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Canadian Gene Cure Foundation
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GENExpressions

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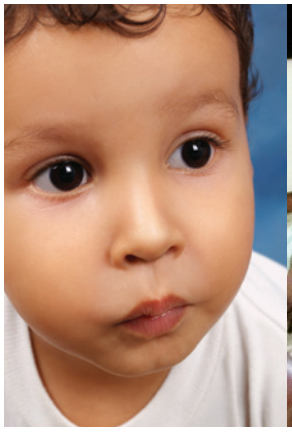
2012 CHAMPIONS OF GENETICS

The Next Generation Grant funds needed research staff for study in childhood eye cancer



Each year, the Canadian Gene Cure Foundation (CGCF) honours researchers who make a difference in the genetics research community — they are named our Champions of Genetics. Champions are senior scientists actively conducting genetic research and mentoring young researchers in the field. Champions nominate early career scientists for Building the Next Generation Grants, funded by the Canadian Institute of Health Research-Institute of Genetics (CIHR-IG) and the CGCF. These three $90,000 grants help bright, young scientists establish their laboratories and reach their research goals.

Helen Dimaras is one of these young scientists, nominated in 2011 by Brenda Gallie, medical geneticist and pioneer in researching cancer in children.

An assistant professor at the University of Toronto in the Department of Ophthalmology, Dr. Dimaras focuses on retinoblastoma (childhood eye cancer), which is a genetic cancer. With CGCF's help, the scientist is continuing research on early detection techniques.

"Our preliminary evidence shows that we can actually detect the cancer earlier than we do now," Dr. Dimaras says. "That would be an advantage if you were to initiate treatment at a time when you can't actually see the

CONTINUED »

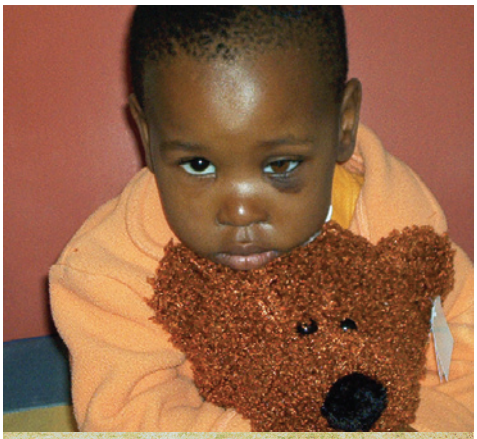
cancer, when the genetic evidence is telling you it's there so you can potentially treat it earlier and have better outcomes."

If detected and treated early enough, retinoblastoma survivor rates are high.

In most Canadian communities early detection of retinoblastoma is common, but that's not true for countries with struggling healthcare systems. And that's one reason Dr. Dimaras is so committed to her research. Early in her career, she was on a Canadian medical team working in Kenya to help that country build a national strategy to improve cancer survival rates of retinoblastoma.

"For me it was really important that whatever research I do wouldn't just be applicable to the eight percent of children that live in the developed world countries. Ninety-two percent of kids with retinoblastoma live in poor countries with few resources," Dr. Dimaras says. "Here in Canada if a parent notices a white reflex (in the eye), they can very easily consult with their doctor. It's not a 10-mile walk to the nearest clinic. But in many developing countries, mothers might not seek help for a white flash because it's not causing the child any distress. Then the tumor just proceeds to grow."

The most common symptom of an affected eye is a pupil that looks white, or has white spots, most noticeable in photographs taken with a flash. A crossed eye is the second most common symptom of retinoblastoma. The cancer most often affects children under six years old, usually between the ages of one and two. Untreated, the disease can spread from the eye via the optic nerve to the brain, the spinal cord, and bones.



90 percent of children
with retinoblastoma
are the first in their families to develop eye cancer.
In 10 percent of cases, a family member has also had the disease.

Retinoblastoma is an old disease, having been documented in children over **2,000** YEARS AGO.

Dr. Dimaras's pilot study at the Hospital for Sick Children has already shown that by identifying biomarkers of a patient's tumour and then searching for those biomarkers in bone marrow and cerebrospinal fluid, they could detect the disease outside the eye, well before a standard biopsy would turn up cancer cells.

With CGCF funding, Dr. Dimaras hired a post-doctoral fellow to assist with research, Dr. Ledia Brunga. Dr. Brunga is also expanding the research to study patients who experienced some form of hearing loss after chemotherapy treatment for retinoblastoma. The goal is to see if these patients have the genetic variants that predispose them to hearing loss after chemotherapy. By identifying high-risk patients, treatment might be fine-tuned to avoid that side effect.

The work is a testament to the value of funding an up-and-coming scientist.

"The funding is pivotal to having my own lab," Dr. Dimaras says. "It was really beneficial to me, allowing me to hire a postdoc — they require less of a researcher's time because they're already well-trained."

Dr. Brunga, a trained doctor from Albania, manages all the clinical and molecular data of the lab. A couple of undergrads are also part of the lab, which furthers their own training, potentially in the clinical genetics field.

As for Dr. Dimaras's future, her focus is doing good science "and making a difference in the lives of the patients."

Young Researchers **IN THE LAB**

Gene Research for a Week (GRFAW) is a program that introduces Canadian high school students to high profile genetics labs across the country.

BY FARAH MUSHARBASH,
GRFAW ALUMNI 2012

When I received my acceptance email from the GRFAW program, I was struck at first by my placement location — McGill. Having applied to McGill and receiving my acceptance offer three weeks after my GRFAW acceptance, I realized that it was an excellent opportunity to do research and explore a prestigious university. I was placed in the lab of Dr. Nada Jabado, an associate professor of pediatrics, a Champion of Genetics, and a fantastic lab host as well. Dr. Jabado's lab provided me with an enriching research experience, and an opportunity to discover what Canada's best university has to offer. The GRFAW program definitely had a positive impact on my future, inspiring me to continue my education at McGill.

