Looking Back on the Future of Genetic Counselling in Canada

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Abstract. This article focuses on how occupational roles and working relationships have changed over time for individuals involved in genetic counselling 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 Génétique are then used to examine role divarication in genetic counselling and the boundary realignment in inter-professional relations among physician and non-physician genetic counsellors. This leads, in a final step, to a summary of what the research shows about the changing face of genetic counselling in Canada and directions for future investigation.

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

Résumé. Cet article examine comment ont évolué les rôles et relations de travail des personnes actives en counseling génétique au Canada. Il brosse d’abord un tableau des consensus qui se sont succédé en ce qui a trait au rôle de la génétique et du counseling génétique en milieu clinique, et poursuit avec la formation de la médecine génétique comme spécialité. Des entrevues menées par l’auteur et les données d’une enquête effectuée par la Canadian Association of Genetic Counsellors/L’Association Canadienne des Conseillers en Génétique sont utilisées pour étudier la diversification des rôles en counseling génétique et les réalignements dans les relations inter-professionnelles entre conseillers médecins et non-médecins. L’article propose finalement un bilan de ce que la recherche fait ressortir quant au visage changeant du counseling génétique au Canada, ainsi que des pistes pour des travaux futurs.

Mots-clés. counseling génétique, professions de la santé, professionnalisation, médecine génétique

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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 medical specialty in Anglo-North American medicine after the Second World War. A number of important milestones are identified in these accounts: the emergence of heredity clinics and the application of Mendelian genetics for purposes of probability guesstimates and empiric risk figure calculations in heredity counselling in the 1940s; the use of geneticists for counselling in clinical and university 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; the “molecular revolution” in laboratory services in the 1980s; and the Human Genome Project in the 1990s leading to discussions of genomic medicine by the turn of the century. The key figures populating these accounts have been the geneticists who were successful in creating career paths for themselves in medicine as staff geneticists, later medical or clinical geneticists, or overseeing laboratory testing services and research over a period of about four decades after the Second World War. Relatively little has been written about the genetic associates, later called genetic counsellors, who were ranked lower than the medical geneticists but higher than nurses and social workers who were recruited to take up 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 attempting to do so, I acknowledge that many aspects of the Canadian history of genetic counselling are integrally linked to the history of genetic counselling in the United States. Nevertheless, I believe that it is important to broaden and extend the view that the history of 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. Rather, I maintain that what look like coherent sets of service arrangements at the multinational level really represent loose networks of resource dependencies, personnel, and organizations which have been re-configured within the context of local health care delivery systems. Accordingly, I wish to contribute to the understanding of how, why, and to what effect local circumstances have affected particular courses of action or inaction in Canada.
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. But the main focus of the article 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 on data collected for the broader purposes of cross-national research I am conducting on the development and growth of genetic health services in Canada and the United Kingdom. The data used in the article comes from open-ended qualitative interviews, which were conducted in Canada with service providers involved in genetic counselling in 18 regional genetics centres over three periods: April 1997 to June 1999 (n=44), September 2001 to May 2003 (n=20), and September 2008 to February 2010 (n=22). Interviewees included 25 PhD geneticists (PhDG), 30 MD geneticists (MDG), 5 nurse genetic counsellors (NGC), and 26 MSc-trained genetic counsellors (MScGC). I have made a conscious effort to include among my interviewees individuals in each service catchment area in Canada. There are absentees, but the absentees are generally smaller centres with links to larger regional genetics centres. As a final point, I have grouped my interview findings around themes and compare the themes with the findings of surveys, beginning in 1991, conducted every five years by the Professional Issues Committee (PIC) of the Canadian Association of Genetic Counsellors/L’Association Canadienne des Conseillers en Génétique (CAGC/ACCG) in order to track the changing work environment in Canada and new occupational self-awareness among non-physician genetic counsellors.

