NEWFOUNDLAND AND LABRADOR
MEDICAL ASSOCIATION

FACT SHEET- Medical Genetics

Medical Genetics

- Medical Geneticists study biological variations and identify gene mutations that cause inherited diseases in families and communities. They specialize in diagnosing and prescribing therapy for patients with genetic-linked diseases.[i]

Dr. Bridget Fernandez

- Dr. Bridget Fernanadez's research interests include inherited forms of pulmonary fibrosis and aneurysms, clinical dysmorphology, autism spectrum disorders and the burden and epidemiologic distribution of genetic diseases in the province. Like many other clinical geneticists, she is interested in characterizing new genetic syndromes and in better understanding the natural history of known syndromes.[ii]

- Dr. Fernandez is a collaborator in a multicenter study headed by Drs. Stephen Scherer (University of Toronto) and Peter Szatmari (McMaster University) looking at the genetics of autism. She is also a team member of the Atlantic Medical Genetics & Genomics Initiative, developed to systematically identify genes and genetic mutations related to monogenic disorders in communities throughout Atlantic Canada. Knowledge generated from the initiative is transferred to local health care providers to improve clinical management.[iii]

Provincial Medical Genetics Program

- The Provincial Medical Genetics Program was established in 1986. The province has never had more than two practicing clinical geneticists at one time. The program is currently staffed by two clinical geneticists and eight genetic counselors. Following Dr. Fernandez's resignation on February 4, 2010, the program will down to one full-time practicing medical geneticist.

- The Medical Genetics Program includes:

    - **Hereditary Cardiac Genetics:** (inherited cardiomyopathies, inherited rhythm disturbances of the heart, Marfan syndrome, other genetic disorders of blood vessels).
    - **Hereditary Cancer Genetics:** (breast/ovarian cancer, colon cancer and gastric cancer syndromes).
    - **Endocrine tumor syndromes:** (clinical care of patients with endocrine tumor syndromes, Multiple Endocrine Neoplasia, von Hippel-Lindau syndrome)
    - **Neonatal services:** Provincial Newborn Screening (to detect children with treatable enzyme deficiencies at birth), consults for infants with birth defects.
    - **Pediatric services:** consults for children with intellectual disabilities, loss of developmental milestones, autism, birth defects, growth abnormalities or seizures where the referring physician suspects a genetic etiology.

- **Metabolic Genetics**: long-term follow-up of children and adults with genetic enzyme deficiencies.
- **Infertility**: genetic testing mandatory for the couple prior to in vitro fertilization if no etiology for the infertility has been identified.
- **Preconceptual genetic counseling**: for couples wanting to have children, where there is a known genetic disorder in the family.
- **High Risk Obstetrics consults**: Provincial Maternal Serum Screening Program, consults for pregnant women with birth defects identified on fetal ultrasound.
- **Neurology**: consults for patients with possible Huntington disease, genetic muscle diseases, genetic diseases of the brain or nerves.

Genetic Diseases in Newfoundland and Labrador

- A genetic disorder is a disease caused by a different form of a gene called a variation, or an alteration of a gene called a mutation. Many diseases have a genetic aspect. Some, including many cancers, are caused by a mutation in a gene or group of genes in a person's cells. These mutations can occur randomly or because of an environmental exposure such as cigarette smoke. Other genetic disorders are inherited, whereby a mutated gene is passed down through a family and each generation of children can inherit the gene that causes the disease.[iv]

- The population of Newfoundland and Labrador is genetically isolated and serves as an excellent resource for identifying of genes implicated in common diseases[v] that could affect up to 60% of our population.[vi] It has been estimated that 90% of the current population has arisen from 20,000 to 30,000 original European settlers of predominantly English and Irish decent.[vii] Mating segregation, combined with low immigration, and geographical isolation of communities resulted in the genetic isolation of the population.[viii]

- When a limited number of individuals bring a disease mutation into a small population, the population grows through natural expansion and (in the absence of significant in-migration) a high proportion of people will carry the chromosome on which a disease mutation is found.[ix]

- A founder effect has been observed in Newfoundland for many genetic disorders, including a variety of hereditary cancers, heart disease, kidney disease and psoriasis. Other genetic diseases, especially those that have a relatively high incidence in the population, include nonsyndromic deafness, various forms of retinal dystrophy, neuronal ceroid lipofuscinosis, neural tube defects and Bardet-Biedl Syndrome.[x]

- Bardet-Biedl Syndrome (BBS) is a rare genetic disease affects six people per million worldwide; however, in Newfoundland and Labrador the incidence is 10 times that of other populations of northern European ancestry.[xi]

