now being paralleled by the medical geneticist” in the clinical detection of genetic carriers of disease and the preparation of genetic prognoses.¹⁵ The “carrier problem” used in bacteriological theory and preventive medicine as a metaphor was here being extended into genetics in such a manner that “pathologic

genes” were themselves to be viewed as “etiologic agents of disease;” agents that, Snyder argued, would increasingly account for an increasing proportion of physicians’ workloads.

An Association of American Medical Colleges (AAMC) report of 1955 marks the first step towards reaching consensus concerning a role for geneticists in medicine in the United States and Canada.¹⁶ Clinical techniques involving the identification of hereditary factors in disease were described in the report as supplementing the practices of “any [medical] specialty that can be named.”¹⁷ Turning to the question of who should teach genetics, workshop participants generally agreed upon a trained geneticist on the staff who “could also have service and research functions.”¹⁸ Thus, a multi-faceted role was envisioned for geneticists working in teaching hospitals. As a “staff geneticist,” the geneticist would provide genetic counselling, 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.

Discussion generated by the recommendations of the report appears in a number of later surveys and reviews on the subject of teaching human genetics in medical education.¹⁹ Comparatively speaking, five subsequent surveys, completed over a period of three decades, show that the proportion of medical schools in the United States and Canada with formal courses in genetics increased from 8.6 per cent in 1953 to 86.5 per cent in 1985.²⁰ 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 as an interdisciplinary problem; a problem frustrating the larger goal “that ‘genetically thinking’ becomes an integral part of [all]

medical practice.”²¹ Furthermore, arguments emerged for what I have elsewhere called a bifurcated ideological construct to shape and inform the means of organizing a “genetics-based approach to medicine.”²² The construct stipulates, on the one hand, that the mandate of genetic medicine is to add a new set of medical procedures to the clinical repertoire of *all* health disciplines. On the other hand, it indicates that when and where service providers are unable to deliver the new procedures, a class of technical specialists (staff geneticists or medical geneticists/clinical geneticist) will be available for consultation and counselling. As one geneticist I interviewed put it:

The happiest day I could envision ... is when the clinical genetics departments close. And geneticists stop seeing the relatively routine kinds of patients. Because when that wonderful day arrives, that will mean that we have a decent teaching program in genetics in medical schools. And they don't need us, except as sources of information; as people who can take the new and exciting developments in research and communicate them to our clinical colleagues. [M]any of the conditions that we follow, we really shouldn't be doing. I think that we should be acting as consultants who know how to establish the appropriate diagnosis using the exotic and not-so-exotic testing procedures that are available. Then the patient should go back to his or her family physician, paediatrician, or whoever, for the follow-up, and that person should be organizing whatever specialties are needed. (MDG199914)

The first concerted effort to monitor the rate and direction of genetic counselling and laboratory services was mounted in Canada in the early 1970s. Members of the Genetics Society of Canada established the Committee on Genetics as it Relates to Social Problems in 1971, 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 confirmed that an increase in demand for genetic counselling had occurred and attributed the increase to innovations in laboratory and obstetric services.²³ 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.²⁴ It was there that the idea of creating a formal institution to maintain and monitor standards of genetic services first surfaced. What emerged was a coalition to form a corporation to be known as the Canadian College of Medical Geneticists/Collège Canadien de Généticiens Médicaux Canadian (CCMG/CCGMC).²⁵ The proposed College was “not [to be] 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.”²⁶

The Royal College of Physicians and Surgeons (Canada) later supported an application by the CCMG/CCGMC in 1989 to create a free-standing medical specialty with a five-year training program. The recommendation was approved by the Credentials Committee and Royal College’s Council so that medical genetics attained status as one of fourteen clinical specialties recognized by the Royal College’s Division of Medicine. However, in doing so, the role of PhD-geneticists was overturned in terms of performing a consultant role. PhD-geneticists continued on in the field with respect to teaching human genetics in teaching hospitals 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 clinics.

Role Divarication and Widening the Remit of Genetic Counselling in Canada

I have, thus far, highlighted the centrality of geneticists who built career paths in medicine over a period of four decades. They drew a clientele of patients on the basis of personal reputations for specialized expertise in a manner that recalls what Victor Thompson described as “personal specialization.”²⁷ Specialist status arose from the *person*, and not the *task*. Indeed, using Thompson’s nomenclature, there was high personal specialization in medical genetics prior to

specialty formation, but only one operative role, i.e., the staff or consultant geneticist. As a result, a formal job classification became viable as a full-time occupation in medicine. “Task specialization,” nevertheless, followed with specialized occupational roles including non-physician genetic counsellors and work rules for counselling and laboratory services being standardized through the Canadian College of Medical Geneticists/Collège Canadien de Généticiens Médicaux Canadian. However, task specialization, in this context, should not be understood to mean that genetic counselling underwent de-skilling. Rather, genetic counselling became more complex and the accompanying roles underwent divarication in a relatively short period of time together with new technological advancements in genetic diagnostics.²⁸ The first such advancement involved the investigation of chromosomal abnormalities (e.g., the syndromes of Downs, Turner, and Klinefelter). A parallel innovation to what came to be known as cytogenetic analysis, biochemical testing, provided new ways of identifying genetic events by revealing abnormal metabolites in body fluids. The introduction of newborn screening programs in state and provincial hospitals in North America followed when a simple and inexpensive metabolite inhibition assay was developed to detect a treatable metabolic disease (phenylketonuria). Laboratory services were then supplemented with other, more complex techniques for culturing foetal cells from amniotic fluid were refined and amniocentesis was employed as an outpatient procedure for obtaining test samples from pregnant women. This was followed in the 1970s by still more procedures in fetoscopy and chorionic villus sampling (biopsy). Staff geneticists provided pre-procedure counselling here to inform the patient of any associated risks. The geneticists would also schedule when and where the procedure would take place. Furthermore, they would review test results and provide counselling in the event of a positive result.

At the same time, it is reasonable to argue that role divarication in genetic counselling and changes in the boundary realignment in inter-professional relations between physician and non-physician counselling is easier to track in Canada than in the United States due to the presence of a national network of Canadian regional genetic centres. A broad cohesive national strategy has never been conceived for building a genetic health service delivery infrastructure in the United States.²⁹ As a result, it is not possible here to describe in any detail. Certainly, specific components of the way genetic counselling activities are coordinated across North America bear similarities at the local and regional level. But it is beyond the scope of this paper to enumerate the complexity and subtlety of the differences. As such, I concentrate on the general features of role divarication in the regional genetics centres developed in Canada and overseen by the CCMG/CCGMC.

