CANADIAN GENETIC DISEASES NETWORK
RÉSEAU CANADIEN DE MALADIES GÉNÉTIQUES

Newsletter of the Canadian Genetic Diseases Network

OPEN READING FRAME

Volume 1; Issue 6 GENES TO THERAPIES December 2003

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

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CANADIAN GENETIC DISEASES NETWORK
RÉSEAU CANADIEN DE MALADIES GÉNÉTIQUES

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Gene Researching for a Week

30 high school students will give up their spring break holiday to study genetics

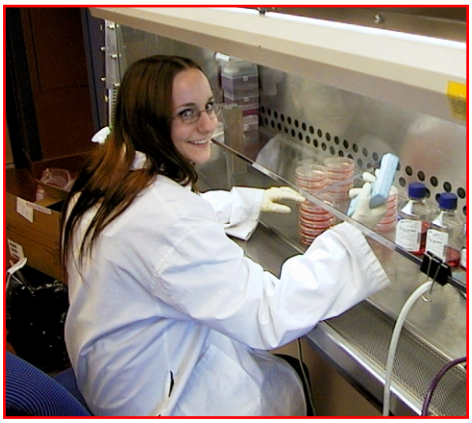
PCR. DNA isolation and purification. PCR. SDS-PAGE. Bioinformatics. Gel electrophoresis. DNA sequencing. Western blotting. Chances are if you're not a genetic researcher, you don't know what these terms mean.

Thirty Canadian high school students are about to find out.

Throughout March and April 2004, thirty high school students from across the country will be joining the genetic research labs of twenty-four CGDN investigators for a one-week crash course in genetics. This marks the second year that the Canadian Genetic Diseases Network, in partnership with the Institute of Genetics, CIHR, has organized *Gene Researcher for a Week – The Ramon Hnatyshyn Youth in Science Initiative.*

Gene Researcher for a Week is a unique opportunity for Canadian high school students to gain first-hand knowledge and experience in genetics and genetic research.

"Gene Researcher for a Week is a unique opportunity for Canadian high school students to gain first-hand knowledge and experience in genetics and genetic research," said **Dr. Tom Hudson**, CGDN scientist and program founder. "CGDN scientists are committed to mentoring the next generation of young researchers and encouraging them to pursue a career in science and genetics."



High school student Crystal Gennick works in a tissue culture hood at the Manitoba Cancer Research & Treatment Centre during her 2003 spring break under CGDN scientist, Dr. Geoff Hicks.

Gene Researcher for a Week was developed out of a strong belief across the genetics community that more must be done to educate Canadians about genetic research. "I spent several years asking my colleagues about how to reach out to people across the country. After many hours of discussion with other CGDN scientists, *Gene*

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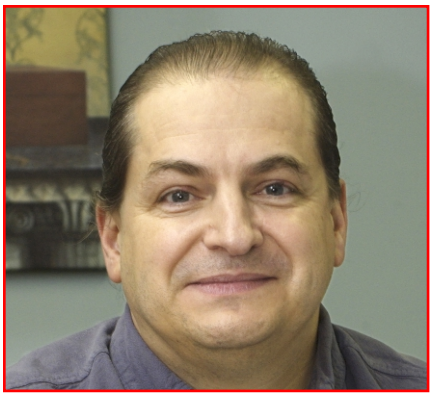
Awards & Achievements



CGDN member Dr. Régen Drouin receives Tier 1 Canada Research Chair

Régen Drouin receives a Tier 1 Canada Research Chair

CGDN network member, **Dr. Régen Drouin**, has been named the Canada Research Chair in Genetics, Mutagenesis & Cancer. In his research, Dr. Drouin uses advanced genome sequencing technology to study DNA damage in skin cells by the sun, enhancing our knowledge of the molecular processes involved in the development of skin cancer. An understanding of these mechanisms could lead to the development of new preventative therapies.

