CANADIAN GENETIC DISEASES NETWORK
RÉSEAU CANADIEN DE MALADIES GÉNÉTIQUES

Newsletter of the Canadian Genetic Diseases Network

OPEN READING FRAME

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Open Reading Frame: *noun.* (1) *in DNA, an open reading frame has no termination codon, or no signal to stop reading the nucleotide sequence and so may be translated into a protein and* (2) *the newsletter of the Canadian Genetic Diseases Network*

In This Issue

Open Reading Frame
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Canadian Genetic Diseases Network
201-2150 Western Parkway
Vancouver, BC V6T 1V6
Phone: (604) 221-7300
Fax: (604) 221-0778
Email: info@cgdn.ca
Website: www.cgdn.ca

New Discovery Sheds Light on Breast Cancer

Tumor size and disease progression are not always related

A team of Canadian researchers, in a study partly funded by the Canadian Genetic Diseases Network, has discovered that the behavior of certain breast cancers is not as easily predicted as once thought. Led by Network Investigators **Drs. Steven Narod** and **William Foulkes**, the team discovered that the presence of either the BRCA 1 or BRCA 2 gene mutation in a patient will significantly impact the pattern of metastasis.

> "This discovery is important because it points to the fact that breast cancer is many diseases," said Dr. Foulkes.

Traditionally, it was believed that the larger a tumor in a breast cancer patient, the more likely it was that the cancer cells had metastasized into the lymph system. The results of this study, which were published in the October 15th, 2003 issue of CANCER, demonstrate that the tumor size in a breast cancer patient with the BRCA1 gene mutation is not necessarily correlated with local disease progression.

"This discovery is important because it points to the fact that breast cancer is many diseases," said Dr. Foulkes. "Until each sub-type of breast cancer is understood, including hereditary and acquired forms of the disease, data from whole populations should not be used to predict disease behavior, such as metastatic potential, response to treatment and survival. Hereditary and acquired forms of breast cancer have biological differences that may require different diagnostics and treatment."

This study was funded by the CGDN, the Fonds de la Recherche en Santé du Québec Cancer Network – Breast and Ovarian Tumour Bank Axis, the Susan G. Komen Foundation, Norwegian Cancer Society and the Norwegian Research Council. ▲

Facts about Breast Cancer

- 1 in 9 women will develop breast cancer during her lifetime.
- Breast cancer is the most frequently diagnosed cancer among Canadian women and accounts for 30% of all cancer cases.
- Since 1986, mortality rates from breast cancer have declined by 20%.

Source: Canadian Breast Cancer Foundation

SNiP's – Short, Newsworthy Points

Three new CGDN Core Facilities

As a result of a recent competition, the CGDN will be funding three new Core Facilities effective October 1st, 2003. The new Facilities include the Microarray Analysis Core Facility, directed by **Drs. Bob Nadon** and **Tom Hudson**, the HPRT Targeted Transgenesis Core Facility led by **Dr. Alan Peterson** and the Mouse Cytogenomics Core Facility, directed by **Drs. Jayne Danska** and **Steve Scherer**.

We would like to congratulate the new Core Facility Directors and welcome them to the CGDN. The CGDN now supports 11 core facilities across Canada, which provide services and advice to the Canadian health research community. For more information about the Network's Core Facilities, visit www.cgdn.ca/eng/core/overview.html. ▲

Twenty-five students receive CGDN summer studentship awards

As a result of a national competition, twenty-five outstanding Canadian science undergraduate students spent the summer of 2003 in CGDN research labs. The students were selected on the basis of outstanding undergraduate marks and a strong interest in research. The experience is rewarding both for the undergraduates but also for the host researchers and their laboratory staff.