The notion of the regional genetics centre that emerged in the 1970s combined ideas about the health needs of populations with an omnibus “genetic approach” to health and illness and has its roots in the United Kingdom.³⁰ The regional genetic centre was the hallmark of the British “integrated genetics service” of the National Health Service of the 1970s, holding regular clinics in the centre and also satellite clinics to which clinical geneticists and genetic nurses would be dispatched to see patients in District General Hospitals. Responsibility for genetic health services would later devolve from regional administration to conurbations of districts. The genetics service was said to be “integrated” with respect to an integration of counselling and laboratory services for a range of largely rare heritable diseases as well as a selection of disorders that are not strictly familial but requiring chromosomal, biochemical, or (later) molecular analysis. Whilst there have been significant differences in the way each regional centre in the UK and Canada have coordinated service activities for their respective catchment areas, two broad

primary areas of counselling could readily be identified from the beginning. The first set falls under a general category of prenatal care in pregnancy and childbirth, and overlaps with the specialist jurisdictional claims of obstetrics and gynaecology. This set of service delivery activities can be distinguished from “general genetics,” which is a catch-all category for clinical activities involving infants, children and adults. As a set of service delivery activities unto itself, it can be further divided into three sub-sets. Activities in the first sub-set overlap with those of neonatology. This involves the diagnosis and management of congenital anomalies and diseases in newborns. The second sub-set takes up broader paediatric concerns and focuses on the diagnosis and management of genetic conditions in children. Finally, the third sub-set deals with, on the one hand, the diagnosis of adult-onset diseases and, on the other, screening for carriers of heritable conditions. In this regard, the character of the jurisdictional interface between genetic health services and other service specialty areas shift paradigmatically depending on whether the patient was a pregnant woman, an infant, a child, or an adult.

By the time medical genetics had gained specialty recognition in Canada, there were eighteen regional genetic centres. In its simplest geographical aspect, regional services consisted of two generalised unit parts: the centre and the adjoining catchment area. The two developed together, each presupposing the other. But while the centre was compact and readily visible, the catchment area was diffuse and difficult of precise observation. The boundaries of regional genetic services in fact appeared in varying degrees of distinctness at the local level according to the repertoire of policy instruments available, the preferences of the dominant physician elites, and the position and power of local bureaucracies to control funding and other kinds of resources. Genetic services, in this context, represent a series of concentric zones around service centres which differ in the degree of attachment of their occupants to the centres, of the frequency of

movement of patients or patient information to and from the centres, and in the extent to which contacts with the centres are, on the one hand, direct, involving the movement of individuals, or, on the other, indirect, involving a circulation of information and specimens rather than people. These zones include an extension to the regional centre in the form of “outreach clinics” in outlying areas to which medical geneticists and assistive personnel are dispatched on a regular basis to hold clinics lasting between one and three days and collect test samples to take back to centralized laboratory facilities. By the end of the twentieth century, each of the provinces was assigned one centre, with the exception of Alberta (two centres), Quebec (three centres), and Ontario (nine centres). In addition, nine provinces have outreach programs: five outreach clinics in British Columbia run out of Vancouver; nine clinics in Alberta, four run out of Edmonton and five run out of Calgary; one in Saskatchewan; three clinics in Manitoba run out of Winnipeg; nine clinics in Ontario, four run out of Sudbury, three run out of North York, one out of Kingston, and one out of London; four clinics in the maritime provinces, all run out of Halifax; and three clinics in Newfoundland run out of St. John’s.

As previously noted, role divarication occurred with the segregation of MD-geneticists and PhD-geneticists counselling in the regional centres at the time of Royal College recognition in the late 1980s. However, in the period of the 1960s and 1970s, staff geneticists working in genetic centres generally took on responsibility for overseeing laboratory investigations as well as providing counselling. For example, individuals I have interviewed who were involved in early cytogenetic analysis reported a standard regimen whereby diagnostic investigation was initiated by a physician verbally communicating a suspected case of chromosomal abnormality to either a geneticist offering genetic counselling services in a university-clinical program or a cytologist/cytogeneticist in charge of a research laboratory. This was often done over the

telephone. If it appeared that chromosomal analysis was in order, a test kit was dispatched, usually by bus, to the physician concerned. The kit contained four Rockefeller tubes, each containing culture media, together with lancets and heparinized micro-blood-collecting tubes. Upon receipt, the culture media would be kept refrigerated until use. Blood would then be obtained from the patient by the physician, introduced to the culture media in each Rockefeller tube, and mixed by gentle agitation. The Rockefeller tubes were then labelled and packed, and shipped back to the laboratory. On arrival at the laboratory, more solutions of chemicals would be added to the tubes by a laboratory technician, and incubated for several days. The resulting cell suspension would then be pipetted on to a slide, which would be examined with a microscope for chromosome spreading. Slides would then be stained, and cells with good spreading of chromosomes counted and photographed under phase contrast. After the photographs were printed, the chromosome figures would be individually cut out and arranged on bristol board. Finally photostat copies would be made and sent together with the cytogeneticist's report to the physician concerned. Beyond this, the geneticist would be available for consultation by the physician.

The division of labour involved in biochemical testing followed a pattern similar to that of chromosome analysis and a new occupational category gradually appeared: biochemical geneticists. Physicians would look for tell-tale signs and symptoms that might be indicative of a metabolic disease. In such cases, a geneticist might be called upon by attending physicians to investigate the family history for clues of a known inborn error in a family member. If it appeared that a laboratory evaluation was in order, blood or urine or, less frequently, epidermal material, was obtained from the patient and shipped to the laboratory. On arrival at the laboratory, the samples would undergo testing for abnormal metabolites. Finally, a laboratory report would

be sent to the physician concerned. Beyond this, the geneticist would be available for consultation.