Canada Research Chairs are awarded to those individuals who advance the frontiers of knowledge in their fields, not only through their own work but also through teaching and mentoring students.

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Canada Research Chairs are awarded to those individuals who advance the frontiers of knowledge in their fields, not only through their own work but also through teaching and mentoring students. Dr. Drouin is the director of the CGDN In vivo DNA Analysis Core Facility and an associate professor in the Faculty of Medicine at the Université de Sherbrooke in Quebec.

Another CGDN member added to the ISI Highly Cited list

We are proud to announce that CGDN investigator, **Dr. David MacLennan**, has been added to the ISI List of Highly Cited Researchers. The list, compiled by the Institute for Scientific Information, recognizes 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.

CGDN Investigator David MacLennan has been named to the ISI Highly Cited List for his substantial contributions to science.

Dr. MacLennan, an Officer of the Order of Canada, is with the Banting and Best Department of Medical Research at the University of Toronto. His research areas include calcium binding and transport across membranes, Brody Disease, cardiomyopathy, malignant hypothermia and central core disease.

For more information, visit www.isihighlycited.com

Student in the lab of Erwin Schurr wins first prize at ASHG

Marcelo Mira, a student in the lab of CGDN Principal Investigator **Dr. Erwin Schurr**, won first prize in the clinical pre-doctoral student award competition at the 53rd Annual Meeting of the American Society of Human Genetics in Los Angeles, CA for her work on the project “Strong association of intragenic SNPs of the 6q25 region with leprosy in two independent samples”.

The CGDN congratulates Dr. Drouin, Dr. MacLennan and Marcelo Mira for their impressive achievements! ▲

Unraveling the mystery of a global epidemic

A CGDN discovery may lead to protection from Malaria

Malaria affects 300 to 500 million people worldwide and is responsible for 2 to 3 million deaths per year. Although it is prevalent in most tropical regions around the globe, the majority of malaria-related deaths occur in Africa.

> "This gene is critical in preventing parasitic replication. Very seldom are genes identified that have this kind of drastic implication for so many millions of people around the world."

CGDN investigator **Dr. Philippe Gros** and his team at McGill University have identified a mutation in the *Pklr* gene that causes a deficiency in pyruvate kinase, an enzyme that converts glucose to energy in red blood cells. This deficiency appears to protect mice against malaria by preventing the malarial parasite from growing in red blood cells.

The findings, which were published in the December issue of *Nature Genetics*, suggest that a similar situation in humans could lead to protection from malaria.

"This study shows that a deficiency in pyruvate kinase protects mice from developing malaria; however, we also know that the same deficiency in humans causes hemolytic anemia," said Dr. Gros. "The next step is to determine if the protection against malaria in mice is indirectly or directly related to anemia."

Dr. Gros and his team will now study whether hemolytic anemia confers resistance to malaria by reducing the number of mature red blood cells in the body, making it more difficult for the malaria parasites to grow.

"Although there are many co-factors that worsen the malarial epidemic, any crack in the armor that has the potential to prevent the malarial parasite from replicating in the host has great value," states Dr. Gros. "This gene is critical in preventing parasitic replication. Very seldom are genes identified that have this kind of drastic implication for so many millions of people around the world."

This study was funded by the Canadian Genetic Diseases Network, CIHR and McGill University. ▲

Facts about Malaria

- Malaria is a life-threatening parasitic disease transmitted by mosquitoes.
- Approximately 40% of the world's population, mostly those living in the poorest countries, is at risk of malaria.
- Malaria is one of the major public health challenges that undermine development in the world's poorest countries.

Source: The World Health Organization's Roll Back Malaria Initiative

Gene Researching

Continued from page 1

Researcher for a Week was born," explained Dr. Hudson.

Because the CGDN and its partners provide travel and accommodation bursaries, the program is accessible to students in both urban and rural areas. With the assistance of Indian and Northern Affairs Canada, the CGDN is hoping to involve aboriginal students in this year's program.