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Courtney Casserly spent the summer in the research lab of **Dr. Dennis Bulman**, a CGDN Scholar, at the Ottawa Health Research Institute. When asked about her experience this past summer, she said, "I would like to thank the CGDN for providing me with the opportunity to work in Dr. Bulman's laboratory. It has been an extremely rewarding experience, and it has allowed me to become much more knowledgeable in my current field of study. Furthermore, it has certainly opened my eyes to future opportunities in genetic research. Thank you again for making this possible!"

CGDN would like to thank the Principal Investigators, Scholars and Associates who hosted summer students in their labs in 2003. ▲

CGDN supporting colleagues in less affluent countries

Spearheaded by one of the Network's founding members, **Dr. Charles Scriver**, the CGDN is sponsoring subscriptions for the *Metabolic and Molecular Basis of Inherited Disease* (MMBID) online version for scientists in less affluent countries. So far, subscriptions have been provided to colleagues in South Africa, Mauritius, Argentina, Iran, Turkey, India, Czech Republic, Mexico, Costa Rica and Brazil. CGDN would like to acknowledge the generous support of McGraw-Hill, the publisher of the MMBID, for helping us to provide this valuable resource to our international colleagues. ▲

Two more Scriver MD/PhD studentships awarded

The Canadian Genetic Diseases Network, in partnership with the Institute of Genetics, CIHR, and the Canadian Gene Cure Foundation is pleased to announce that **Ms. Angela Hyde** and **Mr. Michael Ward** have been awarded the Drs. Walter, Jessie Boyd & Charles Scriver MD/PhD Studentship.

Ms. Hyde started her graduate studies in genetics in 2000 under the supervision of **Dr. Ban Younghusband**, working on a research project entitled "The molecular characterization of colorectal cancer in the Newfoundland population". She started her medical studies at the University of Newfoundland in the fall of 2002.

Mr. Ward started medical school at the University of Toronto in the fall of 2002 and will start graduate work with **Dr. Duncan Stewart** at the Institute of Medical Science in Toronto in January 2004 to work on a project entitled "Genetic and functional studies for primary pulmonary hypertension."

A total of eight Scriver studentships have been awarded to date and were named in honour of Dr. Charles Scriver and his parents, Drs. Walter and Jessie Boyd Scriver for their contributions to medicine and research in Canada. For more information, please visit the website of the Canadian Institutes of Health Research at www.cihr-irsc.gc.ca. ▲

Awards & Achievements

Dr. Michael Hayden is the 2003 recipient of the Henry Friesen Award

CGDN investigator, Johanna Rommens, has been named to the ISI Highly Cited List for her substantial contributions to science.

Michael Hayden receives Henry Friesen Award and is awarded UBC Killam Professorship

Dr. Michael Hayden, Chief Scientific Officer of the Canadian Genetic Diseases Network, has been awarded the Henry Friesen Award by the Canadian Society of Clinical Investigation and the Royal College of Physicians and Surgeons of Canada. The Henry Friesen Award recognizes Canadian scientists who are notable for his or her contribution to biomedical research and are recognized internationally as of the highest caliber.

Dr. Hayden has also received the designation of University Killam Professor from the University of British Columbia. This designation recognizes exceptional faculty members who have distinguished themselves as scholars in research and have received the highest acclaim by the academic community and the general public.

Dr. Hayden is the Director and Senior Scientist of the Centre for Molecular Medicine and Therapeutics in Vancouver and a Professor in the Department of Medical Genetics at the University of British Columbia.

Johanna Rommens recognized on the ISI list of highly cited researchers

CGDN Principal Investigator **Johanna Rommens** is now on the ISI Highly Cited researchers list. This list, compiled by the Institute for Scientific Information, identifies individuals who have made fundamental contributions to the advancement of science and technology in recent decades. These scientists are the most highly cited and comprise less than one-half of one percent of all publishing researchers. The ISI list also includes CGDN members Lap-Chee Tsui and Anthony Pawson. For more information visit www.isihighlycited.com.

Dr. Rommens is a Senior Scientist in the Department of Genetics at the Hospital for Sick Children's Research Institute and an Associate Professor in the Department of Molecular and Medical Genetics at the University of Toronto.

