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Genomic Tools for Diagnosis and Evaluation of Mental Retardation

Status	Current
Competition	Applied Genomics and Proteomics Research in Human Health
Sector	Health
Genome Centre	Genome British Columbia
Project Leaders	Jan Friedman & Marco Marra

Project Description

Improving diagnoses and evaluation of mental retardation

Mental retardation is a life-long disability that affects more than 300,000 Canadians. While the cause of most mental retardation remains unknown, chromosomal abnormalities like Down syndrome are the most frequently recognized cause. “For more than 40 years, we have used a process called karyotyping to detect chromosomal abnormalities,” explains Dr. Jan Friedman of the University of British Columbia. “Unfortunately, karyotyping is a technically demanding microscopic technique that requires highly skilled interpretation. In addition, some children with mental retardation have loss or gain of chromosomal material that is too small to see with karyotyping. We believe that new genomic tests will allow us to identify these chromosomal changes, especially the subtle ones, more effectively than by karyotyping.”

Led by Drs. Jan Friedman and Marco Marra, the Genomic Tools for Diagnosis and Evaluation of Mental Retardation project aims to develop an alternative to karyotyping to identify chromosomal abnormalities in people with mental retardation. The project will evaluate a new testing method to identify chromosomal abnormalities 100 times smaller than those detectable by karyotyping.

The project includes the implementation of a specialized clinical and cytogenetic research database at all participating centres to coordinate collection of clinical data and specimens. And a GE3LS (Genomics and Ethical, Economic, Ecological, Legal and Social) issues component will assure that the nationwide exchange of clinical research data and specimens is done in an appropriate legal, ethical and social context that reflects Canadian and international norms. A Health Technology Assessment component will provide the expertise necessary to determine whether patients, their families, and the Canadian health care system would benefit by replacing routine karyotyping with new technologies for the identification of chromosomal abnormalities in people with mental retardation.