Gene Researcher for a Week attracts a diverse group of students, explained **Dr. Chrystal Palaty**, CGDN's Manager of Scientific Affairs. "Each applicant is interested in being a gene researcher for a week for a different reason. Some of the students have a close family member with a genetic disease and others are just fascinated by the great potential of genetic research and are eager to try techniques that they don't have the opportunity to use in high school classes."

But providing Canadian teenagers with a unique learning opportunity is not the only reason why CGDN scientists host a student. "Don't think that the students are the only ones benefiting from this experience!" explained Dr. Hudson. "My lab is very motivated by having an enthusiastic student with us for a week, someone who is excited to spend her spring break working alongside us." ▲

The Other Side

A Profile of Dr. Elise Héon of the Hospital for Sick Children in Toronto

Dr. Elise Héon, a CGDN investigator and Ophthalmologist-in-Chief at the Hospital for Sick Children, researches inherited eye disorders.

Dr. Héon, the first woman to head a surgical unit at the University of Toronto, is an associate professor of Ophthalmology at the University. She is Ophthalmologist-in-Chief at the Hospital for Sick Children and an associate scientist in Genetics and Genomic Biology at the Hospital for Sick Children Research Institute. Her clinical practice is dedicated to inherited eye disorders including retinitis pigmentosa and retinoblastoma.

Q. Where are you from originally?

A. I was born and raised in Montreal, Quebec. I came to Toronto in 1991 for 2 years for my fellowship in pediatric ophthalmology and my molecular genetics research training. Then, I went to Iowa for 18 months to do more molecular ophthalmology and then to Switzerland for a year.

Q. What made you go into science?

A. I was always attracted to helping people. But when I got into medicine, I realized that I had such a curiosity for researching problems in great depth. It became clearer when I was in ophthalmology that I wanted to make a difference through research and not only through private practice.

Q. Who have been your greatest role models?

A. Brenda Gallie and Edwin Stone, two stellar world-class clinician-scientists who paved the way for me. It's really hard as an ophthalmologist to be a clinician-scientist and they did a lot of the hard work to convince people of the importance of the clinician-scientist in vision science.

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

A. Being a clinician-scientist, meaning balancing both research and patients. It's a challenge to be good at both and you need to be in order to succeed and be respected. Now being Ophthalmologist-in-Chief and leading a department with 50 people while trying to maintain my research life is not easy. The two combined are more of a challenge than I could have ever imagined! But if and when you want to make a difference, I think these huge challenges make it happen.

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

A. I have learned many more than one! The most important is: Never quit! Second: always fight for the good things and believe in what you do. Also, pick you battles. Ed Stone taught me: Never bleed when you swim with sharks! I could go on…

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

A. On a beach with my feet in the water, then off to the mountains for some skiing, but in reality: Right here. I love what I do…I just do too much of it!

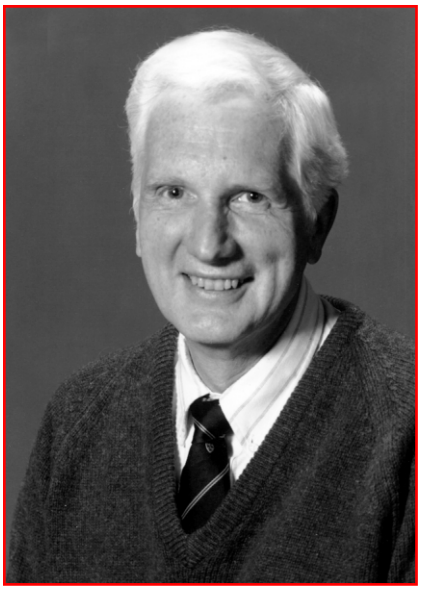
Q. Finish this sentence: "If I wasn't a scientist, I would be…

A …A gardener, or a dog breeder and I am sure that I would have a scientific approach to either!

