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Mapping and Isolation of Genes Influencing Severity of Disease in Cystic Fibrosis

Status	Past
Competition	II
Sector	Health
Genome Centre	Ontario Genomics Institute
Project Leader	Peter Durie and Lap-Chee Tsui

Project Description

An important aspect of the Human Genome Project lies in its promise of better understanding and treatment of human diseases. The more we know about the genes that underlie disease, the more skillfully we can devise treatments. It is rare for a genetic disease to be caused simply by an error in one gene. While a single-gene defect may be the primary cause, the complexity of the human body almost ensures that other genes will be involved. A case in point is the gene which, when defective, causes Cystic Fibrosis (CF). Due to the pioneering work in the 1980s of researchers at the Hospital for Sick Children we know that all Cystic Fibrosis patients are defective in a gene called CFTR, which instructs the cell to make a protein that moves ordinary chlorine ions into and out of cells. Yet, even in patients with the identical genetic change in CFTR, the severity of Cystic Fibrosis can be very different. The reason must be that other genes interact with CFTR to alter the course of the disease.

Our aim was to use the most up-to-date genomic methods to find as many of these other genes as we could. This kind of study thrives on close co-operation between medical doctors and research scientists. It requires very careful diagnosis and description of disease symptoms. Underneath this must lay a strong organization that can identify patients and their families all across Canada, obtain samples from them for genetic analysis, and record and analyse the results.

We collaborated with all 38 Canadian Cystic Fibrosis clinics to establish a study group of over 75% of the entire Canadian CF population. All together, we collected blood samples from almost 2800 CF families, including some from foreign sources. We extracted DNA from these and also established tissue-culture cell samples that can be kept for future research; this is now the world's largest resource for CF genetic studies. Our analysis of the extracted DNA using sophisticated genetic techniques has identified almost 100 genes that potentially could affect the progression of CF.

Fast Facts

<i>Highlighted outcome:</i>	Creation of the world's largest repository of family-based cell lines for clinical-genetic studies; the identification of genes that potentially affect the severity of Cystic Fibrosis
<i>Number of research personnel employed by the project:</i>	19
<i>Number of peer reviewed publications published:</i>	1 plus 4 indirectly
<i>Resources generated:</i>	World's largest repository of family-based cell lines for clinical-genetic studies; the Cystic Fibrosis Mutations Database; the Canadian Consortium for Cystic Fibrosis Genetic Studies
<i>Number of public outreach events held:</i>	27 (public lectures, magazine articles, website, communications with CF families)
<i>Co-funders:</i>	Canadian Foundation for Cystic Fibrosis; Hospital for Sick Children Foundation