It is important to note that, in the early period, personal specialization characterized the way innovations in genetics technologies entered the delivery of health services. As one interviewee explained:

Back at the beginning [in the mid- to late-1960s] it was mainly myself except, well, the laboratory. The first person to be appointed was a PhD cytogeneticist who was really a lab person. So they were at that stage doing the laboratory tests, the chromosome tests, [at] the direct request of the paediatricians or anyone else who wanted them. And, I got tacked on and initially ... we were having to provide [for] our remuneration by doing other activities. So I was doing general paediatrics as well as running a special genetics clinics. In the first instance, it [i.e., genetic counselling] was just a growth phenomenon really. And then, other than secretarial people, the next group that began to evolve was what evolved into being genetic counsellors. But in the instance were often just a nurse or sometimes a science graduate or somebody who just liked dealing with people. You just sort of taught on the spot, and on an in-service basis, to take pedigrees and to run around collecting the background medical records and that sort of thing – the role genetic counsellors now fulfill. But initially it was very much a learning on the spot basis; adding people on that basis was the next bit of growth on the personnel side. (MDG199912)

The auxiliary people mentioned in the above quote represent the second level of role divarication in genetic counselling in the form of non-physician/non-geneticist positions. This level of counselling activity became increasingly important in the delivery of counselling services over time and ultimately produced trained Masters level genetic counsellors who ranked lower than the staff geneticists and physicians but higher than the nurses and social workers who were doing counselling work.. Indeed, non-physician/non-geneticist genetic counsellors have received little attention in history of medical genetics, with a few prominent examples, and merit close attention.³¹

Training programs for Masters level “genetic associates” were first developed at Sarah Lawrence College in Bronxville, New York in 1969.³² By 1975, there were a total of five such

training programs in the United States. The title, “genetic associates” changed to “genetic counsellor” when the National Society for Genetic Counselors was incorporated in the United States in 1979.³³ The following year, the American Board of Medical Genetics agreed to design certifying exams for genetic counselling. Finally, American genetic counsellors went on to establish their own Board, i.e., the American Board of Genetic Counselors in 1992.

On one level, the aim of new training programs was to produce assistive personnel to work under the direction of geneticist-consultants (i.e., “medical” or “clinical” geneticists) and undertake such “routine tasks” in counseling clinics as working up family histories and pedigrees. In addition, the genetic associate was to provide clients with background information about procedures provided in the clinic, including laboratory tests, and counsel on such matters as empiric risk figures concerning the chances of bearing a child with a heritable condition.³⁴ A guiding principle in the early development of the programs followed that of the early geneticist counsellors: the prevention and elimination of genetic disease.³⁵ Counselling with a view to disease prevention informed decisions in the use of different forms of contraception *and*, due to the availability of prenatal diagnosis and therapeutic abortion, decisions to terminate pregnancies of affected fetuses. As noted above, new laboratory technologies for studying chromosomal anomalies and tests for genetic metabolic disease arrived in the 1960s followed by newborn screening programs and routine use of amniocentesis in prenatal diagnosis in the 1970s. Further to this, elective abortion became available in the United States in 1973 and abortion for cause became legal in Canada in 1988. A second (or parallel) guiding principle emerged very early on in the 1970s that said that a primary task of the (Masters level) genetic counselor was to provide what Barbara Bowles Biesecker would call “improved psychological well-being in client adaptation to a genetic condition or risk.”³⁶ It is noteworthy that as early as 1956, Franz J.

Kallmann, then on staff at the New York State Psychiatric Institute of Columbia University picked up on and expanded upon Sheldon Reed's doctrine of non-directiveness, arguing that a counsellor cannot assume that a patient will be realistic about his/her genetic circumstances and that the counsellor should not pit intellect against emotion in the presentation of genetic information. Non-directiveness, the omission of an overt prevention focus, and the goal of assisting patients with their psychological needs and to adapt to their biological circumstances became enshrined in a widely circulated definition of genetic counselling written by a subcommittee of the American Society of Human Genetics under the chair of McGill University's Clarke Fraser in 1974:

Genetic counseling is a communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to: (1) comprehend the medical facts including the diagnosis, probable course of the disorder, and the available management, (2) appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives, (3) understand the alternatives for dealing with the risk of recurrence, (4) choose a course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards and act in accordance with that decision, and (5) to make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.³⁷

In addition to promoting a guiding principle of non-directiveness, the definition acknowledged the emergence of a new occupational role "out of ways in which the new responsibilities are fulfilled rather than merely acquiring or delegating new tasks."³⁸

There were a few genetic counsellors with American Masters degrees working in Canada by the end of the 1970s.³⁹ Furthermore, a Canadian MSc genetic counselling program was established at McGill University in Montreal in 1985 and in 1986 the Canadian College of Medical Geneticists/Collège Canadien de Généticiens Médicaux Canadian formed a sub-committee to look into the role of clinical support personnel and their certification. In response,

the recently established provincial Association of Genetic Counsellors of Ontario called a meeting to discuss certification and the relationship between genetic associates and the College. A “Committee to Investigate the Formation of a National Society” was subsequently formed to open up the discussion on a national level. This resulted in the formation of the first executive board of the Canadian Association of Genetic Counsellors/L’Association Canadienne des Conseillers en Genetique (CAGC/ACCG) in 1987. A draft set of bylaws was written in 1989, and letters patent for the CAGC were signed in 1990. Lastly, in 1997, a combined CCMG/CAGC liaison committee was formed to facilitate communication between the two organizations.

It is important to note that the period of 1985 through to the end of the century represents the movement towards task specialization as counselling duties become shared between physician (i.e., clinical geneticists) and non-physician counsellors. That being said, questions remain to this day about how far should non-physician counsellors be allowed to exercise their own judgement before they infringe on areas that are legally the domain of the physician?⁴⁰ In the 1997 to 1999 phase of interviews I have conducted, interviewees typically talked about the inter-professional physician-counsellor relationships in terms of “teams.” The responsibility of the counsellors was to gather all the preliminary information on the patients who were booked for counselling. All the patients would be discussed at a pre-counselling meeting with the clinical geneticist. The counsellors would usually see patients with a clinical geneticist. Or they would do preliminary intake and then the clinical geneticist would come in with them to see the patient.

