

Major genetic breakthrough in rare form of ovarian cancer

The discovery of mutations in a single gene in multiple cases of small cell carcinoma of the ovary, hypercalcaemic type (SCCOHT), the most common undifferentiated ovarian cancer to strike women under 40, by a team led by **Dr. William Foulkes**, represents a huge breakthrough. The findings are published in *Nature Genetics*.

“Though rare, SCCOHT is important because the average age of victims is 25, with some as young as 2 years old. The oldest known patient was a woman of 48. It kills about half of those who get it within five years,” explains Dr. Foulkes, who is head of the Cancer Genetics Laboratory at the LDI and of the Program in Cancer Genetics at McGill.

Using whole exome sequencing (WES), a revolutionary method for diagnosis and taxonomy that was only developed over the past five years, Dr. Foulkes and collaborator

Dr. Jacek Majewski of the Department of Human Genetics at McGill, succeeded in identifying a mutation in the chromatin-remodelling gene SMARCA4 that was common to three families with at least two members afflicted with SCCOHT. Further testing of a total of 40 cases revealed that this was the only important genetic alteration in any of the tumors.

“Treatment options remain limited to poisoning this cancer with DNA damaging agents,” said Dr. Foulkes. “However, these are unlikely to be as effective as using novel therapies that function by modulating expression of the mutant gene. Developing a drug that targets SMARCA4 could have widespread benefits, as this gene has been implicated in various primary cancers, including kidney and pediatric brain tumors.”

Patients with a family history of early on-set ovarian cancer can now be screened for this genetic mutation and given counselling about available options.

Importantly, this study classifies SCCOHT as a malignant rhabdoid tumor – in essence, a type of tumor usually seen in the brain that, in this instance, sprouts in the ovary. This is much more than a mere academic achievement, as pathologists depend on recognizing the architecture of a cancer in order to accurately diagnose it. Now they will be able to diagnose this cancer with a simple antibody test.

Developing novel therapies targeted at particular biomarkers is among the strengths of the cancer research programs at the LDI and Segal Cancer Centre. The principle underlying personalized medicine in cancer, which is a major focus at the JGH, is to identify those biomarkers that characterize each patient’s cancer and to prescribe therapies that directly target the genetic foundations of the malignancy. Treatment can be directly targeted when the cause of a cancer can be isolated to a particular gene, leading to far better outcomes for patients.

Critical to the research that went into this study were extensive international collaborations involving clinician-scientists across Canada and from as far afield as Australia, Germany, France, Greece, Slovenia, the United States, and the United Kingdom. Essential support for several aspects of this project was supplied by the McGill University and Genome Quebec Innovation Centre.

[“Germline and somatic SMARCA4 mutations characterize small-cell carcinoma of the ovary, hypercalcaemic type”](#) by Leora Witkowski et al, is published in *Nature Genetics* online ahead of print publication.

“One of the most satisfying parts of the project was working directly with women and their families from all around the world,”

- Dr. William Foulkes

This “open[s] the door for development of specific therapies targeted to SCCOHT, of which there currently are none.”

- Brooke LaFlamme,
Assistant Editor, *Nature Genetics*

Dr. William Foulkes awarded 2013 O. Harold Warwick Prize

Dr. William Foulkes, head of the Cancer Genetics Laboratory at the LDI and Director of the Program in Cancer Genetics in the Departments of Oncology and Human Genetics at McGill, is the 2013 winner of the O. Harold Warwick Prize, awarded by the Canadian Cancer Society to a scientist whose research has had a major impact on cancer control in Canada.

Dr. Foulkes is a James McGill Professor of Medicine, Human Genetics and Oncology at McGill, and previously held an FRSQ *Chercheur national* award. His lab works on several aspects of inherited susceptibility to cancer, often generating data from individuals and families that present at his clinic. He is best known for his work on the clinico-pathological features of hereditary breast cancer, for his discovery of founder mutations in cancer susceptibility genes, and for his work on DICER1. Most recently, he identified the cause of small cell carcinoma of the ovary, hypercalcaemic type.

CACMID honours Dr. Mark Wainberg

Dr. Mark Wainberg, head of HIV/AIDS research at the LDI and Director of the McGill AIDS Centre, has been selected as the recipient of the 2014 John G. FitzGerald – CACMID Award by the Canadian Association for Clinical Microbiology and Infectious Diseases. This prestigious honour recognizes a Canadian microbiologist who has significantly advanced the field of medical microbiology. Dr. Wainberg is world renowned for his contributions to the development of antiretroviral therapies to treat HIV/AIDS, for his work on drug resistance, his advocacy on behalf of AIDS patients, and his on-going efforts to help discover a cure.

