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Regulatory Genetics: Identification of Regulatory Polymorphisms in the Human Genome

Status	Past
Competition	Competition I
Sector	Health
Genome Centre	Genome Quebec
Project Leaders	Thomas Hudson & Daniel Sinnett

Project Description

At the start of this project, not much was known about the association of gene regulation with certain diseases. However, early research had suggested that most relevant mutations reside in regulatory regions. The objective of the Regulatory Genetics Project (RegGen) was to identify genetic variations that affect gene regulation and potentially cause common, complex diseases.

The Regulatory Genetics program has been very successful. We have discovered genes with a regulatory 'haplotype' – variations that are clustered into blocks of DNA – several of which are implicated in common diseases, such as asthma and leukemia. These will be the basis for a wide range of scientific projects in pharmacology, genetics, medicine, cellular biology and computation biology that will further the understanding of complex diseases and drug response.

We have also associated several SNPs – variations within a single nucleotide – with diseases. These findings can be developed for further use in public health (risk identification for chronic diseases) and in a clinical setting (diagnosis, pharmacogenetics, and drug therapies). Although additional validation will be required, the timely identification of people at risk for a chronic disease has the potential for a large economic savings. Industry will likely be involved in the development of specific screening kits.

The project has developed several technologies and software that are used worldwide and have so far generated about \$300k in licensing revenues. Most importantly, we've built an inter-university group of collaborators who now work in the important field of gene regulation in relation to diseases. RegGen's scientific contributions have increased our insight into the field of regulatory genetics and enriched the academic environment, not only at McGill and the Université de Montréal, but also internationally.

Fast Facts

Highlighted outcome: discovery of genes with a regulatory ‘haplotype’ – variations that are clustered into blocks of DNA – several of which are implicated in common diseases, such as asthma and leukemia

Number of research personnel employed by the project: 30

Number of peer reviewed publications published: 14 articles, 1 book, 50 abstracts, and 51 invited presentations

Number of patents in process or obtained: 1 disclosure, 1 filed, 1 published and 1 commercial license in place