One MSc-trained genetic counsellor explained:

We get paged when the family arrives. I go out; bring them into a room. The first thing I do typically with families is explain to them why they’re in genetics. Oftentimes they don’t have a clear concept of what we do, so I start there. I then illicit from them any specific concerns that they have coming into clinic and [establish] what their agenda is. Once I’ve established what our agenda is, letting them know that there’s flexibility ‘cause

they're here to learn information and hopefully we can adjust our agenda so that theirs is in synch as well. I then obtain the medical history, so pregnancy history, birth history, developmental history of that child. I review the medical history that I've obtained from the chart with the family to verify the information is correct and find out if there's anything that we don't know, uh things that have been done at outside institutions. I take a family history; oftentimes dependent upon what the referral is I will illicit certain information from the family considering different diagnoses in our mind. So for example if a child has cleft palate I may be concerned about the consideration of Stickler syndrome so I would be looking for specific information when taking that family history to try to pull out whether or not that is actually present. Once I've done all of that, we go across the hall to start the physical exams; so height, weight, head circumference. Then I go back and I then discuss the case with Dr. ***** [clinical geneticist] so I review with him the information I've obtained pulling out that information – I mean I'm not going to review absolutely every piece; I pull out those pieces that are pertinent and review the family history with him. Bring things to his attention and we then discuss what our plan is going in to see the family. He will do the physical assessment then on the child and we typically come then back out while the family returns to this room, we then may go and discuss the case together, consider different diagnoses, consider plan of care and come up with an approach for that family. If it's a straightforward diagnosis, whether it's likely not to be significant complex medical issues, I don't like the work complex, but if there are issues that I can answer for the family so it's not a vague picture, it's not that there is no diagnosis, typically I finish up with the families on my own in terms of doing the counseling, discussing the condition, discussing recurrence risk, discussing prenatal options. However, if we don't have a diagnosis for a child there's more medical management issues, then Dr. ***** will come in with me and we tend to jointly counsel the family together. So he will discuss medical plan of care and I proceed with my role to be that of making sure that it is clear to the family, what's going on and making sure they understand how it will impact them as a family or impact their child and sort of make sure that they're leaving at a place that they're comfortable with. After the families leave, uh the chart typically comes back to me to facilitate getting the referrals made, filling out any requisitions for testing that we didn't do at the time when the patient was leaving. A letter is written by myself to both the physician and the family and then given to Dr. ***** to review. If there's no diagnosis, Dr. ***** will write the letter to the physician listing the differential the majority of the time and I will write the letter to the family. (MScGC19996)

In the 2008 to 2010 interviews, the description of inter-professional relationships placed greater emphasis on the relative autonomy of the genetic counsellor. Interviewees still referred to teamwork with a supervising geneticist, but the geneticist may or may not be involved in seeing the patient directly. Genetic counsellor interviewees frequently talked about increasing workload

and as having “their own patients.” The complexity of inter-professional relations is illustrated in the following quotes from a MSc-trained genetic counsellor in a large regional genetics centre:

[In the beginning of my employment] I was in charge of prepping her [i.e., medical geneticist] clinics. Whenever she was acting as a medical geneticist, I was doing whatever she needed for her patients. I was prepping her clinics; I was conducting the patients for their testing or to share some results if the patient had indicated the patient would like them over the phone, or reminding them of appointments, asking them if they would be interested in a follow-up with her because after our assistance they were due for follow-up. Now, I kind of supervise a new person. ... And so I'm still involved with Dr. *****'s general genetics patients, but I now see my own patients. ... So I see two to three patients per clinic and I have my clinics on Wednesday afternoons, at the same time that Dr. ***** has her clinics, because she co-signs my letters.

... So I go in with my patient and say what I have to say, then I come back, come out of the room, and then basically fill Dr. ***** in, in terms of what I said, the patient's questions, so she gets a general feeling, and then she comes into the room to basically, what she calls, “say hi,” and make sure the patient is satisfied. If the patient had any type of questions that I wasn't able to answer, she's there. ...

... As counsellors, we have the time to see the patient for a good hour or 40 minutes and basically take the time to say “Okay, why do you think you are here for?” “What do you expect from this appointment so that I can try my best to have you satisfied with the information I'm willing to give you?” “Do you understand what's your situation?” “Would you like an explanation of that?” So we can translate something very complicated into simple words to make sure the patient is aware of what's going on with him/her and the patient feels more, with more knowledge about what's happening and this is not this weird thing that the doctor that I don't really know. And that's what counsellors do, so we provide education to the patient. (MScGC20093)

Variations on description of the relative autonomy of the genetic counsellor in her/his range of duties tended vary with the size of centre and its catchment area. Some genetic counsellors reported ordering genetic tests and referring patients to specialty clinics. Still, in probing the interviewees, I found it was always the case that a physician's signature was mandatory. I was told repeatedly: “it's a legal issue;” “genetic counsellors are not independent practitioners; “we're not licensed;” “there's CAGC – but they're not a governing body; it's a professional organization, they administer the tests that allow us to say we're certified genetic counsellor'.”

One genetic counsellor summed up the predicament in the following manner:

All health regulation is done provincially in Canada. And so every province has their own regulations around that as opposed to a national body. Well, there's two avenues that you could potentially go. One is: you could form a genetic counsellor college ...like a nursing college or any of the other more established health professions. But the trouble is our [low] numbers. ... I mean the main [area of concern] is making a diagnosis. And I think the wording in Ontario is even stricter in the sense of saying communicating a diagnosis, and we certainly do that all the time. But, you know, for example, an amniocentesis is done and the results come back showing the baby has Down's syndrome. I see that as we are making a diagnosis in that situation. And so you could argue, you know, are we contravening the Medical Act by telling the family that the baby has Down's syndrome. This is part of the issue. And we've kind of gotten around that in our clinic in some ways, over time, by having the physicians co-sign. So within the team, there's always a co-signature even though that particular physician doesn't actually see the patient or does not meet the patient, has no knowledge other than the letter that comes to them.

The second avenue that I've been pushing other than...because I don't feel we can get licensure in B.C. given the numbers, is potentially going down the road of delegation of function, which Halifax has done. But our physicians here ... I don't know if they don't want to relinquish control or it's more of a quality issue; I mean it's a bit of both I think. Where they don't feel that they want to delegate function to us. ... Within the province if you said okay, this group is now delegated to, you know, say see a triple screen and follow-up that result, whatever it may be, that's not...you know, and it would save, be more efficient in terms of not having to need a co-signature. It would also remove that physician from any liability associated with that case if anything went awry. Because technically, with their signature on a letter, if anything did go wrong and a suit happened, you know they would be potentially liable as well. (MScGC20103)

Surveys conducted every five years by the Professional Issues Committee (PIC) of the Canadian Association of Genetic Counsellors/L'Association Canadienne des Conseillers en Genetique between 1991 and 2006 provide valuable insights into a changing work environment in Canada and new occupational self-awareness among genetic counsellors.⁴¹ Respondents to the surveys mostly worked in regional genetic centres in urban settings (80.9% in 1995; 92% in 2006). A minority of respondents lived and worked in community clinics or private health care. The surveys nonetheless indicate significant variation and change in the range of official job titles over the fifteen year period.

