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GENES TO THERAPIES

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Canadian Researchers Lead Mapping of Human Chromosome 7 Key Milestone Achieved in Bid to Identify New Disease Genes

Led by Canadian researchers **Steve Scherer, Johanna Rommens, and Lap-Chee Tsui**, an international research team has published a detailed and annotated map of human chromosome 7. This collaboration involved 90 scientists in 10 countries, including Dr. Ben Koop at the University of Victoria and Celera Genomics.

"The support of the CGDN, and in particular its Victoria and Toronto DNA Sequencing facilities, was crucial for all aspects of the chromosome 7 project", stated Dr. Scherer.

Chromosome 7 is the seventh largest chromosome in the human genome. "If the genome was the size of the Earth, chromosome 7 would be the size of Canada", remarked Dr. Scherer. In fact, it contains some 158 million nucleotides and 1,445 of the estimated 28,000 genes in the genome. Three hundred sixty of these genes have been linked to diseases such as leukemia, colorectal cancer, renal cancer, deafness, ADHD, autism, epilepsy, and obesity.

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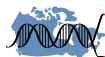
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Gene Plays Important Role in Heart Disease Network Research Pinpoints Genetic Mechanism for Heart Failure

An international team of scientists, including **Dr. David MacLennan** of the Canadian Genetic Diseases Network, has identified a genetic mutation that causes heart failure and premature death in people in their teens and 20s.

"We've shown a clear link between the disruption of calcium regulation caused by a genetic mutation in the PLN gene and progression to dilated cardiomyopathy," says Dr. David MacLennan, a

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CANADIAN GENETIC DISEASES NETWORK
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Steven Narod Appointed Tier 1 Canada Research Chair

Dr. Steven Narod, an Investigator of the Canadian Genetic Diseases Network, has been appointed to a Tier 1 Canada Research Chair for Public Health Sciences. Dr. Narod, a recognized leader in the field of breast cancer genetics, will receive the award over a seven-year period.

"This appointment to the Canada Research Chairs program recognizes Dr. Steven Narod as a world-leader in breast cancer genetics. The Canadian Genetic Diseases Network congratulates Dr. Narod for this accomplishment and acknowledges his contribution in making Canada a leader in genetic research", stated **Dr. Michael Hayden**, Scientific Director of the Canadian Genetic Diseases Network.

The Canada Research Chairs program is a \$900 million infrastructure improvement program created by the federal government. The program aims to propel Canada to the highest levels of research excellence and to become a world-class research centre in the global, knowledge-based economy.

Dr. Steven Narod's work on identifying the link between mutations in the genes BRCA1 and BRCA2 and hereditary breast cancer has opened the door for diagnostic screening that will improve health outcomes for breast cancer patients. His new research is directed at translating genetic knowledge into effective prevention and management strategies for breast and ovarian cancer. ▲

Heart Disease

Continued from page 1

professor in the Banting and Best Department of Medical Research at the University of Toronto. "Scientists at U of T and elsewhere had already learned how this process functioned in mice but this is the first time we've seen the same mechanism in people."

The research team first identified a family in which several members had developed early onset dilated cardiomyopathy. Family members who carried the normal PLN gene were healthy while those who carried a copy of the mutant PLN gene developed dilated cardiomyopathy and heart failure.

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PLN works by affecting the ability of the heart to regulate calcium. Muscle contractions are triggered by an influx of calcium into the heart. Relaxation occurs when that same calcium is pumped out of the heart. The movement of calcium out of the heart is controlled by a calcium pump, and PLN is the protein that regulates this pump.

Dr. Peter Liu, Director of the Heart and Stroke Foundation of Ontario, compared PLN to the thermostat in

your home. "PLN is the equivalent of the calcium thermostat in the heart cell. When there is a mutation in the switch regulation, it leads to heart failure. This research represents a piece in the jigsaw puzzle that heart specialists have been looking for in terms of one of the master regulators of heart function."

Previous research has identified seven or eight different gene mutations that can contribute to heart failure. **Dr. Michael Hayden**, Scientific Director of the Canadian Genetic Diseases Network, emphasized that this discovery is important in understanding how these genes work and contribute to heart failure. "This is a wonderful result. It is hoped that this result highlights pathways that will have relevance for many Canadians who suffer from heart failure."

The scientists will now use the knowledge of how PLN works to determine how other genes involved in calcium regulation might also induce heart failure. ▲

Research Labs Host High School Students During Spring Break

Sixteen high school students from across Canada learned what it's like to be a gene researcher. These students participated in a pilot program that placed up-and-coming scientists into Canada's top laboratories to encourage their interest in medical genetics research. The program is called "Gene Researcher for a Week" and it is a partnership between the Canadian Genetic Diseases Network and the CIHR Institute of Genetics.



