MEDICAL GENETICS IN CANADA
Evolution of a Hybrid Discipline

ESSAYS ON THE EARLY HISTORY
H.C. Soltan, Editor
Chapter 12. Newfoundland and Labrador

Since the mid 1900s, many and varied persons have contributed to investigations in medical genetics in Newfoundland and Labrador. Rather than name them one by one, most will speak in their own words through references listed in Appendix B1. The bibliography commences with some papers and observations published prior to the establishment of the Faculty of Medicine at Memorial University in 1967 (1-5). The chronologic order reflects the sequence of the investigator’s activity in the Province and, for some, their retirement or departure from Newfoundland. Without the Memorial University Faculty of Medicine (49), the majority of these scientists would never have moved into the Province, and the granting agencies such as the Medical Research Council (MRC), for example, would not have awarded the support credited in these papers. In contrast to the situation in some other Provinces, the Newfoundland Government has no intraprovincial source of research funds for human genetics. Major support must be obtained from funding agencies based outside of Newfoundland.

Preplanning for development in genetics for the Faculty of Medicine included consultation by the Dean (Ian Rusted) and Associate Dean (Ken Roberts) with John Edwards, Clarke Fraser, Victor McKusick, Arno Motulsky, James Neel, Howard Newcombe, Charles Scriver, and others. Since 1967, faculty with research interests in biochemical genetics,
cytogenetics, epidemiology, immunology, and population genetics, as well as physician specialists concerned about the medical effects of genetic disease, have gradually been recruited. A letter to the president of the Medical Research Council, Dr. Malcolm G. Brown, introducing Memorial University’s 1972 proposal for support to establish a Medical Research Council of Canada Genetics Group in the Faculty of Medicine began as follows:

Virtually every medical scientist who has visited this university in recent years has agreed with us that we have a unique opportunity to carry out research in genetics which would contribute significantly to the Canadian Research scene. It would also be relevant to social needs and therefore provide an important service to the province.

The submission was coordinated by Ken Roberts with sections by L.F. Bernini of Leiden (human biochemical genetics), George R. Fraser (subsequently appointed as Professor of Medical Genetics at Memorial University), Richard B. Middleton (microbial genetics), C.S. Mellor (genetics in psychiatry), and with input from John Darte, W. Marshall, Ian Rusted, Bruce Sells, and others. The initial budget request for portions of professors’ salaries, for postgraduate and graduate students, research assistants, and support staff, as well as for equipment and supplies for five sections or laboratories, was in range of $270,000 for 1972/73, increasing to about $430,000 by 1974/75. Competition for recognition and establishment as an MRC Genetics Group came from McGill University. One hears that after the site visits, the decision was tipped by one vote in favor of funding the Group at McGill.

Since 1974, three senior medical geneticists have taken a turn as Professor of Medical Genetics at Memorial University. George Fraser and Clarke Fraser were each awarded Development Grants by the MRC. Bill MacDiarmid combined chairing and developing the Department of Medicine with service and teaching in clinical genetics (30, 40). Although “emeritus” since 1985, Clarke Fraser continued to return for genetics clinics and teaching. His recommendations helped obtain a 1987 agreement from the Deputy Minister of Health to allocate monies in the range of $150,000 in the 1988 Provincial budget to fund support staff for a unified genetics clinic at Memorial University, with outreach clinics.
A number of papers contain data alerting the reader to the social affects and distribution of genetic disease in the province (5, 32, 38, 42, 48, 64, 81, 82, 99, 136, 153, 154). Others attempt to translate concepts in human genetics into lay language, to stir up interest, and to encourage lay people and primary health care workers to ask questions about familial genetic risk prior to a genetic crisis (72, 108, 131, 166, 171). Near the tip of the Northern Peninsula in St. Anthony (almost 1000 kilometres by road from St. John’s), the Grenfell Association increasingly attracts physicians who combine research in genetics with provision of diagnosis and continuing care for members of local kindreds (10, 11, 13, 28, 51, 52, 56, 57, 70, 87, 89, 94, 102, 114, 115, 116, 120, 121, 126, 127, 133, 134, 138, 143, 153, 154, 175, 180).