As noted above, prior to the establishment of MSc level training programs in genetic counselling, the first non physician/geneticist personnel recruited into the counselling clinics

were mostly nurses. The replacement of nurses with MSc level genetic counsellors by retirement was something planned for by regional centre administrators, beginning in the mid- to late 1980s.⁴² Still, it is interesting to note the early preference of nurse respondents to the PIC surveys was to retain the designation “nurse” in their title. While a significant portion of the respondents (44%) to the 1991 survey chose the genetic counsellor designation, 68% of the respondents with nurse included in their present titles said they preferred this to remain the case. (See Table 1.) The “genetic counsellor” designation subsequently outnumbers all other classifications by 2006 and important questions arise concerning levels of occupational autonomy, routinization of tasks, and medical decision-making.

Table 1. Official Job Titles of Counsellors in Canadian Genetics Centres

	1991 N=65 (%)	1995 N=63 (%)	2001 N=67 (%)	2006 N=117 (%)
Genetic Counsellor	26.2%	57.1%	74.6%	79.4%
Nurse	23.3%			0.9% (N=1)
Public Health Nurse/ Assistant		4.8%	3.0%	0.9% (N=1)
Genetic Nurse Coordinator		6.3%	4.5%	0.9% (N=1)
Genetic Associate	18.5%	9.5%	3.0%	2.7%
Genetic/Clinic Coordinator	13.9%			
Clinical Coordinator		1.6% (N=1)	1.5% (N=1)	
Genetic Assistant	6.2%			
Genetic Consultant		1.6% (N=1)	1.5% (N=1)	0.9% (N=1)
Senior Genetic Counsellor/Supervisor/Coordinator		6.3%	1.5% (N=1)	1.8%
Research Associate/Assistant			1.5% (N=1)	1.8%
Other: Coordinator, Clinical Programs, Paediatrics		1.6% (N=1)		

Other: Coordinator, EA Trial, Genetic Counsellor		1.6% (N=1)		
Other: Hereditary Disease Nurse		4.8%		
Other: Volunteer		1.6% (N=1)		
Other: Patient Service Director		1.6% (N=1)		
Other	11.9%		7.5%	11.6%

Source: CAGC PIC Surveys 1991, 1995, 2001, 2006

Many longer service genetic nurses and early genetic counsellors that I have interviewed talked exclusively about working under the supervision of clinical geneticists and performing such administrative tasks as directing clerical staff and managing budgets, answering telephone calls and screening referrals, making appointments and obtaining medical records, medical intake, working up family histories and pedigrees, blood drawing, letter writing and writing up consults, and organizing clinics and outreach clinics. Individuals hired in the late 1980s and early 1990s, however, talked more about growing responsibility with respect to the associates/nurse/counsellors providing patients with background information about procedures offered in the clinic, including laboratory testing. By contrast, interviewees who had more recently entered the field talked about the responsibility for overseeing the health of patients' families as well as the individual patient and the use of computer databases. As one interviewee put it: "We provide recommendations for management but then we do rely on the family physician or paediatrician to follow that child and provide the appropriate care as needed. We're kind of there as a resource but [it is] not [the case] that we are providing primary care for that individual." (MScGC20103)

This is highlighted in the 1991 PIC survey. While 50.8% of survey respondents reported doing primary counselling tasks with the MD-geneticist, 79.4% reported periodic primary counselling alone. Furthermore, significant numbers of respondents did follow-up counselling

(92.1%), making referrals (84.1%), post-patient conferences (63.5%), education (including 79.4% educating health professionals), supervision (15.9%), and physical exams (15.9%). By 1995, the survey design ceased to include questions about professional responsibilities and instead focused on primary areas of service delivery. (See Table 2.)

Table 2. Primary Areas of Service Delivery for Canadian Genetic Counsellors

	1995	2001	2006
Prenatal Counselling	70.10%	67.16%	48.70%
Prenatal/Maternal Screening	36.80%	38.81%	32.17%
Teratogen Exposure	30.0%	31.34%	14.78%
Paediatric (Genetic) Counselling	9.0%	50.75%	39.13%
Adult (Genetic) Counselling	51.80%	61.19%	41.74%
Cancer Genetics	21.70%	37.31%	36.52%
Neurogenetics	18.70%	23.88%	7.83%
Molecular Genetic Diagnostics	30.50%	25.37%	9.57%
Cytogenetic Diagnostics	0.0%	26.87%	10.43%
Biochemical Genetic Diagnostics	0.0%	10.45%	7.83%
Public Health/Newborn Screening	11.50%	7.46%	0.87%
Specialty Disease Clinic	10.50%	5.97%	7.83%
Other	5.0%	7.83%	17.39%

Source: CAGC PIC Surveys 1995, 2001, 2006

A comparison of PIC survey findings indicates a cresting in 2001 and a plateau by 2006 for “other” responsibilities, where there is an increase from 7.83% to 17.39%. The 2006 respondents provided the following areas as other: administration, education, research, cardiac genetics, congenital anomaly surveillance, pre-implantation diagnosis, and “other” specialty clinics. Further questions revealed an increase in primary roles in clinical activities combined with teaching. The number of respondents with MSc training increased from 62.7% to 82.9% and

those pursuing faculty appointments had increased from 4.2% to 54.4%. In addition, respondents seeking more administrative duties had increased from 6.0% to 24.8% and those citing “burn-out” as a reason for leaving counselling altogether had decreased from 45.5% to 6.8%.