Crystal Gennick works in a tissue culture hood at the Manitoba Cancer Research and Treatment Centre. Crystal spent her break week learning about gene research with Dr. Geoff Hicks.

Students interested in the program were invited to submit an essay describing why they wanted to be a gene researcher for a week. 330 students applied an essay, and 16 were selected into the program. "I was impressed

by the number of applications we received and the level of enthusiasm displayed by our applicants", remarked **Dr. Tom Hudson**, Associate Scientific Director of the CGDN.

Fifteen gene researchers hosted students during the spring break from March 3, to April 25. Hosting laboratories were located in Vancouver, Edmonton, Calgary, Winnipeg, Montreal, and Toronto.

Meghan Trenholm, a grade 12 student from Riverview High School in Riverview NB, spent her break week in Dr. Hudson's lab at the Genome Quebec

Centre for Innovation in Montreal. "The program gave me a whole new appreciation for gene researching – how much work goes into it, how interesting it can be, and how rewarding and beneficial it is", remarked Trenholm.

Rachel Woynorowski, a grade 12 student at Ross Sheppard High School in Edmonton, spent her break in Dr. Mike Walter's lab at the University of Alberta.

"The program gave me a whole new appreciation for gene researching – how much work goes into it, how interesting it can be, and how rewarding and beneficial it is"

"The chance to work in the lab allowed me to put my theoretical knowledge regarding genetics into practice. The ability to learn the procedures helped me to better understand the theories behind some of the concepts in genetics. It was a great opportunity to talk to people and students in the genetics field and get general information regarding career and school paths", stated Woynorowski.

With the success of the program, CGDN and the Institute of Genetics hope to gain additional support to expand the program in 2004. ▲

Pfizer Pledges Support for the Scriver MD/PhD Scholarship Program

Pfizer Canada Inc. has pledged \$120,000 to support the Scriver MD/PhD Scholarship program. The program, which aims to increase the number of students enrolled in MD/PhD training programs in Canada and to encourage graduates to pursue research careers, provides support to MD/PhD students focusing in genetics.

"We partner with over 1,000 clinical researchers, including family doctors, who through their combined medical and research expertise, are helping to raise Canada's collective standard of care, and add to this country's reputation for scientific excellence," stated Dr. Bernard Prigent, Vice President, Medical, Pfizer Canada. "We're extremely proud to be a sponsor of this very worthwhile program."

In addition to funding, Pfizer will provide an annual forum for the scholarship recipients to meet and discuss their research. "At Pfizer Canada, we know first-hand the tremendous contribution clinical researchers make to advances in healthcare science. We partner with over 1,000 clinical researchers, including family doctors, who through their combined medical and research expertise, are helping to raise Canada's collective standard of care, and add to this country's reputation for scientific excellence," stated **Dr. Bernard Prigent**, Vice President, Medical, Pfizer Canada. "We're extremely proud to be a sponsor of this very worthwhile program."

"We are pleased by Pfizer's participation in the Scriver MD/PhD scholarship program. Their support will significantly further the development of clinician scientists, whose unique training will facilitate the translation of genetic discoveries into diagnostic tests, therapies, and ultimately positive health outcomes for Canadians", stated **Dr. Ron Woznow**, CEO of the Canadian Genetic Diseases Network.

The Scriver MD/PhD Scholarship Program is named in honour of **Drs. Jessie Boyd, Walter, and Charles Scriver**. It was founded by the Canadian Genetic Diseases Network, the Canadian Gene Cure Foundation, and the CIHR Institute of Genetics. ▲

Human Chromosome 7

Continued from page 1

The project to map and sequence chromosome 7 began when Dr. Scherer and Dr. Rommens were training in Dr. Tsui's lab at the Hospital for Sick Children in Toronto. "We developed the mapping resources to complement ongoing projects that aimed to map and identify the Cystic Fibrosis (CF) gene. These same resources eventually contributed to our mapping of chromosome 7", stated Dr. Scherer. Indeed, the three and their collaborators have mapped or identified more than 20 disease genes on the chromosome.

Dr. Michael Hayden, Scientific Director of the Canadian Genetic Diseases Network, remarked, "The completion of this map is a major scientific achievement. It will facilitate the development of new diagnostic tests and innovative treatments for human diseases linked to chromosome 7." Already the mapping project has led to the development of molecular diagnostic tests for a number of genetic disorders, including those for patients with kidney tumors and adult-onset citrullinemia.

