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Genetic Dissection of Complex Traits Using Phenotypic and Expression Analysis of Recombinant Congenic Mouse Strains

Status	Past
Competition	Competition II
Sector	Health
Genome Centre	Genome Quebec
Project Leaders	Emil Skamene

Project Description

Most human diseases are multifactorial and genetically complex. These complexities confound many studies to investigate the basis for inherited predisposition to disease in humans. One approach for identifying genes controlling complex traits and narrowing the search for candidate genes is “mouse-man homology”, where genetic information derived from mouse models is applied to identify candidate genes in the human.

Our team proposed to use Recombinant Congenic Strains (RCS) to dissect complex traits and accelerate and broaden the scope of target discovery, in collaboration with academic institutions and investigators. The project brought together a network of Canadian researchers and industrial partners for the purpose of identifying novel targets to treat common human diseases. The synergy between the different research groups has contributed to the retention in Canada of highly qualified personnel and trainees.

Our findings could lead to better access to early diagnosis and the development of new treatments for common human diseases. Even though most of the projects involving the RCS platform are still at early stages, a few are starting to point out direct applications.

For instance, one team has demonstrated that early replication of malaria parasites can be reduced by the administration of cysteamine. Directly arising from a gene discovery effort in RCS mice, this discovery could have a strong impact on the prevention and treatment of one of the most deadly diseases world-wide and its commercial potential is now being investigated. In addition, another team is working of the development of a treatment for severe pyruvate kinase deficiency in humans using one of our strains as a mouse model.

We anticipate that the RCS platform will continue to lead to more discoveries, which will, in turn, lead to applications for diagnosing and treating common human diseases.

Fast Facts

Highlighted outcome:	the advancement of our understanding of the genetic regulation of several complex human diseases
Number of research personnel employed by the project:	63.34 person years (calculated as sum of FTEs per quarter per category x 0.25 year)
Resources generated:	world-wide distribution and use of 36 recombinant congenic mouse strains derived from our gene discovery platform, available to the research community upon request; genotyping, phenotyping and expression data coming from the RCS set, which will be made available to the scientific community once commercial potential is protected
Number of patents in process or obtained:	1 patent disclosure, 1 provisional patent, 1 patent filed, 33 commercial licenses in place, 1 company formed
Co-funders:	Emerillon Therapeutics Inc. (formed by parent company Xenon Genetics Research Inc.)