The interviews that I have conducted with genetic counsellors suggest two explanations for the changes in PIC survey responses about main areas of service delivery. On a high level of generality, this may reflect the vertical segmentalization associated with the broad primary areas of service delivery outlined in the previous section of this article. This is borne out in interviews about the jurisdictional authority of segments that form with “prenatal counselling,” “paediatric (genetic) counselling,” and “adult (genetic) counselling.” But counselling families is not straightforward. Interviewees talked about the problems associated with shepherding patient and families with complex problems through the system. In many cases the clinical geneticist would be the main source of referral. But where it is not considered a medical “emergency,” genetic counsellors would “facilitate” the process “with the help of our administrative assistants,” talking about this in terms of a “clinic referral”:

... there's a fine line because, when we say *we* make referrals, we know perhaps of the particular physician who may have an interest. ... Oftentimes we come to know specialists who have an interest in particular genetic condition. We write the indication for referral on the form. The form then goes to our administrative staff to be completed and actually send that referral form over or arrange the appointment. There's a role called the information coordinator. They do bookings with clinics. They phone the family to inform them of when they've been booked to see such and such or for a renal ultrasound or whatever the case may be. We give them the form with the medical information completed on it but our information coordinators are the individuals who actually do the bookings. (MScGC19995)

Secondly, the PIC survey data respondents specificities for “other” and the presence of a new area – “cancer genetics” – hints at something more. Indeed interviewees talked about movement between two modalities, one succeeding the other, in which networked innovation and the

circulation of standards of counselling practice is beginning to diffuse horizontally across the jurisdictional boundaries of service specialisms not usually associated with genetic health services. Further to what is already noted above, genetic counsellors described themselves as being parts of networks, acting as advocates for patients seeking clinical care. This can take the form of counsellors working in particular clinics and units, offering a kind of expertise based on their own chosen specialization or research interest. For example:

[The genetic counsellors here] develop different networks depending on what *their area of special interest is*. Or what their clinical caseload is. So the counsellor who works in the cranial-facial unit knows the plastic surgeons who are best at repairing cleft lip and palate, versus the plastic surgeons who are best at repairing hand anomalies, versus the plastic surgeons that we send our patients to for tongue reductions. ... The problem is that the networks are always changing. This physician is going down to the States. This physician is leaving. Or they say that they're not seeing that kind of thing anymore. So it's a very fluid kind of thing. ... We're active proponents for the patients. We're really advocates on their behalf to find out who is most interested [in them]. ... That's part of our job. (MScGC19982)

This interview took place in 1998. Similar points were raised in interviews conducted in 2009 and 2010 with genetic counsellors working in hybridized clinics for neurofibromatosis, cystic fibrosis, infertility, specialized teratogen concerns, familial cancers, familial cardiovascular, and familial ophthalmological conditions. At the same time, the interviews in 2009 and 2010 tended to talk more about administration and the research interests of the genetic counsellors who were increasingly becoming involved in case management, teaching, and journal publication. One interviewee reflected:

... a major part of our role is trying to hold everything together because the amount of work coming in and the counsellors – I think we have a better ability than the physicians to keep ourselves and the clinic organized, keep track of who the patients are, what tests need to be done, who needs to be seen and more of the counsellors' responsibility is becoming yeah, holding that all together and if we drop the ball then things may not get done. Or things may get missed. ... And that's even more probable with the specialty clinics; the genetic counsellor tries to

keep all the different pieces of the team working together. But even in a general clinic where it's mostly just genetic counsellors and geneticists, that's still our role. (MScGC20101)

Conclusions

In *Rethinking Expertise*, Harry Collins and Robert Evans usefully lay out what they call a “periodic table of expertises” in which levels of expertise operate within domains of specialist expertise.⁴³ Specialists immerse themselves in domains of specialist expertise “so as to acquire specialist tacit knowledge.”⁴⁴ The highest level of experts are those with “contributory expertise;” the expertise that is needed to do an activity “with competence.” Contributory expertise is self-sustaining. It can “be taught to new recruits and is passed on from generation to generation by apprenticeship and socialization; someone who has the contributory expertise can pass it to someone who does not have it.”⁴⁵ Below this is “interactional expertise,” which refers to the ability to only master the language of the domain of specialist expertise.

In the early history of genetic counselling in Canada, the notion of contributory expertise can readily be seen in the practices of PhD- and MD-geneticists functioning as, first, heredity counsellors, then staff geneticists in teaching hospitals, and finally medical geneticists in a service specialism. They took up work as a class of technical experts functioning in consultation with physicians from across the spectrum of long-standing service specialisms. At the same time, they operated under a bifurcated ideological construct that aimed to diffuse technical artefacts and knowledge horizontally across the jurisdictional boundaries of *other* service specialisms. Other service specialists can be seen as holding interactional expertise. This has largely remained constant over time. By contrast, the position of non-physician genetic counselling has changed in

medicine. An intrinsic technical logic and language evolved after 1960 that provided a kind of *lingua franca* spoken by all providers of counselling services including assistive personnel. Genetic counselling as a service specialism matured alongside genetic testing and gained a semblance of unanimity over its basic reference points and arrived at a meaning directly tributary to current acceptance of the term. At the same time, there was the segregation of MD-geneticists and PhD-geneticists counselling in the late 1980s with PhD-geneticists losing privileges to counsel. On the other hand, non-physician counselling continues in the form of genetic nurses and MSc trained genetic counsellors working with clinical geneticists in multi-disciplinary teams in regional genetics centres. Second, and concurrently, MSc trained genetic counsellors enter the class of contributory technical experts when they work as liaisons in clinics which may be run by specialist from specialisms other than medical genetics. Importantly, they are responsible for providing technical expertise in the clinic – *without homogenization* (i.e., they do not lose self-identity as *genetic* counsellors).

An argument follows that goes something like this: In the first place, the jurisdictional interface between medical genetics and other specialties is being reinforced and strengthened by the knowledge and technological expertise in genetics that genetic counsellors take with them into hybridized situations. Networked innovation can be seen to have diffused horizontally, as the advantages are recognized in different types of service situations. As networked innovation proceeds, collegial recognition of a divarication of expertise offering genetic counselling is reinforced and fortified. Genetic counselling is here to be regarded as inclusive in such circumstances as opposed to exclusive in the occupational hub culture of medicine. The interviews and CAGC/ACCG survey data outlined in the article point to increasing autonomy on

the part of genetic counsellors working in multidisciplinary teams with respect to diffusing technical knowledge across service specialisms other than medical genetics. The nature of the work done by the genetic counsellor involves a liaison function with the regional genetic centre. The genetic counsellor working in hybridized clinics acts as a conduit of specialist expertise.

To conclude, a notable implication here for future historical study is how local coordination of genetic counselling in hybridised clinics is negotiated, first, in the face of national supervision and, second, despite differences in contributory expertise among the cross-jurisdictional partners. “Collaboration” is a term that may here be useful insofar as it indicates different actors aiming at shared goals. Indeed, as this article shows, far from merging into a homogenous entity, the different actors working in hybridized situations maintain different degrees of distinction in line with their disciplinary backgrounds. The key point is this: Despite significant disciplinary differences there remain localized zones of activity in which a coordinated set of actions is deployed as genetic counselling. As such, the intricate and complex problem of attending to similarity as well as to variation among local versions of service provision and, in particular, to change in the degree of homogeneity over time becomes a matter of importance for future investigation of the changing face of genetic counselling in Canada.