Post-doc recognized by Society of Toxicology

Alicia Bolt, a post-doctoral fellow in Dr. Koren Mann's lab was awarded second place in the Metals Specialty Section at the 2014 Society of Toxicology meeting for her poster presentation, *Tungsten Alters Bone Homeostasis by Decreasing Osteogenesis and Increasing Adipogenesis*.

Quebec invests to improve clinical research in cancer

The Quebec government has awarded \$9.2 million over four years to the Quebec-Clinical Research Organization in Cancer (Q-CROC), as part of the Fonds de partenariat pour un Québec innovant et en santé (FPQIS). Along with matching funds from the pharmaceutical industry, and the previously announced \$15 million investment by the federal government over five years, a total of \$32 million is being devoted to advancing personalized medicine in cancer.

Personalizing therapies based on molecular profiling and biomarker discovery of the individual genetic structure and mutations within a malignant tumor is revolutionizing cancer diagnostics and treatment. The Segal Cancer Centre and LDI are encouraging this revolution, helping to redefine different cancers as groups of rare sub-diseases and translating this new approach to clinical care by identifying treatments that directly target the genetic foundations of the cancer. Patients

have a higher probability for a successful outcome when tumor-specific molecular profiles are targeted.

A total of \$32 million has been invested in PreThera Research, a public-private national centre of excellence for personalized medicine in cancer

Q-CROC will work very closely with the newly created National Centre of Excellence PreThera Research for the creation

of a clinical and molecular database. These activities will be synergized with those of the Partnership for Personalized Medicine in Cancer, in which Q-CROC and many of its collaborators are already actively involved.

“Our private and public sector partners are proud to be key players in the effort to establish the necessary technological and human infrastructures to achieve personalized medicine in cancer,” affirmed **Dr. Gerald Batist**, Director of the Segal Cancer Centre and co-founder of Q-CROC. “This subsidy will provide resources crucial to the start-up and efficient conduct of clinical trials in several healthcare establishments, while equipping physicians with important clinical decision-making tools through the implementation of state-of-the-art IT platforms designed to integrate biomarker usage and targeted cancer treatments in their practice.”

Recruiting for clinical study on caregivers of advanced cancer patients

Caregiving for a loved one with advanced cancer is often rewarding but it can also be a stressful experience, eliciting a wide range of emotions as well as extreme fatigue. **Jamie Penner**, a registered nurse and doctoral candidate studying psychosocial oncology, is recruiting participants for a pilot study to determine whether small amounts of physical activity may alleviate feelings of stress and anxiety and keep caregivers healthy to continue providing care.

"Physical activity, even in small amounts, is proven to have a number of health benefits, including making people generally feel better, more energetic, and less stressed," Ms. Penner observed. "Many caregivers report that they have little time to pay attention to caring for themselves. Our study intends to examine whether an individually tailored physical activity program can be of benefit to family caregivers of patients with advanced cancer."

Study participants will receive coaching and support to engage in short periods of activity that they enjoy and can incorporate in their routines. Examples include walking, stretching, or yard work. The type and duration of activity will be tailored to the participant's personal circumstances, needs, interests, and abilities. The program entails two home visits, of approximately 30 minutes each, one week apart, followed by six weekly telephone calls of about 15 minutes each.

To participate, a person must be 18 years of age or older, able to speak and read English, and be involved in caring at home for a person with stage 3 or 4 cancer.

Family caregivers who may be interested in participating, or clinicians treating individuals whose family caregivers may be interested in participating, are asked to contact Jamie Penner at 514-340-8222, ext. 3426, or jamie.penner@mail.mcgill.ca.

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To submit information or for media enquiries, contact: Tod Hoffman at: thoffman@jgh.mcgill.ca; 514-340-8222, ext. 8661.

Merck Canada announces \$2 million grant to support personalized treatment in cancer

At the 6th Annual National Conference to Defeat Cancer, organized by Coalition Priorité Cancer, Merck Canada reinforced the company's continued commitment to the Quebec life sciences research innovation sector by announcing a \$2 million grant to the Quebec Clinical Research Organization in Cancer (Q-CROC).

The grant will go towards biomarker-driven clinical research for personalized medicine in cancer, notably in the framework of a public-private partnership put forward by Q-CROC and the newly-formed Centre of Excellence PreThera Research.

