

GENEXPressions

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3RD ANNUAL CANADIAN
HUMAN & STATISTICAL GENETICS MEETING

Making a Difference with GENE RESEARCH

A Canadian Gene Cure Foundation funded research project led to a medical breakthrough with dramatic results for a young woman from Quebec. Now 25 years old and working at a full-time job, Karine suffered for most of her childhood and adolescence from several medical conditions and needed daily insulin injections for diabetes. The discovery changed everything for the better.

Within a few days of Karine's birth in 1988, her mother Hélène knew something was wrong. Karine wasn't eating or drinking, so Hélène decided to return to the hospital. Within a week, Karine was transferred to the Neonatal Intensive Care Unit at the Montreal Children's Hospital. Doctors looked for many different conditions, eventually diagnosing baby Karine with diabetes.

In the years that followed, doctors discovered other medical problems in Karine. At age two, she was put on a drug for epileptic seizures, and at age six, she was diagnosed with autism spectrum disorder.



Karine (left) and Rosemarie Grabs (right)

A DISCOVERY THAT CHANGES EVERYTHING

At age 16, Karine started to see Dr. Laurent Legault for her diabetes. **Dr. Constantin Polychronakos**, an endocrinologist, was also involved in her care.

A strong proponent of individualized medicine, Dr. Polychronakos's research focused on the one percent of diabetic children in Canada, numbering about 1,000, who are able to produce insulin, making them candidates for a pill treatment instead of daily injections of insulin.

"It's the next best thing to curing their diabetes," Dr. Polychronakos says.

His initial research confirmed that these children, whose disease is caused by a rare genetic mutation, can be treated with oral medication rather than painful injections. Oral medication allows children to produce their own insulin, which assures near-perfect control of blood sugar, and leads to better health overall. With insulin injections, blood sugar control is impossible.

The Canadian Gene Cure Foundation awarded Dr. Polychronakos \$65, 000 in 2005 for his research

into Type 1 juvenile diabetes. With the grant, Dr. Polychronakos's lab set out to review the DNA samples that it had collected over the years from children diagnosed with Type 1 diabetes. The research team's goal was to find a genetic mutation that would permit the switch in some patients.

After about a year of research, Dr. Polychronakos called Karine's mother, Hélène, with news—a member of his lab team, Rosemarie Grabs, had identified a gene mutation in Karine. The mutation made her a perfect candidate to try an oral anti-diabetic treatment (glyburide pills) instead of daily insulin injections.

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In consultation with Hélène, the medical team planned a two-week transition, during which Karine would be weaned off insulin injections and start taking glyburide pills three times a day.

Drs. Legault and Polychronakos called Hélène every day to check her daughter's blood sugar tests until she completed the transition. The dose was then stabilized at two pills, two times a day.

BEYOND EXPECTATIONS

To be free of the responsibility that comes with daily injections was a tremendous relief for Hélène. But what else happened was far greater than anything she ever

It's the next best thing to curing juvenile diabetes.

- DR. POLYCHRONAKOS

expected. Up until the age of 16, Karine was not a very social or expressive child.

"You could say that the new treatment was a sort of liberation for Karine. Her behaviour and demeanor completely changed for the better," Hélène says.

Karine now works five days a week at L'école Aérotechnique de St-Hubert on

a project sponsored by the Centre de réadaptation en déficience intellectuelle (CRDI). She has also developed her hobbies and interests: Hélène says she's a whiz at jigsaw puzzles, and knows virtually every song on the radio—artist, title and lyrics.

"My life was pretty difficult before Karine started her new treatment," Hélène says. "I was the one who had to inject her with insulin every day for 17 years. It was very stressful at times. But once she started her new treatment, I had more time to spend with my other daughter, Stéphanie, and with my partner. Honestly, it changed all our lives for the better."

GRFAW student profile

Growing the Next Generation of Researchers: Meet Michelle Ning



Michelle Ning, GRFAW Alumni

They say seeing is believing, and so it was for Michelle Ning when she first laid eyes on DNA while working in a research lab at Simon Fraser University.

The year was 2008 and Ning was a grade 11 student in Port Coquitlam, British Columbia, taking part in the Gene Researcher for a Week program. Designed to expose students to potential careers in genetic research, the program offers the opportunity to conduct hands-on experiments in the labs of influential scientists.

Ning was placed in the lab of SFU molecular biologist Dr. Jack Chen,

who works with the nematode *C. elegans* to identify novel genes, structural variations, and regulatory mechanisms through bioinformatics and data mining.

