Genetics

CIHR IRSC

The Canadian Institutes of Health Research (CIHR) is the Government of Canada's agency for health research. Through CIHR, the Government of Canada invested approximately $243 million in 2006-07 across Canada in research on genetics.

The Facts

- The human genome is made up of 3 billion (3,000,000,000) bases (letters) of DNA, split into 24 distinct chromosomes.
- This information…
  - would fill two hundred 500-page telephone directories.
  - would take a century to recite, if recited at a rate of one letter per second, 24 hours a day.
- The human genome contains about 30,000 genes.
- Between humans, our DNA differs by only 0.1%, or 1 in 1,000 bases.
- Genes contain instructions for how to make proteins which are responsible for carrying out all of the cell functions (examples – facilitating chemical reactions, controlling growth and transporting substances through the body).
- Many diseases stem from problems associated with proteins (examples – too many proteins produced, too few proteins, the protein is the wrong shape or "misfolded").
- The proteome is the complete set of proteins produced by the genome at any one time, approximately 1,000,000.
- Proteomics is the study of proteins – what proteins look like (structure), interactions between proteins and the types of proteins expressed in healthy vs. diseased tissues.

Canadian Institutes of Health Research
160 Elgin St., 9th Floor, Ottawa ON K1A 0W9
www.cihr-irsc.gc.ca

About CIHR

The Canadian Institutes of Health Research (CIHR) is the Government of Canada's agency for health research. CIHR's mission is to create new scientific knowledge and to catalyze its translation into improved health, more effective health services and products, and a strengthened Canadian health-care system. Composed of 13 Institutes, CIHR provides leadership and support to more than 11,000 health researchers and trainees across Canada.

Finding Solutions

How do medications work?

Drug researchers can learn a lot from the medications that we already use. CIHR-supported researchers Drs. Charlie Boone and Brenda Andrews of the University of Toronto are building a collection of "chemical-genetic interaction" profiles, descriptions of how therapeutic chemicals affect gene activity. These profiles, which the researchers are generating using genetically altered yeast cells, could be used in the future to identify new drugs, determine how those drugs work, and anticipate possible negative side effects. These profiles have already uncovered how a common breast cancer treatment and an anti-HIV drug work.

Conducting respectful research

Who "owns" the DNA samples that researchers study? CIHR-supported researcher Dr. Laura Arbour of the University of British Columbia studied this important issue and provided a number of recommendations that helped shape the new *CIHR Guidelines for Health Research Involving Aboriginal Peoples*. According to Dr. Arbour, researchers must view biological samples obtained from aboriginal communities as "DNA on loan", and any research carried out using these samples must reflect the needs and priorities of the community. Researchers should also take steps to involve community members in the research process and share any study results with the community.

Using genetic technology wisely

Researchers at Laval University are looking for the best ways to put our genetic know-how to work in the fight against breast cancer. Genetic screening may help save many lives, but it is expensive and time-consuming. Dr. Jacques Simard and his team are trying to find an effective way to identify high-risk populations that would benefit most from screening for mutations in BRCA1 and BRCA2, two genes commonly linked to breast cancer. In a study of 256 French-Canadian families from around Quebec, Dr. Simard identified eight new mutations linked to breast cancer and tested the effectiveness of three mathematical models for predicting breast cancer risk. His results indicate that doctors should test patients with a family history of breast cancer for common breast cancer gene mutations, and then use the mathematical models to determine which patients should receive further testing.

The Researchers

Dr. Stephen Scherer – Scanning the genetic horizon

Dr. Stephen Scherer, a CIHR-funded researcher at Toronto's Hospital for Sick Children, is pretty good at predicting the future in his research field. But every now and then, something surprises him. Last year, he and his colleagues discovered much more variety in the human genome than previously thought.

Geneticists thought the genome contained two copies of most of its genes. But Dr. Scherer and his colleagues recently published a new map of the human genome showing that individuals often have multiple copies of a single gene, or even have missing genes. These repetitions and deletions are called copy number variations, or CNVs; it appears they account for much of the genetic diversity from person to person.

"I cannot believe that two years ago we didn't know that this type of variation existed in the genome," he says. "That's something that just hit us over the head, and now it's changing the whole field."

Dr. Scherer and his team are now investigating how CNVs may contribute to disease. He's part of an international team scanning the human genome in search of the genetic causes of autism. Their effort has already pinpointed several CNVs which appear to play a role in this developmental disorder. The findings may produce improved diagnostic tests and a better understanding of how various genes interact to cause autism.

Dr. Scherer's lab will be using similar technology to study the genetic roots of many diseases, from schizophrenia to cancer. He's also focused on developing "personalized" DNA scanning technology, which may someday make full-genome diagnostic scans a medical reality.

"It's quite likely that in ten years or so we may be able to offer to sequence your newborn child's DNA before you leave the hospital," says Dr. Scherer. Who would have been able to predict that?