



Another project brought to you by **GenomeCanada**

The Contribution of Genetic Modulators of Disease Severity in Cystic Fibrosis to other Diseases with Similarities of Clinical Phenotype

Status	Current
Competition	III
Sector	Health
Genome Centre	Ontario Genomics Institute
Project Leaders	Peter Durie & Julian Zielenski

Project Description

Canada is a world leader in research on cystic fibrosis (CF). Drs. Peter Durie, a pediatrician and senior scientist and Julian Zielenski a geneticist at the Hospital for Sick Children's Research Institute plan to build on this research strength, by investigating the genetics of other diseases with similar phenotypes – observable physical characteristics, which may be genetically determined.

Drs. Durie and Zielenski are project leaders of *The contribution of genetic modulators of disease severity in cystic fibrosis to other diseases with similarities of clinical phenotype*.

This project will apply knowledge about the genetic factors (so called modifier genes) that influence the severity of CF to other diseases that are clinically similar to CF. These diseases include a single-gene disorder affecting the liver (α 1-antitrypsin deficiency), and multifactorial conditions such as pancreatitis due to alcohol abuse and chronic obstructive pulmonary disease due to smoking. The project will analyse mutations in the Cystic Fibrosis Transmembrane Conductance Regulator gene (CFTR) as well as selected modifier genes that are found to influence the severity of disease in patients with CF as well as blood-circulating proteins, in order to identify disease biomarkers, which can help predict disease severity and progression. Diagnostic and prognostic tests will be developed, and genetic test-based risk identification could lead to behaviour modification and disease prevention among those at risk for the diseases.

Enormous human suffering and prohibitive healthcare costs are associated with alcohol abuse and tobacco smoking. This project is expected to yield results of world wide importance, such as development of genetic tests of disease susceptibility that will be useful in future research projects and in development of preventative strategies to modify behaviour in high risk populations. This in turn should lead to reduced morbidity and mortality and more efficient healthcare. Important components of the project are ethical issues associated with genomics research, as well as industrial, economic and social benefits.