NOTES

¹ For a general overview, see Peter S. Harper, *A Short History of Medical Genetics* (Oxford: Oxford University Press, 2008); Peter S. Harper, *Landmarks in Medical Genetics: Classic Papers with Commentaries* (Oxford: Oxford University Press, 2004). See, also, Hubert C. Soltan, ed., 1992. *Medical Genetics in Canada. Evolution of a Hybrid Discipline: Essays on the Early History* (London, ON: University of Western Ontario Regional Medical Genetics Centre, 1992); Peter A. Coventry and John V. Pickstone, "From What and Why did Genetics emerge as a Medical Specialism in the 1970s in the UK? A Case-History of Research, Policy and Services in the Manchester Region of the NHS," *Social Science & Medicine* 49 (1999): 1227-38; William Leeming, "The Early History of Medical Genetics in Canada," *Social History of Medicine* 17(2004): 481-500; William Leeming, "Ideas about Heredity, Genetics, and 'Medical Genetics' in Britain, 1900-1982," *Studies in History and Philosophy of Biological and Biomedical Sciences* 36 (2005): 538-58; William Leeming, "Tracing the Shifting Sands of 'Medical Genetics': What's in a Name?" *Studies in History and Philosophy of Biological and Biomedical Sciences* 41(2010): 50-60; Nathaniel Comfort, "Polyhybrid Heterogeneous Bastards": Promoting Medical Genetics in America in the 1930s and 1940s," *Journal of the History of Medicine and Allied Sciences* 61 (2006): 415-55; M. Susan Lindee, *Moments of Truth in Genetic Medicine* (Baltimore: Johns Hopkins University Press, 2006).

² The titles “medical geneticist” and “clinical geneticist” are somewhat interchangeable. Historically speaking, the term “clinical geneticist” has more currency after 1990 in clinical settings.

³ I am grateful to The Canadian Association of Genetic Counsellors for access to these survey findings. I am particularly grateful to David Koehn and President Tillie Chiu for their assistance in this matter.

⁴ Lee R. Dice, “Heredity Clinics: Their Value for Public Service and for Research,” *American Journal of Human Genetics*, 4 (1952): 9.

⁵ C. Nash Herndon, “III. Heredity Counselling,” *Eugenics Quarterly*, 1 (1954): 1-2.

⁶ James V. Neel, J. V., “The Meaning of Empiric Risk Figures for Disease or Defect. Proceedings of the Heredity Counseling Symposium Held at the New York academy of Medicine Building (November 1, 1957),” *Eugenics Quarterly*, 5 (1958): 38-41.

⁷ Clarence P. Oliver, “The Collection of Records in the Study of Human Heredity,” *Dight Institute of the University of Minnesota Bulletin*, 2 (1945): 11.

⁸ Sheldon C. Reed, “Counseling in Human Genetics, Part II.” *Dight Institute of the University of Minnesota Bulletin*, 7 (1951): 6.

⁹ Sheldon C. Reed, “Heredity Counseling: The Role of the Doctor.” *Eugenics Quarterly*, 3 (1956): 106.

¹⁰ Reed, “Counseling in Human Genetics, Part II,” p.7.

¹¹ Sheldon C. Reed, “Counseling in Human Genetics, Part III.” *Dight Institute of the University of Minnesota Bulletin*, 8 (1953): 15.

¹² Reed, “Heredity Counseling: The Role of the Doctor,” p. 106.

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- ¹³ Reed, "Counseling in Human Genetics, Part III," p. 19. See, also, Sheldon C. Reed, "A Short History of Genetic Counseling," *Social Biology*, 21 (1974): 336.
- ¹⁴ James V. Neel, "Some Applications of the Principles of Genetics to the Practice of Medicine," *Medical Clinics of North America*, 35 (1951): 532.
- ¹⁵ Laurence H. Snyder, "Old and New Pathways in Human Genetics," *American Journal of Human Genetics*, 3 (1951): 13.
- ¹⁶ Association of American Medical Colleges, Subcommittee on Objectives of Teaching, "The Teaching of Pathology, Microbiology, Immunology, Genetics," *Journal of Medical Education*, 30 (Suppl., Sept. 1955): 1-23.
- ¹⁷ AAMC, "The Teaching of Pathology, Microbiology, Immunology, Genetics," p. 19.
- ¹⁸ AAMC, "The Teaching of Pathology, Microbiology, Immunology, Genetics," p. 22-3.
- ¹⁹ See James V. Neel, "The Functions of a Department of Genetics in a Medical School," *Journal of Medical Education*, 3 (1957): 493-500; Francis Hickman, "Comprehensive Planning of Human Genetics Education: A Position Statement," *American Journal of Medical Genetics*, 8 (1981): 243-247; Barton Childs, "Genetics in the Medical Curriculum," *American Journal of Medical Genetics*, 13 (1982): 319-24; Ronald G. Davidson and Barton Childs, "Perspectives in the Teaching of Human Genetics," *Advances in Human Genetics*, 16 (1987): 79-119; J. M. Graham, J. I. Rotter, V. M. Riccardi, P. A. Baird, J. Benkendorf, J. Bodurtha, B. Childs, R. G. Davidson, C. A. Huether, K. Laing, R. F. Murray, H. Nadler, W. E. Nance, M. Nesbitt, C. R. Scriver, E. M. Short, R. L. Summitt, and H. V. Toriello, H.V. "Report of the Task Force on

Teaching Human Genetics in North American Medical Schools,” *American Journal of Human Genetics*, 44 (1989): 161-65.

²⁰ See M. D. Levine, M. D., 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, F., eds., *Genetic Counseling. A Monograph of the National Institute of Child Health and Human Development* (New York: Raven Press, 1977), pp. 349-68; G. Gordon Robertson and John C. Haley, “Genetics in the Medical Curriculum,” *Journal of the Association of American Medical Colleges*, 21 (1946): 351-53; C. Nash Herndon, “Human Genetics and Medical Education,” *Journal of Medical Education*, 29 (1954): 13-19; Barton Childs, Carl A. Huether, and Edmond A. Murphy, “Human Genetics Teaching in U.S. Medical Schools,” *American Journal of Human Genetics*, 33 (1981): 1-10; Vincent M. Riccardi and Roy 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.