Farah is now at McGill studying biomedical sciences and is starting a job at a nanobioengineering lab.

FEATURED Host Lab

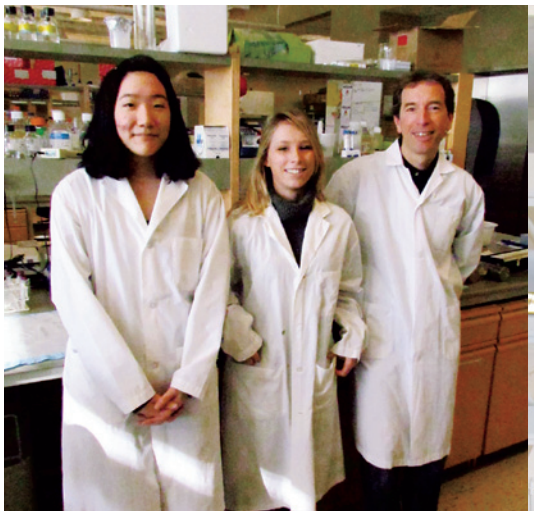
Over 70 human genetics labs have hosted GRFAW students from across the country, providing over 9,000 hours of training since 2003, the year the program began.

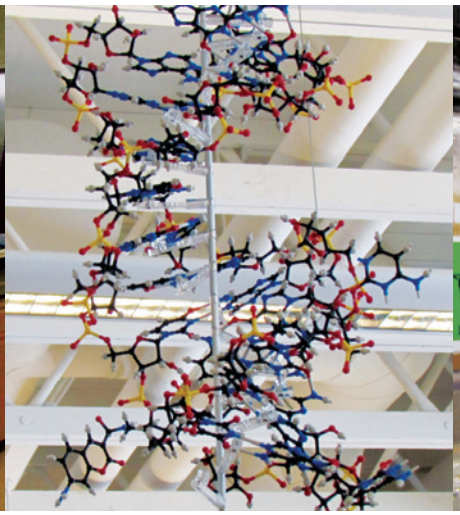
The Blackburn Cardiovascular Genetics Laboratory at the Robarts Research Institute, The University of Western Ontario, is a molecular genetics lab focusing on cholesterol and lipid disorders that lead to heart disease.

"[GRFAW] students have a good introduction to DNA, lipids and atherosclerosis. Specifically, we teach the students, what DNA does and where it lives, how it's extracted and what you can do with it in the sense of looking for mutations and working with it via techniques we use in the lab," says Matthew Ban, a researcher in Dr. Robert Hegele's Blackburn Cardiovascular Genetics Laboratory.

Researchers extract DNA from blood samples, studying the molecule with high-tech gear, such as a thermal cycler, an instrument that amplifies DNA fragments. The lab also sequences DNA to look for mutations causing disease-the scale of which has increased greatly over the last few years.

To personalize the experience, the lab often has students swab their own cheeks, extract DNA, amplify it, and search their genetic code for changes or polymorphisms. The students also have the chance to visualize DNA through gel electrophoresis, an instrument that separates DNA molecules of different sizes and allows researchers to see the molecules with ultra-violet light — a technology unavailable to high school students.





THE SCRIVER
MD/PHD PROGRAM

Established in 2002, the Scriver MD/PhD scholarship honours the work of Charles Scriver, a pediatrician and geneticist who created the first biochemical genetics lab and clinical program in Canada, at McGill University. Dr. Scriver developed a new screening test for phenylketonuria (PKU), a genetic disease that causes brain damage, seizures and mental disabilities if left untreated. The test is now used in countries around the world.

The scholarship, in partnership with the Canadian Gene Cure Foundation and the Institute of Genetics (CIHR-IG), supports the next generation of clinician-scientists.

JASON MAYNES, 2002 SCRIVER MD/PHD AWARD RECIPIENT

Anesthesiologist at the Hospital for Sick Children, Investigator in Molecular Structure and Function, SickKids Research Institute

Dr. Maynes graduated from the University of Alberta with a MD/PhD in 2006. As a PhD student, Dr. Maynes studied biophysics in the lab of Michael James, Canada's first protein crystallographer. The James lab has contributed vital research to understanding RNA viruses, such as hepatitis A, polio, hepatitis C, and rabbit hemorrhagic disease.

For Dr. Maynes, the CGCF funding lightened the economic burden of continuing research, especially as a PhD student.

"If you have family, kids or responsibilities like that, it can be economically challenging to continue," says Dr. Maynes, a father of two young children at the time.

Today, as a researcher, Dr. Maynes studies how anaesthetics affect the cell, particularly important research for understanding the effect of anaesthesia and the developing brain. The work is a perfect blend of science and clinical medicine.

"Anaesthesia itself can be very physics- and pharmacology-based," Dr. Maynes says. "So it reflects my research background and clinically, one of the biggest issues right now in anaesthesia is anaesthetic neurotoxicity. Those two things together meshed rather well for me: I can apply science and biophysics to problems I see as a clinician. And the only way to do that effectively is by going through an MD/PhD program."

Since 2002, the Canadian Gene Cure Foundation
has awarded 16 scholarships
— a total of $658,000 —
through the MD/PhD program.

The Canadian Gene Cure Foundation is a national charity and is grateful for gifts in any amount.

To donate online go to **www.genecure.ca/en/donate** Tel: **250.708.4203** Email: **info@genecure.ca**

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