

In the last 40 to 50 years the field of medical genetics has grown from genetic counseling services and biological laboratories to a comprehensive discipline in mainstream medicine. Accelerated discoveries in molecular genetics and immense advances in research technology are leading the way toward new methods of diagnosis, treatment and prevention of genetic disease and drug related adverse events. This timeline aims to provide an historical summary on the progress of the discipline with a focus on the significant research conducted in Newfoundland and Labrador.

1967: Faculty of Medicine established at MUN

Late 1960's - 1970's, clinical studies allowed genetic researchers to identify and diagnose diseases and syndromes

1970's: Through visiting rural communities, high incidence of various conditions were noticed in some families



Identifying family pedigrees were paramount in studying genetic conditions and involved traveling to communities, interviewing patients and their families, and going through paper records (i.e. birth, death and marriage certificates)

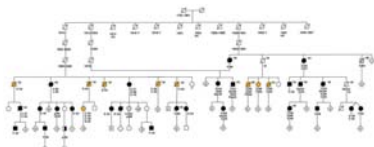
Cytogenetics - study of structure of chromosome material - was a predominant form of genetic research

Cytogenetic maps - diagrams identifying chromosomes based on banding patterns - allowed researchers to look for chromosomal alterations

One of the first significant genetic studies in NL used cytogenetics to identify the cause of severe disabilities among residents of Sandy Point: a pericentric inversion on chromosome three. This condition is later coined Allderdice Syndrome, after Dr. Penny Allderdice

1979: Ocular Genetics Clinic begins

1980's: Compilation of large family pedigrees allowed researchers to collaborate on linkage studies and map specific genes, which later gave rise to gene identification



Gene mapping studies identify closely linked markers for presymptomatic diagnosis of gene carriers, and eventual cloning of the disease gene

1980's: Clinical studies in genetics remain predominant

DNA samples had to be analyzed outside of the province

DNA was examined through linkage analysis



Advances were being made in molecular cytogenetics, which combines molecular biology with cytogenetics

1985: Familial Polyposis (FAP) study begins

Two rare recessive disorders with increased frequency were identified:

- *Bardet Biedl Syndrome (BBS)
- *NL Rod-Cone Dystrophy (NFRCD)



DNA sequencing allowed researchers to determine the number of mutations in genetic isolates

1986: Multiple Endocrine Neoplasia (MEN 2) begins

1987: Multiple Endocrine Neoplasia (MEN 1) study begins

Colorectal cancer work begins

DNA samples from individuals in kindreds with genetic diseases including BBS, Huntington, polycystic kidney and retinoblastoma were shared with national and international collaborators for RFLP analysis and mapping the human genome

1988: Provincial budget provided funding to support staff for unified genetic clinic at Memorial University, with outreach clinics

Late 1980's characterized as period of accelerating change in applications of new molecular technologies

1990's: Onset of molecular genetics studies

Beginning of molecular genetics of BBS led to recognition of role of cilia in development

Human Genome Project begins

1990's: Hereditary Non-Polyposis Colon Cancer (HNPCC) project begins

DNA microarrays are invented

1993: First HNPCC gene identified



1995: Automated sequencing technology allows Genome project to accelerate

Newfoundland acquires DNA sequencing lab

2000: Population based genetic research leads way towards personalized medicine

Genetic research begins to branch from studying single gene diseases (i.e. Huntington's disease) to complex diseases (i.e. cancer, arthritis and obesity)

Researchers study candidate genes as opposed to conducting association studies

2001: The sequence of the human genome is released and "post-genomic era" officially begins

Colorectal Cancer (CRC) study begins

2003: A research team, led by Memorial University rheumatologist, discovers new gene for psoriatic arthritis

2004: Pharmacy Research Database (PRD) is funded through Atlantic Innovation Fund; this marks the beginning of the Population Therapeutics Research Group

2006: Memorial University partners in the Atlantic Medical Genetics and Genomics Initiative (AMGGI), a pan-Atlantic effort to identify gene mutations that cause inherited diseases in Atlantic Canadian communities

2007: Memorial genetic researchers help find genetic marker for colon cancer

Rare stomach cancer mutations identified in CDH1

Identification of gene mutation responsible for congenital indifference to pain (CIP)