BACKGROUND: HEREDITY COUNSELLING, GENETIC COUNSELLING, AND THE INTER-PROFESSIONAL RELATIONS OF INDIVIDUALS OFFERING COUNSELLING

In the 1940s, scientists such as Herluf H. Strandskov (University of Chicago) and Laurence H. Snyder (University of Ohio) made reputations for themselves providing heredity-related family counselling, i.e., “heredity counselling.” The Heredity Clinic of the University of Michigan opened its doors in 1941, the same year that the Charles Fremont Dight Institute of the University of Minnesota and a clinic at the Bowman Gray School of Medicine were established. Other heredity counselling services were set up through the 1940s at the Laboratory of Human Genetics, University of Utah; the University of Texas; and, in Canada, the Hospital for Sick Children, Toronto.

In essence, heredity counselling consisted of using Mendelian genetics to provide answers to questions from physicians and families about
hereditary traits in families.\textsuperscript{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.\textsuperscript{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 were either directly sought after by the resident researchers or came to the clinics through individuals interested in the subject of human inheritance.\textsuperscript{7}

The approach to heredity counselling that came to best exemplify the sense of mission of heredity counsellors in the 1950s was that of Sheldon C. Reed of the Dight Institute of the University of Minnesota. Reed argued that it had never been the policy of the Institute to give advice to “clients” \textit{per se}. Rather, counselling was provided in order to enable clients to make clear-cut decisions, “a personal matter between the husband and wife.”\textsuperscript{8} Counselling was based on a doctrine of what Reed called “non-directiveness,” which presumed that the clients of what he 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.”\textsuperscript{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.\textsuperscript{10} At the same time, he believed that physicians would largely be unwilling to make the type of commitment necessary for genetic counselling.\textsuperscript{11} Nevertheless, the patient should have 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.”\textsuperscript{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.\textsuperscript{13}

Other 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 physicians in medicine. James V. Neel, director of the Heredity Clinic at the University of Michigan, used the metaphor of “teamwork” to explain: “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." More forcefully, Laurence H. Snyder, then dean of the medical school at the University of Oklahoma, asserted that the “decisive role” of the “medical geneticist” in the detection of genetic carriers of disease and the preparation of genetic prognoses paralleled the role played by the bacteriologist in facilitating the control of infectious disease.

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. 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.” 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. 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 offering formal courses in genetics increased from 8.6% in 1953 to 86.5% in 1985. Arguments emerged here for what I have elsewhere called a bifurcated ideological construct to shape and inform the means of organizing a “genetics-based approach to medicine.” The construct stipulates, on the one hand, that the mandate of a genetics-based approach to 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 (e.g., staff geneticists) will be available for consultation and counselling.

The first concerted effort to monitor the rate and direction of genetic counselling as a clinical service 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 22 individuals known to be providing genetic services in 13 cities confirmed that an increase in demand for genetic counselling had
occurred and attributed the increase to innovations in laboratory and obstetric services.\textsuperscript{21} 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.\textsuperscript{22} It was there that the idea of creating a formal institution to maintain and monitor standards of service 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 (CCMG/CCGMC).\textsuperscript{23} 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.”\textsuperscript{24}

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 14 clinical specialties recognized by the Royal College’s Division of Medicine. However, in doing so, the role of PhD-geneticists was overturned in favour of MD-geneticists performing a counselling role in Canada. PhD-geneticists continued on in the field with respect to, first, teaching human genetics in medical schools and, second, the delivery of laboratory services independent of the counselling services. As a rule, however, they ceased to counsel patients.

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 genetic counselling 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.”\textsuperscript{25} 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, later medical/clinical, 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 being standardized through the Canadian College of Medical Geneticists/Collège Canadien de Généticiens Médicaux. 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
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accompanying roles underwent divarication in a relatively short period of time together with new technological advancements in genetic diagnostics. 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 provincial hospitals followed when a simple and inexpensive metabolite inhibition assay was developed to detect a treatable metabolic disease (phenylketonuria). Laboratory services were then supplemented when 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). 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.