With the annotated map published, Dr. Scherer says that his lab will continue to focus on studying diseases linked to chromosome 7. He speculates that it will take years to study the remaining 1,000 genes on chromosome 7 that have not been linked to a human genetic disorder. ▲

Scholarships for Young Scientists



Rt. Hon. Don Mazankowski, Board Chair of the CGDN, talks to nearly 100 of Vancouver's business and community leaders at the fundraising dinner.

Rt. Hon. Don Mazankowski was the guest of honour at a March 4th fundraising dinner hosted by the Canadian Gene Cure Foundation. The dinner was attended by nearly 100 business and community leaders in Vancouver. Mr. Mazankowski entertained guests with stories about election campaigns, his early days in Parliament, and his role as Deputy Prime Minister of Canada. The take-home message he left with guests was that gene research is important to the well-being of all Canadians. The monies raised from the event will be used to fund an undergraduate scholarship in genetics. ▲



Left to Right: Dr. Ron Woznow, CEO of the CGDN, Dr. Indira Samarasekera, Vice-President of Research at UBC, Paul Stinson, Executive Director of BC Biotech, and Dr. Michael Hayden, Scientific Director of the CGDN.

Summer Student Program in Full Swing

Summer is here and undergraduate students are once again hustling and bustling throughout CGDN research labs. CGDN will be awarding studentships to 20 promising, young researchers in 2003. The awards provide an excellent opportunity for science undergraduate students, who possess a strong interest in genetics, to gain valuable research experience and to develop a network within the research community.

Dr. Rachel Wevrick is a CGDN Scholar and a supporter of the summer student program, "The studentship program gives students an opportunity to work on an independent research project, to work with a scientist, and to publish papers. My student, Megan O'Neill, will spend her summer researching a gene deleted in Prader-Willi Syndrome." This year, students in the program will be undertaking projects related to obesity, asthma, glaucoma, arthritis, and other genetic disorders.

Students were judged on their academic credentials and the alignment of their proposal to CGDN's strategic research objectives. "The caliber of students that applied in the first competition was excellent. 15 students have been selected into the program to date and another 5 students will be selected in May", remarked **Dr. Chrystal Palaty**, Manager of Scientific Affairs and Training at the CGDN. Past award winners have gone on to graduate studies, medical school, and also MD/PhD training programs. The summer student program runs from May to September. ▲

ANNOUNCEMENTS

Vancouver Core Facility Expands
**Now offering ES Cell work
 and Frozen Embryo
 Implantation**

The CGDN Transgenic Mouse Core Facility is now offering 2 new services, ES cell work and frozen embryo implantation. The facility serves both academic and commercial clients, and offers quality service and competitive prices. For more information, access the website (http://cmmt.ubc.ca/core_fac/trans-genic/) or contact Dr. Blair Leavitt at (604) 875-3845 or bleavitt@cmmt.ubc.ca.



Canadian
 Bioinformatics
 Workshops

Organized by the CGDN and hosted nation-wide, this highly acclaimed workshop series is now in its 5th year. Join us in 2003 for one or more of the following:

Bioinformatics

May 26 – June 7, 2003, Ottawa.
 Competition Closed.

**Bioinformatics: Introduction to
 Programming**

June 16 – 28, 2003, Vancouver.
 Apply by April 26, 2003.

Proteomics

July 7 – 12, 2003, Vancouver.
 Apply by May 16, 2003.

Genomics

August 18 – 23, 2003, Calgary.
 Apply by June 20, 2003.

For further details or to apply, visit
www.bioinformatics.ca



GenInfo is a bimonthly electronic newsletter concentrating on new policy statements on human genetics from international, regional and national sources. GenInfo is intended to facilitate communication and to inform policy makers, researchers, health professionals, and the general public. Subscription to GenInfo is free, however email address is required in order to obtain updates of new reports. Any interested individuals can subscribe online to GenInfo at <http://www.humgen.umontreal.ca/en/GenInfo.cfm>.



**The GEEE! in
 Genome**

The Prime
 Minister of

Canada, the Right Honourable Jean Chrétien, on April 25, 2003, opened The Geee! in Genome, Canada's first national travelling exhibition on the science of genomics, at the Canadian Museum of Nature (CMN) in Ottawa. This unique and provocative bilingual exhibition will visit about nine cities across Canada over a three-year period.

The Geee! in Genome exhibition is a major feature of a national public education project on genomics – the study of genes and their functions. To find out more about the exhibition visit: <http://nature.ca/genome/>