- The province's annual incidence rate of juvenile Type 1 diabetes also represents the highest known incidence of the disease in North America. Similar trends have been noted in psoriasis, where the prevalence of disease is 5- to 10-fold higher in Newfoundland and Labrador than in most other Caucasian populations.[xii]

Hereditary Cancer

- When mutations occur in the damage-controlling genes, cells can grow out of control and cause cancer. For most people who develop cancer, the cancer-causing gene mutations happen over the course of a lifetime, leading to cancer later in life. However, some people are born with a gene mutation that they inherited from their mother or father.[xiii]

- The medical community uses the term "genetic susceptibility" to describe the high risk for cancer in people with an inherited mutation. Experts believe that about 10% of most cancer types are due to inherited gene changes. People with an inherited gene change have a 50% chance of passing the mutation on to each of their children.[xiv]

- Medical Geneticists use genetic testing to identify some of these inherited gene mutations to determine whether some people have an increased chance of developing the disease.[xv]

Heart Disease

- Most people think of heart disease as one condition. But in fact, heart disease is a group of conditions affecting the structure and functions of the heart and has many root causes.[xvi]

- When it comes to heart disease, lifestyle usually trumps genetics; however, the risk of heart disease is significantly increased if close family members – parents, siblings or children – developed heart disease before age 55 or, in the case of female relatives, before menopause. The risk of stroke is increased if close family members had a stroke before age 65.[xvii]

- Identifying unique gene mutations that predispose people to congenital heart disease or risk factors such as high cholesterol, high blood pressure and diabetes, allows medical geneticists to better predict a patient's susceptibility to heart disease and stroke. It also enables them to recommend appropriate therapies such as implantable defibrillators to prevent sudden cardiac arrest in patients with congenital heart disease.[xviii]

[i] Royal College of Physicians and Surgeons, Directory of Fellows, Specialty Description, 2010.
[ii] Faculty of Medicine, Memorial University of Newfoundland and Labrador, http://www.med.mun.ca/Medicine/Faculty/Fernandez,-Bridget.aspx
[iii] Atlantic Medical Genetics and Genomics Initiative (AMGGI), http://www.genomeatlantic.ca
[iv] Medline Plus, A service of the U.S. National Library of Medicine, National Institutes of Health, http://www.nlm.nih.gov/medlineplus/geneticdisorders.html
[v] Human Molecular Genetics, Proton Rahman, Albert Jones, Joseph Curtis, et al. The Newfoundland population: a unique resource for genetic investigation of complex diseases, Vol. 12, Review Issue 2 R167–R172, 2003
vi Heutink, Peter and Oostra, Ben A., Gene finding in genetically isolated populations, Institute of Clinical Genetics, Erasmus MC, Rotterdam, The Netherlands, Oxford University Press Human Molecular Genetics, 2002, Vol. 11, July 17, 2002, No. 20 2507–2515
[vii] PATRICK S. PARFREY, WILLIAM S. DAVIDSON,1 and JANE S. GREEN, Clinical and genetic epidemiology of inherited renal disease in Newfoundland, Department of Medicine and Biochemistry, Kidney International, Vol. 61 (2002), pp. 1925–1934,Perspectives in Renal Medicine, 2001.
[viii] American Journal of Human Genetics, A Founder Effect in the Newfoundland Population Reduces the Bardet-Biedl Syndrome I (BBS1) Interval to 1 cM, Young, Woods, Parfrey, et al., 65:1680–1687, 1999.
ix Human Molecular Genetics, Proton Rahman, Albert Jones, Joseph Curtis, et al. The Newfoundland population: a unique resource for genetic investigation of complex diseases, Vol. 12, Review Issue 2 R167–R172, 2003
x American Journal of Human Genetics, A Founder Effect in the Newfoundland Population Reduces the Bardet-Biedl Syndrome I (BBS1) Interval to 1 cM, Young, Woods, Parfrey, et al., 65:1680–1687, 1999.
xi Ibid.
xii Human Molecular Genetics, Proton Rahman, Albert Jones, Joseph Curtis, et al. The Newfoundland population: a unique resource for genetic investigation of complex diseases, Vol. 12, Review Issue 2 R167–R172, 2003
xiii FORCE: Facing Our Risk of Cancer Empowered, http://www.facingourrisk.org
xiv FORCE: Facing Our Risk of Cancer Empowered, http://www.facingourrisk.org
xv Canadian Cancer Society, 2010
xvi Heart and Stroke Foundation of Newfoundland and Labrador
xvii Heart and Stroke foundation of Canada
xviii Ibid.