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## Genetic Determinants of Human Health and Disease

<b>Status</b>	Past
<b>Competition</b>	Competition II
<b>Sector</b>	Health
<b>Genome Centre</b>	Ontario Genomics Institute
<b>Project Leaders</b>	Katherine Siminovitch

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### Project Description

Our research program explored gene variants causing and/or modulating a number of common multigenic disorders, including Alzheimer's, inflammatory bowel disease, cancer (breast, endometrial, prostate, ovarian, melanoma) and osteoporosis. We also developed novel biometric and computational tools for expediting disease gene discovery.

The disorders were selected on the basis of their prevalence and very significant morbidity, the availability of large patient populations, significant prior knowledge obtained by team members, and the urgent need for and potential major socioeconomic and medical impact of new diagnostics and therapeutics for any one of these conditions.

Results of our project have created new knowledge relevant to the medical care of a number of common debilitating diseases. The project findings include the discovery of several genes and proteins involved in inflammatory bowel disease and Alzheimer's disease, as well as gene variants associated with risk for a set of common cancers and metabolic bone disease.

These data provide a platform for the development of tools for the diagnosis of certain conditions and for identifying molecular targets for therapeutic intervention. Discoveries of genetic variants associated with such common cancers as prostate and breast cancer have important clinical implications, all of this knowledge paving the way for predictive or diagnostic tests relative to these diseases.

In addition to knowledge relevant to the diagnosis and treatment of specific diseases, this program has provided new information and methods to improve technical platforms for genotyping, gene expression profiling and the computational management and statistical analyses of clinical and genetic datasets. Such tools are key to the exploitation of the human genome sequence for the purposes of disease gene discovery and, ultimately, for translation of genomic information to clinical and therapeutic applications.

## **Fast Facts**

***Highlighted outcome:*** new knowledge relevant to the medical care of a number of common debilitating diseases, including Alzheimer's, inflammatory bowel disease, cancer (breast, endometrial, prostate, ovarian, melanoma) and osteoporosis

***Number of research personnel employed by the project:*** 57

***Number of peer reviewed publications published:*** 16 journal articles, 20 book chapters or contributions to collective works, 23 abstracts or notes, and 109 invited presentations

***Number of patents in process or obtained:*** two applications filed for methods for diagnosing Inflammatory Bowel Disease, and two novel patents granted related to intestinal peptides