The notion of the regional genetics centre that emerged in the 1970s combined ideas about the health needs of populations with an omni-bus “genetic approach” to health and illness and has its roots in the United Kingdom. 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. The genetics service was said to be “integrated” in the sense of an integration of counselling and laboratory services for a range of largely rare heritable diseases. While there have been significant differences in the way each regional centre in Canada and the UK have co-ordinated service activities for their respective catchment areas, two broad primary areas of counselling can readily be identified. 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 subsets. Activities in the first subset overlap those 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 genetic conditions in children. Finally, the third subset 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. Again, in the early period, personal specialization characterized the way innovations and innovators in genetics technologies entered the delivery of health services. As one of my interviewees 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 special genetics clinics. In the first instance, 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 it was 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)

In its simplest geographical aspect, the regional centre has consisted of two generalized unit parts: the centre and the adjoining catchment area. The two developed together, each presupposing the other. But while the centre has been compact and readily visible, the catchment area has been diffuse and difficult of precise observation. The boundaries of regional genetic services in fact have 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 health services, in this context, have represented 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 have also included 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
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days. By the end of the 20th 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 had 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 run out of Saskatoon; 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 began 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. I want to stress here that it was not simply the case that the technology advanced and technological advancement drove specialty formation. The division of labour followed patterns of specialization dictated by the Royal College. From here, further complexity occurred with the introduction of non-physician genetic counselling positions. It is important here to note that non-physician genetic counsellors have received little attention in the history of medical genetics, with a only few prominent examples.28

NON-PHYSICIAN GENETIC COUNSELLORS IN CANADA

Training programs for Masters level “genetic associates” were first developed at Sarah Lawrence College in Bronxville, New York, in 1969. By 1975, there were a total of five such training programs in the United States. The title, “genetic associate” changed to “genetic counsellor” when the National Society for Genetic Counselors was incorporated in 1979.29 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 goal of the new training programs was to produce assistive personnel to work under the direction of geneticists and undertake such tasks in counselling clinics 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. In addition, the genetic associate was to provide patients with background information about procedures provided in the clinic, including requisitioning of laboratory tests, and counsel on such matters as empiric risk figures concerning the chances of bearing a child with a heritable condition.30 A guiding principle in the early development of the programs followed that of the early geneticist
counsellors: the prevention and elimination of genetic disease.\textsuperscript{31} 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 goal of the genetic associate was to provide what Barbara Bowles Biesecker would call “improved psychological well-being in client adaptation to a genetic condition or risk.”\textsuperscript{32} 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.\textsuperscript{33}

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.”\textsuperscript{34} At the same time, it needs to be stated that Fraser was more intent on envisioning a role for general practitioners in the delivery of counselling services—not 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.

There were a few genetic counsellors with American Masters degrees working in Canada by the end of the 1970s. 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 formed a subcommittee 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 Génétique (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.

The period of 1985 through to the end of the century saw the movement towards shared responsibilities between physician (i.e., clinical geneticists) and non-physician counsellors. However, 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. In the 1997 to 1999 phase of interviews I have conducted with clinical geneticists, genetic nurses, and genetic counsellors, interviewees typically talked about the inter-professional physician-counsellor relationships in terms of “teamwork.” It was the responsibility of the counsellors to gather all the preliminary information on the patients who were booked for counselling. All the patients would subsequently be discussed at a pre-counselling meeting with the clinical geneticist. The counsellors would generally see patients with a clinical geneticist. Alternatively they would perform 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.... 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 elicit 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…. Oftentimes dependent upon what the referral is I will elicit certain information from the family considering different diagnoses in our mind…. 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… 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 there are issues that I can answer for the family so it’s not a vague picture. Typically I finish up with the families on my own in terms of doing the counselling, discussing the condition, discussing recurrence risk, discussing prenatal options. However, if we don’t have a diagnosis for a child and there’s more 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 make sure that they’re leaving at a place that they’re comfortable with. After the family leaves, 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. The majority of the time I will write the letter to the family. (MScGC19996)

