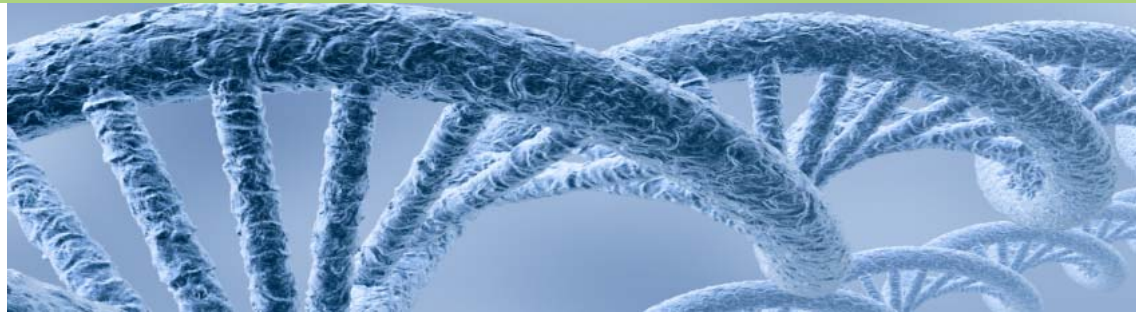


Research About – Genetics

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 nearly 12,000 health researchers and trainees across Canada.

Through CIHR, the Government of Canada invested approximately **\$262.5 million** in 2007-08 in genetics-related research across Canada.



The Facts

- The human genome contains about 30,000 genes.
- Genes contain instructions for how to make the proteins that carry out all of a cell's functions, such as facilitating chemical reactions, controlling growth and transporting substances through the body.
- Canadian scientists have been leaders in the study of human genetic diseases for the last 40 years, and have participated in the identification of many important disease genes, including those for cystic fibrosis, muscular dystrophy, Alzheimer's disease, breast cancer, inherited forms of blindness and many types of congenital malformations.
- The information obtained from the Human Genome Project allowed Canadian researchers recently to identify genes associated with common diseases, including Alzheimer's disease, asthma, diabetes and coronary artery disease.
- Canadian investigators have also been international leaders in the discovery of genes required for the normal development of many organs, and in recent years have also characterized genes important to many types of stem cells.



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Finding Solutions

Keeping GPs in the gene loop

CIHR-supported researchers are helping keep family physicians up-to-date on the state of genetic research. Dr. June Carroll of the University of Toronto and Drs. Judith Allanson and Brenda Wilson of the University of Ottawa created GenetiKit to help doctors answer patients' questions about the genetic causes of disease. The toolkit contains information aids about the risk of inheriting breast or colorectal cancer, and a table outlining the possible consequences of genetic testing. With trials complete in Ontario, the researchers are evaluating the project and gathering information on how doctors rate the toolkit.

Reference checks for proteins

A research team from the Institut de recherches cliniques de Montréal (IRCM) has developed a new way to unlock the secrets of previously puzzling proteins. The procedure involves tracking how the mystery proteins interact with protein partners whose functions are already understood. Identifying the functions of these unknown proteins is important: diseases often are caused by aberrations in the structure of key cellular proteins. The team is led by CIHR-supported researcher Dr. Benoit Coulombe.

Genetic discovery prompts life-saving surgeries

Memorial University researchers have discovered a gene responsible for a deadly heart condition highly prevalent in Newfoundland and Labrador. Knowledge of the location of the gene responsible for arrhythmogenic right ventricular cardiomyopathy has made it possible to identify carriers. Doctors are now implanting defibrillators in adult carriers so that a shock is delivered to the person's heart should ventricular fibrillation – which is potentially fatal – occur. "We have inserted nearly 100 defibrillators in mutation carriers, and clearly demonstrated that lives were saved," said Dr. Sean Connors, a cardiologist and member of the research team.

The Researchers

Dr. Michael Hayden – Avoiding medicine that harms

Imagine taking a medication that's supposed to help you, but that weakens your heart or destroys your hearing instead. Adverse drug reactions (ADRs) like these are far from rare: ADRs are the fifth-leading cause of death in the developed world. Children are at particular risk; in Canada, one-in-seven hospitalized children experiences an ADR.

But research by CIHR-supported geneticist Dr. Michael Hayden and colleagues could dramatically change this. They're creating the world's first gene-based screening tests to identify children susceptible to ADRs.

"These tests will be one of the first examples of truly personalized medicine," says Dr. Hayden, a researcher at the Vancouver-based Centre for Molecular Medicine and Therapeutics.

Every year in Canada, there are an estimated 200,000 severe ADRs which in turn cause between 10,000 and 22,000 deaths. Children are the most susceptible, says Dr. Hayden, because when it comes to pharmaceuticals they're not "small adults" but rather biologically different based on their developmental age.

"The problem is that 75% of existing drugs are used in children but we don't conduct clinical trials with these drugs in children,"



he says. "Essentially we're treating children as guinea pigs."

In order to identify the gene sequences associated with ADRs in children, Dr. Hayden teamed-up with Dr. Bruce Carleton, a pharmacology researcher at the University of British Columbia and the Children's Hospital in Vancouver, to create a unique pan-Canadian research program, called the Genotype-specific Approaches to Therapy in Childhood ADR network. The network brings together 2,300 pediatricians and 10 children's hospitals, whom together treat 75% of Canadian children.

The network has collected ADR data, including DNA samples, from more than 6,000 pediatric patients. The researchers have scoured these genetic samples looking for the DNA sequences, or markers, that are predictive of ADRs.

"Already in the earliest phase of the research we're finding DNA markers that significantly change the risk for children getting these drugs," says Dr. Hayden.

The next step is the creation of screening tests so that a child's or adult's genetic fingerprint can be used to make sure that the medications they receive help – not hurt.



For more information, go to
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