Royal Society of Canada's McLaughlin Medal awarded to Robert Korneluk

The Royal Society of Canada has awarded the prestigious McLaughlin Medal to **Dr. Robert Korneluk** in recognition of his contributions to human molecular genetics and his research on diseases including cancer, diabetes, neurodegeneration and eye disease.

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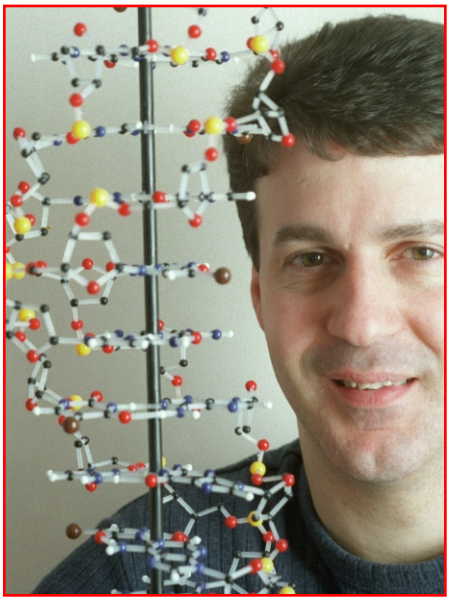
Dr. Korneluk is a Professor of Pediatrics and of Biochemistry, Microbiology and Immunology at the University of Ottawa. He is also the Director of the Solange Gauthier Karsh Laboratory and the Apoptosis Research Centre at the Children's Hospital of Eastern Ontario and a Principal Investigator of the Canadian Genetic Diseases Network.

The CGDN congratulates Dr. Hayden, Dr. Korneluk and Dr. Rommens for their impressive achievements. ▲

Scientists Discover Gene Related to Severe Form of Teenage Epilepsy



Dr. Scherer and his colleagues discovered a second gene responsible for teenage-onset epilepsy.

Scientists can now account for 90% of Lafora epilepsy because of a recent discovery made by a team of scientists led by **Dr. Steve Scherer**, a Principal Investigator of the CGDN and **Dr. Berge Minassian**.

> "The discovery of this gene has enormous implications for patients and families who are affected with this disease," said Dr. Michael Hayden, Scientific Director of the CGDN.

A 1998 discovery identified EPM2A, the first gene implicated in Lafora disease, which is present in 50% of cases. With the discovery of this new gene, NHLRC1, and studying its relationship to EPM2A, Dr. Scherer and his colleagues can now account for 90% of Lafora cases.

The team believes that the EPM2A and NHLRC1 genes work together to prevent neurons in the brain from accumulating too many carbohydrates. If either of the genes is missing, the result is epilepsy.

"The discovery of this gene has enormous implications for patients and families who are affected with this disease," said **Dr. Michael Hayden**, Scientific Director of the CGDN. "It has the potential to lead to new diagnostic and therapeutic treatments for Lafora Disease."

The study was funded by the Canadian Genetic Diseases Network, the Canadian Institutes for Health Research, Genome Canada, the Centre for Applied Genomics at the Hospital for Sick Children and the Hospital for Sick Children Foundation. ▲

What is Lafora Disease?

- Lafora disease is an autosomal recessive disorder that appears in the teenage years.
- Patients develop progressive myoclonus epilepsy (seizures), followed by intellectual decline and death within ten years.
- Approximately 1% of Canadians have a form of epilepsy.

Source: MMBID & Epilepsy Canada.

The Other Side

A Profile of Dr. Eric Shoubridge of the Montreal Neurological Institute.

Dr. Shoubridge, one of the newest CGDN Principal Investigators, researches mitochondrial DNA mutations and how they lead to a wide range of human diseases.

"The most important thing I have learned in science is to keep an open mind and keep doing experiments."