"This important contribution further solidifies our position as a provincial interface for clinical research to help bridge the gap between industry, government, healthcare establishments and the research community in Quebec. The grant from Merck Canada will allow us to get closer to our ultimate goal of providing cancer patients in Quebec with access to the most advanced and personalized medicine," said **Dr. Gerald Batist**, co-founder of Q-CROC and Director of the Segal Cancer Centre.

Dr. Thombs wins inaugural Principal's Award for Research

Dr. Brett Thombs, Senior Investigator at the LDI, William Dawson Scholar and Associate Professor of Psychiatry at McGill University, has been awarded the inaugural version of the McGill Principal's Prize for Outstanding Emerging Researchers. The Prize was established to honour faculty members within 10 years of their doctoral degree who have distinguished themselves through exceptional contributions to research in their field. Dr. Thombs arrived at McGill in 2006 following his PhD at Fordham University and a postdoctoral fellowship at Johns Hopkins University. He is known internationally for his efforts to improve the psychological well-being of those living with chronic diseases, his evidence-based critiques of how depression screening is employed in medical settings, and his work on how research methodology can influence the validity of results.

9th Annual Psychiatry Research Day

This year's 9th Annual Department of Psychiatry Research Day featured talks offering new perspectives on how categorizing mental illness as brain disease affects diagnosis and treatment of mental health problems. The event was attended by a large audience of clinicians and researchers.

Speakers from the LDI included:

- **Dr. Amir Raz** on "The taking of a pill is sometimes more meaningful than the actual pill you are taking ;"
- **Dr. Suparna Choudhury** on "Are we our brains? Cognitive neuroscience and the politics of brain-based identities;"
- **Dr. Laurence Kirmayer** on "Knowledge translation in global mental health: Mental health literacy, political economy, and indigenous psychiatries."

Psychiatry Research Day at the JGH is now in its 9th year presenting research from the Institute for Community and Family Psychiatry

The discussant was Dr. Ian Gold, Canada Research Chair in Philosophy and Psychiatry at McGill.

Diabetes project wins first prize at regional science fair

Alexandra Cohen, a secondary V student at St. George's School, who has been mentored in **Dr. Lawrence Rosenberg's** lab at the LDI over the past two years, won the Hydro-Quebec First Prize for best project at the Montreal Regional Science & Technology Fair, as well as a \$3000 scholarship to McGill at the provincial science fair in Terrebonne, and an invitation to the Intel International Science & Engineering Fair in Los Angeles. Her project involved extending the half-life of the islet neo-genesis-associated protein (INGAP) to enhance its insulin-producing capabilities and realize its potential as a treatment for Type 1 and 2 diabetes.

Dr. Carmen Loisel appointed Co-Director of Segal Cancer Centre

Dr. Carmen G. Loisel has been appointed as Co-Director for Strategic Orientation/Academic of the Segal Cancer Centre, where she will provide strategic leadership in patient experience innovation, research and in the recruitment and mentoring of the Segal's new clinical and research staff.

Dr. Loisel completed doctoral training in Nursing and Psychology at the University of Wisconsin-Madison. She is Director of McGill University Oncology Nursing as well as Associate Professor at the McGill Ingram School of Nursing, where she is the Christine and Herschel Victor/Hope & Cope—McGill Chair in Psychosocial Oncology and has a long list of academic accomplishments in trans-disciplinary fields.

Register NOW: 5th Annual LDI Scientific Retreat

The 5th Annual Scientific Retreat will be held at the Holiday Inn—Midtown, 420 Sherbrooke West, on Friday May 30, beginning at 8:45 a.m.

The keynote address will be delivered by Dr. Morag Park, Director of the Goodman Cancer Research Centre at McGill. She will speak on "Cancer Research in the Post Genomic World: Back to the Basics."

Rebecca Reich, Business Development Director at Mitacs will discuss how they can help academics establish partnerships outside of academia.

Speakers from the LDI's axes include Dr. Prem Ponka (hemovascular), Dr. Marc Fabian (cancer), Dr. Susan Kahn (epidemiology), Dr. Robin Cohen (psychosocial), and Dr. Lawrence Kleiman (HIV/AIDS). Dr. Colin Crist will deliver the new PI talk.

Eight trainees, selected through competitive review, will give presentations on their research. Posters will also be presented for competitive judging. The day concludes with an awards presentation.