Q. Do you have any idiosyncratic lab quirks?

A. I have too many – I would rather not go there! ▲

SNiP's – Short, Newsworthy Points



CGDN investigator Charles Scriver for whom the MD/PhD program is named.

Journal of Neuroscience Highlights New CGDN Core Facility technology

The technology of CGDN's new HPRT targeted transgenic facility was highlighted in the November 12th, 2003 volume of the *Journal of Neuroscience* in an article entitled *"A Combinatorial Network of Evolutionary Conserved Myelin Basic Protein Regulatory Sequences Confers Distinct Glial-Specific Phenotypes"*. **Dr. Alan Peterson**, the HPRT Core Facility Director, is the principal author on this study, which also includes CGDN member, **Dr. Tom Hudson** as a collaborating author.

This publication describes the identification and functional characterization of four conserved regulatory sequences, which control the transcription of myelin basic protein (MBP). MBP is an essential component of the white matter which coats nerves in the central and peripheral nervous system, enabling impulses to be conducted between the brain and other parts of the body. Understanding the complex signals control how and when MBP is produced may provide therapeutic insights for activating this protein to promote healing for damaged situations.

More information about this Core Facility is available on the CGDN website www.cgdn.ca/eng/core/hprt_targeted_transgenesis.html

Pfizer Hosts Drs. Walter, Jessie Boyd & Charles Scriver MD/PhD Studentship Recipients in Montreal

On November 28th, 2003, Pfizer Canada Inc. hosted recipients of the Drs. Walter, Jessie Boyd & Charles Scriver MD/PhD Studentship in Montreal for a research day with representatives from Pfizer. It was an opportunity for the students to present their research, network with Pfizer scientists, and in many cases, it was their first opportunity to meet each other.

During their visit to Montreal, **Mr. H. Arnold Steinberg** hosted a dinner for the students and **Dr. Charles Scriver**, after whom the studentship is named. Supporters of the Drs. Walter, Jessie Boyd & Charles Scriver MD/PhD Studentship were also in attendance including the Canadian Genetic Diseases Network and Pfizer Canada. **Dr. Ron Woznow**, CEO of the CGDN, remarked "This evening provided the scholarship recipients with an opportunity to speak with Dr. Charles Scriver in an informal setting. The evening also gave us the opportunity to meet with the recipients and be inspired by their dedication."

The Drs. Walter, Jessie Boyd & Charles Scriver MD/PhD Studentship program is an initiative of the Canadian Gene Cure Foundation, the CGDN and the Institute of Genetics, CIHR and was established to address the shortage of genetic clinician-scientists in Canada. It was named for the Scriver family in honour of their dedication to translating discoveries made in the lab to the bedside, improving the health of patients. ▲

For your calendar

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
Application deadline closed.

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.

CGDN's 13th Annual Scientific Meeting

The 2004 Annual Scientific Meeting of the CGDN will be held from May 27th – 30th, 2004 at the Talisman Mountain Resort, one and a half hours from downtown Toronto, Ontario. CGDN investigators, core facility directors, associates, post-doctoral fellows, graduate students, Board Members and industrial partners will come together for four days of scientific presentations, poster sessions, networking and collaborative opportunities. For more information, please call Megan Airton at (604) 221-7300 ext. 110 or e-mail mairton@cgdn.ca.

Enhancing health. Enriching Lives.

The Canadian Genetic Diseases Network is the primary catalyst in advancing collaborative medical genetics research in Canada. The unique efforts of our researchers have resulted in significant contributions to the ongoing prevention, diagnosis and treatment of human disease.

The Canadian Genetic Diseases Network is made possible through funding from the Networks of Centres of Excellence Canada. The Networks of Centres of Excellence Canada is a joint initiative of the Natural Sciences and Engineering Research Council, the Canadian Institutes of Health Research, the Social Sciences and Humanities Research Council and Industry Canada. ▲