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Structural and Functional Annotation of the Human Genome for Disease Study

Status	Current
Competition	III
Sector	Health
Genome Centre	Ontario Genomics Institute
Project Leader	Rob Hegele

Project Description

Now that the human genome has been sequenced, the next step is to undertake the complete structural and functional annotation of genes associated with diseases, according to Dr. Robert Hegele, endocrinologist and scientific director of the London Regional Genomics Centre at the Robarts Research Institute. Dr. Hegele is project leader of Structural and Functional Annotation of the Human Genome for Disease Study, an innovative project which aims to bridge new biological knowledge with medical applications.

Any two humans are 99.9% identical at the level of their DNA sequences. But recently, new forms of genomic variation have been appreciated above and beyond single nucleotide polymorphisms. These include large scale variations, such as copy number changes, insertions, deletions, duplications and rearrangements, and they may be much more widespread than was previously appreciated. In this project, collaborator Dr. Steve Scherer of the Hospital for Sick Children will define and superimpose these large scale genomic variations over top of the existing “first draft” of the human genome sequence map.

Another form of genome variation occurs through a process called “alternative splicing”, which gives rise to multiple versions of a protein encoded by a single gene. Also, some parts of the genome previously thought to be dormant are now known to code for active proteins functioning in the body.

Collaborators Dr. Ben Blencowe, Dr. Tim Hughes and Dr. Brendan Frey of the University of Toronto will define and integrate these new forms of genomic variation into the current human genome sequence map.

The project will therefore deliver a “new improved edition” of the human genome map; one that annotates and characterizes large-scale copy number variants, alternative splicing profiles of genes in selected tissues and previously unknown genes and other functional elements.

Dr. Hegele and collaborators will then apply the annotated genome map with its rich trove of new biological information to unravel the genetic basis of diseases that extract a huge social and economic toll in Canada, such as diabetes, heart disease and breast cancer.

The data generated from the project will be made available, free of charge, on the Internet, in order to accelerate biomedical discovery, including the diagnosis and treatment of common diseases.