²¹ Arno 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: Academic Press, 1979), pp. 78; Hickman, “Comprehensive Planning of Human Genetics Education.”

²² Following the example of Robbins and Johnston, I use the phrase “professional 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 chapter. It is not here used in its broader sense of worldview, i.e.,

as a *Weltanschauung*. See David Robbins and Ron Johnston, R. (1976) “The Role of Cognitive and Occupational Differentiation in Scientific Controversies,” *Social Studies of Science*, 6 (1976): 349-368. See, also, William Leeming, “Professionalization Theory, Medical Specialists and the Concept of ‘National Patterns of Specialization’,” *Social Science Information*, 40 (2001), pp. 468-469.

²³ James R. Miller, “Report of Genetic Counselling Services in Canada,” *Genetics Society of Canada Bulletin*, 2 (1972): 21-23.

²⁴ William Leeming, “The Early History of Medical Genetics in Canada,” *Social History of Medicine*, 17 (2004), p. 494-95.

²⁵ The Canadian College of Medical Geneticists/Collège Canadien de Généticiens Médicaux Canadian preceded similar developments in the Netherlands, the United States, Finland, Sweden, Germany, France, and Denmark.

²⁶ Steering Committee, “The Canadian College of Medical Genetics,” *Genetics Society of Canada Bulletin*, 6 (1975), p. 32.

²⁷ Victor A. Thompson, *Modern Organization* (New York: Knopf, 1964), pp. 25-7.

²⁸ William Leeming, “Tracing the Shifting Sands of ‘Medical Genetics’: What’s in a Name?,” *Studies in History and Philosophy of Biological and Biomedical Sciences*, 41 (2010): 50-60.

²⁹ Cf. Jane S. Lin-Fu and Michele A. Lloyd-Puryear, “Access to Genetic Services in the United States: A Challenge to Genetics in Public Health,” in M. J. Khoury, W. Burke, and E. J. Thomson, eds., *Genetics and Public Health in the 21st Century: Using Genetic*

Information to Improve Health and Prevent Disease (Oxford: Oxford University Press, 2000), pp. 273-89.

³⁰ William Leeming, "Ideas about Heredity, Genetics, and 'Medical Genetics' in Britain, 1900-1982," *Studies in History and Philosophy of Biological and Biomedical Sciences*, 36 (2005): 538-558.

³¹ I am thinking of the work of Regina H. Kenen and, more recently, Alexandra Stern on genetic counsellors in the United States. See Regina H. Kenen, "Genetic Counseling: The Development of a New Interdisciplinary Occupational Field," *Social Science and Medicine*, 18 (1984): 541-49; Regina H. Kenen, "The Human Genome Project: Creator of the Potentially Sick, Potentially Vulnerable and Potentially Stigmatized," in I. Robinson, ed., *Social Consequences of Life and Death under High Technology Medicine* (Manchester: Manchester University Press, 1994), pp. 49-64; Regina H. Kenen, "Opportunities and Impediments for a Consolidating and Expanding Profession: Genetic Counseling in the United States," *Social Science and Medicine*, 45 (1997): 1377-86; Regina H. Kenen and Anne Smith, A., "Genetic Counseling for the Next 25 Years: Models for the Future," *Journal of Genetic Counseling*, 4 (1995): 115-123. See, also, Alexandra M. Stern, "A Quiet Revolution: The Birth of the Genetic Counselor at Sarah Lawrence College, 1969," *Journal of Genetic Counseling*, 18 (2009): 1-11.

³² Alexandra M. Stern, "A quiet revolution: the birth of the genetic counselor at Sarah Lawrence College, 1969," *Journal of Genetic Counseling*, 18 (2009): 1-11.

³³ Audrey Heimler, “An oral history of the National Society of Genetic Counselors,” *Journal of Genetic Counseling*, 6 (1997): 315-36.

³⁴ Y. Edward Hsia, “Approaches to the Appraisal of Genetic Counseling,” in Herbert A. Lubs and Felix de la Cruz, F. (eds.), *Genetic Counseling. A Monograph of the National Institute of Child Health and Human Development* (New York: Raven Press, 1977), pp. 53-82; T. E. Kelly “Genetic Counseling,” in T. E. Kelly T. E. (ed.), *Clinical Genetic Counseling* (Chicago, IL: Year Book Medical Publishers, 1986), pp. 343–364.

³⁵ Seymour Kessler (ed.), “Introduction,” *Genetic Counseling: Psychological Dimensions* (New York: Academic Press, 1979), pp. 1-11.

³⁶ Barbara Bowles Biesecker, “Goals of Genetic Counseling,” *Clinical Genetics*, 60 (2005): 323-330.

³⁷ F. Clarke Fraser, “Genetic Counseling,” *American Journal of Human Genetics*, 26 (1974): 636–661.

³⁸ F. Clarke Fraser, “Genetic Counseling,” p. 100. At the same time, it needs to be noted that Fraser was more intent on envisioning a role for general practitioners than non-physician counsellors. That being said, he wrote enthusiastically about the promise of multidisciplinary teamwork in “medical genetics units,” specialty recognition for genetic counselling in the Canadian province of Saskatchewan and by the Royal College of Physicians of Edinburgh (Scotland), and the start-up of the two training programs for genetic associates at Sarah Lawrence College and Rutgers University.

³⁹ Susan Zeesman and Susan Creighton, “The CAGC: a history in progress.” Unpublished document circulated (c. 2000) within the Canadian Association of Genetic Counsellors.

⁴⁰ Kenen, “Genetic counseling,” p. 548.

⁴¹ I am grateful to The Canadian Association of Genetic Counsellors for access to these survey findings. I am particularly grateful to David Koehn and President Tillie Chiu for their assistance in this matter.

⁴² See William Leeming, *Medical Specialization and Medical Genetics in Canada (1947 and After)*. Unpublished PhD thesis, York University, Toronto, 1999.

⁴³ Harry Collins and Robert Evans, *Rethinking Expertise* (Chicago: University of Chicago Press, 2009).

⁴⁴ Collins and Evans, *Rethinking Expertise*, p. 14.

⁴⁵ Collins and Evans, *Rethinking Expertise*, p. 35.