In the 2008 to 2010 interviews I have conducted, the descriptions of inter-professional relationships place great emphasis on the relative autonomy of the genetic counsellor. Interviewees still referred to teamwork with a supervising geneticist, but the geneticist is often described as being less involved in seeing patients. Genetic counsellor interviewees frequently talked about their workloads increasing and as having their “own patients.” The complexity of inter-professional relations is illustrated in the following quotes from an MSc-trained genetic counsellor in a large regional genetic 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. (MScGC20093)

Variations on interviewees’ discussion of the relative autonomy of the genetic counsellor tend to vary with respect to the size of the regional centre and its catchment area. Some genetic counsellors reported requisitioning 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 exams that allow us to say we’re certified genetic counsellors.” 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…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…. Within the province if you said okay, this group is now delegated to, you know, say to see a triple screen and follow-up that result, whatever it may be, it would 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, they would be potentially liable as well. (MScGC20103)

Surveys conducted every five years by the Professional Issues Committee (PIC) of the CAGC/ACCG between 1991 and 2006 provide insights into a changing work environment in Canada and new occupational self-awareness among genetic counsellors. Respondents to the surveys have mostly worked in regional genetic centres in urban
settings (80.9% in 1995; 92% in 2006). A minority of respondents have 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 15-year period.

As noted earlier, the first non-physician personnel recruited into the counselling clinics were nurses and social workers. The replacement of nurses with MSc-trained genetic counsellors by retirement was something planned for by regional centre administrators, beginning in the mid- to late 1980s.\(^\text{37}\) Still, it is interesting to note that 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 that this remain the case. (See Table 1.) The “genetic counsellor” designation subsequently outnumbered all other classifications by 2006.

### Table 1

<table>
<thead>
<tr>
<th>Official Job Titles of Counsellors in Canadian Genetics Centres</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
</tr>
<tr>
<td>Genetic Counsellor</td>
</tr>
<tr>
<td>Nurse</td>
</tr>
<tr>
<td>Public Health Nurse/ Assistant</td>
</tr>
<tr>
<td>Genetic Nurse Coordinator</td>
</tr>
<tr>
<td>Genetic Associate</td>
</tr>
<tr>
<td>Genetic/Clinic Coordinator</td>
</tr>
<tr>
<td>Clinical Coordinator</td>
</tr>
<tr>
<td>Genetic Assistant</td>
</tr>
<tr>
<td>Genetic Consultant</td>
</tr>
<tr>
<td>Senior Genetic Counsellor/ Supervisor/Coordinator</td>
</tr>
<tr>
<td>Research Associate/Assistant</td>
</tr>
<tr>
<td>Other: Coordinator, Clinical Programs, Paediatrics</td>
</tr>
<tr>
<td>Other: Coordinator, EA Trial, Genetic Counsellor</td>
</tr>
<tr>
<td>Other: Hereditary Disease Nurse</td>
</tr>
<tr>
<td>Other: Volunteer</td>
</tr>
<tr>
<td>Other: Patient Service Director</td>
</tr>
<tr>
<td>Other</td>
</tr>
</tbody>
</table>

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While 50.8% of survey respondents in the 1991 PIC survey reported doing primary counselling tasks with the clinical 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%). It is interesting to note that 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

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


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 level 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, there is referencing to 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 clearly 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 a 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 specificities for “other” and the presence of a new area—“cancer genetics”—suggest that something more is going on. Indeed, my interviewees have talked about movement between two modalities, one succeeding the other, in which networked innovation and the circulation of standards of counselling practice are beginning to diffuse horizontally across the jurisdictional boundaries of medical specialties not usually associated with medical genetics. Further to what has already been said 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 hybridized 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)

Similar points were raised in interviews conducted with genetic counsellors working in clinics for neurofibromatosis, cystic fibrosis, infertility, specialized teratogen concerns, familial cancers, familial cardiovascular, and familial ophthalmological conditions. At the same time, interviews in 2009 and 2010 specifically underscored the importance of genetic counsellors in clinic administration and case management. One interviewee summarized:

… 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. (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 are shown to operate within specialized domains. Specialists immerse themselves in domains of specialist expertise “so as to acquire specialist tacit knowledge.” 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.” Placed 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, contributory expertise can readily be seen in the practices of geneticists functioning as, first, heredity counsellors, then staff geneticists in teaching hospitals, and finally medical/clinical geneticists in a recognized medical specialty. They have taken up work as a class of technical experts functioning in consultation with physicians from across the spectrum of long-standing
medical specialties. Importantly, they have operated under a bifurcated ideological construct that aims to diffuse technical artefacts and a “genetics-based approach to medicine” horizontally across the spectrum of different medical occupational roles and specializations. At the same time, the construct stipulates that when and where service providers are unable to deliver the new procedures, a class of technical specialists (e.g., staff geneticists) will be made available for consultation and counselling. In this context, the role of genetic associate first entered inter-professional relations with a level of interactional expertise with respect to applied human genetics in clinical settings. With time, MSc-trained genetic counsellors entered the class of contributory technical experts. Indeed, an intrinsic technical logic and language evolved after 1970 that provided a kind of lingua franca spoken by all experts in genetic counselling services including MSc-trained genetic counsellors. This is particularly evident in the case of MSc-trained genetic counsellors working as liaisons in hybridized clinics that involve specialists from specialties other than medical genetics. While the other specialists may hold levels of interactional expertise with regard to the genetics of the specific disease group they are serving, it is the genetic counsellors who are ultimately knowledgeable in the genetics of disease and a genetics-based approach to medicine. Furthermore, it is noteworthy that the genetic counsellors operate in such service arrangements without losing their occupational identity as genetic counsellors.

An argument follows that goes something like this: In the first place, the jurisdictional interface between medical genetics and other medical specialties is being reinforced and strengthened by the knowledge and technological expertise in genetics as science that genetic counsellors in a variety of occupational roles have taken with them into a range of clinical settings. Networked innovation can be seen to have diffused horizontally as the advantages are recognized in different types of service arrangements. As networked innovation has proceeded, collegial recognition of the expertise of medical geneticists and genetic counsellors has been reinforced and fortified in medicine. Further to this, genetic counselling as a whole is to be regarded as inclusive in such circumstances as opposed to exclusive in the occupational hub culture of medicine because of the bifurcated ideological construct that it upholds.

To conclude, a notable implication for future historical study is how local coordination of genetic counselling in regional genetic centres and hybridised clinics has been negotiated, first, in the face of national oversight and, second, despite differences in contributory and interactional 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. However, as this article shows, far from merging into a homogenous entity, the different actors working in hybridized
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clinical situations maintain different degrees of distinction in line with their specialized backgrounds. A key point is this: Despite significant 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 overall similarity as well as to variation among local versions or styles of service delivery over time becomes a matter of importance for future investigation of changes in the face of genetic counselling in Canada—and elsewhere.

ACKNOWLEDGEMENTS

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NOTES


2 The titles “medical geneticist” and “clinical geneticist” are somewhat interchangeable. Historically speaking, the term “clinical geneticist” has more currency after 1980 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.
10 Reed, “Counseling in Human Genetics, Part II,” p.7.
17 AAMC, “The Teaching of Pathology, Microbiology, Immunology, Genetics,” p. 22-23.
20 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.
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23 The Canadian College of Medical Geneticists/ Collège Canadien de Généticiens Médicaux preceded similar developments in the Netherlands, the United States, Finland, Sweden, Germany, France, and Denmark.


34 F. Clarke Fraser, “Genetic Counseling,” p. 100.


38 Harry Collins and Robert Evans, Rethinking Expertise (Chicago: University of Chicago Press, 2009).
40 Collins and Evans, *Rethinking Expertise*, p. 35.