With the help of a PhD student working in Dr. Chen's lab, Ning learned basic scientific protocols and how to identify mutant and normal wild strains of the transparent nematode.

But the real magic happened when she was given the opportunity to amplify DNA with the help of a PCR machine. PCR machines use the polymerase chain reaction to create multiple copies of DNA, giving researchers a critical mass to analyze.

After using a pipette to drop the amplified DNA into specially prepared gels, Ning watched in amazement as they were exposed to UV light.

"I had no idea you could actually see bands of DNA. It was just really cool. It was magic," recalls Ning. "It made it real for me. It made it into something you could study for its influence on so many things in life and the experience solidified my interest in this area."

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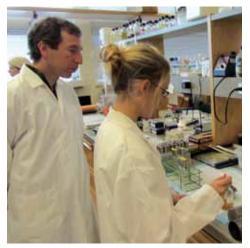
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Dr. Stephen Rader at UNBC

So when it came time to go to university, she knew just what she wanted to study: cell biology and genetics.

During her time at the University of British Columbia, Ning worked in the lab of an evolutionary geneticist studying stickleback fish and also prepared literature reviews for a medical geneticist at B.C. Children's Hospital.

Four years later, with a BSc from the University of British Columbia under her belt, Ning landed a job as a community manager for health care professionals.

She works for Vancouver-based Sosido, an online, worldwide community of healthcare professionals sharing peer-reviewed published clinical research and practical clinical knowledge. It's Ning's job to assess scientific findings and get that information to health professionals who could benefit from that knowledge.

While Ning says she knew after university that work in a genetics lab wasn't for her, she is grateful for the way it shaped her thought processes.

"I had no idea you could actually see bands of DNA.

It was just really cool.

It was magic!"

— MICHELLE NING, GENE RESEARCH FOR A WEEK ALUMNUS

"I would never give up any of my research experience," she says. "It teaches skills like attention to detail and how to think rationally."

That's why Ning encourages high school students to take advantage of the GRFAW program, even it they aren't sure they want to be researchers.

"There is no opportunity to do this anywhere else—unless your parents happen to own a genetics lab," she jokes.

HOW TO APPLY

We are looking for high school students, age 16 and over, who are passionate about science and intrigued by genetics. We encourage students with an overall average of at least 85%, who can write a brilliant essay to demonstrate their excitement about science, who have advanced communication skills, and who can convince a teacher to write a stunning reference to apply. Gene Researcher for a Week (GRFAW) program is an opportunity to gain insight into the world of genetic diseases through a mentorship in one of Canada's top genetic labs. This program is designed to provide a learning experience for students that have not had extensive lab experience from other programs or work experience opportunities.

The GRFAW mentoring program is a great opportunity to connect with the scientific community. Students can apply online before December 31st at:

www.genecure.ca/en/programs/gene-researcherfor-a-week/student-application/

WANTED: Host Research Lab Teams

We're looking for research teams who want to share their passion and knowledge with Canada's next generation of researchers. Since 2003, the Canadian Gene Cure Foundation has been selecting motivated and ambitious students in Grades 11 and 12 who have an innate curiosity for science, human genetics, and human genetics research to participate in our **Gene Researcher for a Week program.** Initiated by the Canadian Genetics Disease Network in 2003 as a way to introduce the field of human genetics research as a career path to Canada's brightest young people, we have an increasing demand for placements and need your help.

Please consider hosting one or two students in your lab this coming spring. Labs must provide as much hands on learning as possible, however, we find that students attend meetings, lectures and discover great learning is also accomplished in observation. The best mentoring includes a healthy mix of experiences provided by a lab team. To find out more, contact Tracy at 250.708.4203 or tzeisberger@genecure.ca

MARK YOUR CALENDAR

3RD ANNUAL CANADIAN HUMAN & STATISTICAL GENETICS MEETING

- VICTORIA, BC!



The Canadian Institutes of Health Research-Institute of Genetics (CIHR-IG), in collaboration with the Canadian Gene Cure Foundation (CGCF), is pleased to sponsor the Third Annual Canadian Human and Statistical Genetics Meeting. This joint event will be held at the Fairmont Empress, Victoria, BC, May 3-6, 2014.

Visit http://epigen3.mcgill-cihr-ig.ca/ for updates on speakers and registration.

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The Canadian Gene Cure Foundation is a charitable organization and is grateful for gifts of any amount in support of the Foundation's goals and programs, which include research grants, the Champions of Genetics Program, and the Gene Researcher for a Week Program.

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