Readers familiar with the home base of various authors will note the extent of collaboration outside the province, the country, and the Continent (31, 35, 59, 91, 103, 128, 135, 147, 149, 158, 163, 174, 178, 181). Collaboration often commences after a lecturer’s or candidate’s visit to Memorial University (26, 35, 39, 43, 44), or after presentation of a paper at a conference outside of Newfoundland. In some instances, local geneticists did not contribute to the writing of the paper, although they may have facilitated the investigation through their personal contact and preliminary investigation of the proband’s family (34, 77, 83, 84, 92).

The St. John’s hospitals provide tertiary care for all Newfoundland patients, thus University specialists have an opportunity to follow kindreds for probands referred from all corners of the province. A number of publications include maps that can be used to illustrate “founder effect” (10, 14, 34, 80, 92, 152). Others include pedigrees illustrating dominant (6, 7, 51, 52, 59, 120, 142), recessive (83, 126) or X-linked (10) inheritance, or segregation of balanced chromosome rearrangements (19, 89, 128).

The extensive multi-generation pedigrees obtained through some investigations suggest that one can do no better, whether at home or abroad, than to start with a proband of Newfoundland ancestry if one’s goal is the investigation of segregating genetic markers. In 1987, DNA samples from individuals in kindreds with specific genetic diseases, or syndromes, including Biedl-Bardet, Huntington, polycystic kidney, and retinoblastoma, are being shared with collaborators in Mainland Canada, the U.S.A. and England for RFLP analysis and mapping the human genome.
The special bibliography to this chapter includes three 1987 references: Barbara Yaffe’s statement about the difficulties of recruiting and keeping specialists in Newfoundland (160), the response from a rural surgeon who chose Newfoundland over his native British Columbia (161), and an article by John and Kathy Sheldon (162). Primary health care workers are indispensable in the evolution of a productive university genetics group because they are the ones who make referrals after recognizing or suspecting a genetic condition. Over the years, they care for, and come to know, family members of all ages and degree of relationship. The Sheldons have served the people around Virgin Arm, New World Island since 1964. Sheldon has referred members from numerous kindreds with segregating genetic disease, including ocular albinism (11), a mild form of haemophilia, and familial polyposis. “These cases alone will provide more than enough medical interest to last a life-time” (162).

It is our intent that the reader, whether in an ivory tower or burdened by social concern - whether an established investigator or a potential student - will be curious enough to review some of the articles in our extended bibliography (Appendix B1) in areas of personal interest. Hopefully a few will react by contacting the Dean of Medicine at Memorial University in St. John’s or a specific author to investigate possibilities for genetics research, teaching, study, and service in Newfoundland and Labrador.

Penelope W. Allderdice
and W.H. Allderdice

The chronological bibliography of human and medical genetics in Newfoundland is Appendix B1.
Penelope Witte Allderdice and William Howard Allderdice

Penny Allderdice entered human cytogenetics through postdoctoral training with O. J. Miller in New York in 1965. Miller’s group excelled in human gene mapping through cytogenetic analysis of somatic cell hybrids. In 1973, Penny was appointed to the Faculty of Medicine at Memorial University in St. John’s, Newfoundland, where she directs the provincial Cytogenetics Laboratory. Service on the National Research Council provided the opportunity to participate in research policy development across Canada while her own research was supported by the Medical Research Council and a Basil O’Connor Starter Research Award. The Newfoundland and Labrador Women’s Health Education Project alerted the Allderdices to the significant level of functional illiteracy and general health knowledge in the adult population and led to the development of a genetics participatory education project, “Ask Your Family Tree,” funded by Canada Health and Welfare’s Health Promotions Directorate. William Allderdice teaches economic development at Memorial University. Recently, the Allderdices reviewed their interactions in Newfoundland in a joint project: “Transforming Theory into Preventive Genetics in Rural Communities.”
HUMAN GENETICS IN NEWFOUNDLAND AND LABRADOR - ORIGINAL ARTICLES (References for Chapter 12)

1909 to 1967

1969

1970

1971

1972

1973


1974


1975


1976


1977


1978


1979


1980


1981


1982


1983


1984


108. Worrall G: Genetic defects to the third generation are sought. Med Post Sept 18, 1984; p92.


1985


1986


1987


1988


1989


1990


1991