Dr. Shoubridge is one of the newest CGDN Principal Investigators, invited to join the Network in 2003. His research focuses on understanding mitochondrial DNA mutations and how they lead to a wide range of human diseases.

Q: Where are you from originally?

A: Toronto. (Actually, the outskirts of Toronto.)

Q: What made you go into science?

A: I don't think there was a specific event or person that catalyzed my interest in science. I have always been fascinated by biology, and it never occurred to me that I would be anything but a biologist. The thrill of scientific discovery just can't be beaten.

Q: Who has been your greatest role model?

A: My PhD supervisor, Peter W. Hochachka. He was quite simply irrepressible. He was also a bit of an iconoclast. To Peter, everything was possible. He was fascinated by biochemical puzzles, and he refused to be bounded by techniques. If a problem was worth investigating, you simply learned the techniques that would permit you to tackle it. To use his words, he "pulled the reptilian scales from my eyes."

Q: What has been one of your biggest challenges in your career?

A: Not having enough time to think, and finding the resources required to do long-term experiments that might take several years to pay off. These are the biggest challenges in research today.

Q: What is the most important life lesson that you've learned?

A: I won't tackle the life question, but the most important thing I have learned in science is to keep an open mind and keep doing experiments. Nearly every eureka moment I have had came from following up an unexpected experimental finding.

Q: If you could be anywhere in the world right now, where would you be?

A: Hmmm… It's Friday afternoon and I have a mountain of paperwork sitting in front of me, none of which is particularly interesting. Somewhere in the South Seas.

Q: Finish this statement: If I wasn't a scientist, I would be…

A: … a cellist. I would love to be able to play the Bach concertos for solo cello.

Q: Do you have any interesting idiosyncratic lab quirks?

A: If I did, I wouldn't say. You will have to ask the people in the lab. Actually, much to my chagrin, I hardly get to do lab work anymore; I live vicariously through my students. ▲

For your calendar

The 13th CGDN Annual Scientific Meeting

When: May 27th – 30th, 2004

Where: Talisman Mountain Resort
Kimberley, Ontario
Visit www.talisman.ca

Who: CGDN Investigators & Core Facility Directors, Network Associates Scholars, post-doctoral fellows, graduate students, Board Members, collaborators, industrial partners and invited guests

Why: For four days of scientific presentations, poster sessions, networking and collaborative opportunities

More info: E-mail Megan Airton at mairton@cgdn.ca or phone (604) 221-7300 ext 110.

BioLIGHT 2003

The first Biophotonics Primer and Workshop is taking place from November 13th – 16th, 2003 at the Four Points Sheraton in Gatineau, Quebec. Organized by Vitesse Re-Skilling Canada Inc. and the Canadian Institute for Photonics Innovation, the workshop will focus on the subjects of Medicine and Health, Biosciences, Environment, Security and Defence. More details are available at www.vitesse.ca.

Life Sciences Symposium with Dr. Francis Collins

A free public lecture featuring Dr. Francis Collins of the National Human Genome Research Facility entitled *"DNA at 50: The Best is Yet to Come"* is taking place on Wednesday, October 29th from 10am – 12:30pm at the Chan Centre at UBC. The lecture will be followed by a moderated panel discussion featuring BC's top genomics and genetic scientists. Tickets are not required, but please RSVP to info@genomebc.ca or call (604) 637-4390. ▲

bioinformatics.ca

Canadian Bioinformatics Workshops

2004 marks the 6th season series of the Canadian Bioinformatics Workshops. The series, coordinated by CGDN as part of its training mandate, offers courses in computational biology at locations across Canada.

The 2004 course schedule is listed bellow:

BIOINFORMATICS
February 16 – 28, 2004 in Vancouver

DEVELOPING THE TOOLS
May 3rd – 9th, 2004 in Montreal

PROTEOMICS
July 19th – 24th, 2004 in Calgary

GENOMICS
August 16th – 21st, 2004 in Vancouver

For more information and registration deadlines, visit www.bioinformatics.ca.