



Genome Québec

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Press Release

Québec Researcher Catches DNA “Spelling Mistakes” Linked to Breast Cancer

Montréal, March 27, 2013 – Working with an international consortium conducting research on breast, ovarian and prostate cancer among 200,000 people, Professor Jacques Simard, researcher at the CHU de Québec Research Centre and holder of the Canada Research Chair in Oncogenetics at Université Laval, was actively involved in the discovery of DNA “spelling mistakes” linked to breast cancer. These mistakes – known as genetic variations – are directly involved in the risk of developing breast cancer.



The research was part of an international study on an unprecedented scale performed by the largest international consortia, the Collaborative Oncological Gene-Environment Study (COGS), coordinated by Professor Douglas Easton of Cambridge University. Given the scope of this international study and the impact of its findings, it will be featured in a series of articles in the prestigious science journal *Nature Genetics* on March 27, 2013. The results will also be presented in three additional articles published simultaneously in *PLoS Genetics* and *The American Journal of Human Genetics*.

Giant leap forward in breast cancer research

For over 20 years now, Professor Simard has been working on genetic susceptibility to breast cancer. These latest research findings helped identify 49 new genetic variations involved in the risk of developing this disease, almost tripling the number of variations now known. In the vast majority of breast cancer cases affecting the general population, it is the interaction between these genetic variations and other environmental and lifestyle factors that is involved in the development of the disease.

Professor Simard explains: “The vast amount of genetic information gathered through this study makes it one of the most significant breakthroughs in recent years in terms of understanding the inherited risk factors of breast cancer. It is now possible to develop a DNA profile where 5 percent of women have one in four chances of developing the disease.” By understanding the genetic causes and their interaction with environmental factors, Professor Simard is paving the way for better prediction of breast cancer risk.

Québec technological expertise at the heart of this achievement

Half of the samples used in the study – over 100,000 – were analyzed in Montréal by genomic experts working at the McGill University and Génome Québec Innovation Centre. Génome Québec President and CEO, Marc LePage, notes: “It was Professor Simard’s leadership that secured the involvement in the project of the McGill University and Génome Québec Innovation Centre. He positioned the Centre’s expertise and attracted many international teams, who could have had their findings analyzed anywhere in the world. This is unequivocal proof that we have built a genomic infrastructure of international repute. We are proud of our role in this worldwide study and applaud the work of all the researchers who participated, including Professor Mark Goldberg of the Research Institute of the McGill University Health Centre, who spearheaded the portion of the study on the interaction between genetic and environmental factors and its impact on the risk of developing breast cancer.”



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The \$1.5 million investment by the Quebec Breast Cancer Foundation played a key role in enabling Québec researchers to take part in the international consortium. Francine Cléroux, Executive Director of the Foundation, explains: “It is a major scientific breakthrough in breast cancer research and we are thrilled with the news. This discovery will make it easier to identify those women most at risk for the disease and offer them early screening and targeted prevention approaches.”

Other partners include the Ministry of Higher Education, Research, Science and Technology and the Canadian Institutes of Health Research (CIHR), which funded the international interdisciplinary research team working on the inherited risk of breast cancer spearheaded by Professor Simard.

About the McGill University and Génome Québec Innovation Centre

The McGill University and Génome Québec Innovation Centre is a world-class research facility for genomics and proteomics. Founded in 2002, the Centre has developed world-renowned expertise in complex genetic disorders, such as cardiac disease, asthma and Type 2 diabetes and cancer, and has become a resource and networking site for various research initiatives in human health, forestry, infectious diseases, agriculture and environment. Major funding for the McGill University and Génome Québec Innovation Centre is provided by the Government of Canada through Genome Canada, and the Government of Québec. For more information, visit www.genomequebec.com

About the Quebec Breast Cancer Foundation

The Quebec Breast Cancer Foundation is a non-profit organization with a mission to fund breast cancer research, promote breast health through education and awareness activities, and provide support for those affected by the disease. For additional information, please visit www.rubanrose.org or observationdesseins.org.

About the Canadian Institutes of Health Research

The Canadian Institutes of Health Research (CIHR) is the Government of Canada's health research investment agency. CIHR's mission is to create new scientific knowledge and to enable its translation into improved health, more effective health services and products, and a strengthened Canadian health care system. Composed of 13 Institutes, CIHR provides leadership and support to more than 14,100 health researchers and trainees across Canada.

About the CHU de Québec

Comprising the CHUL, l'Hôtel-Dieu de Québec and l'Enfant-Jésus, Saint-François d'Assise and Saint-Sacrement hospitals, the CHU de Québec is the leading health care establishment in Québec and one of the largest university medical centres in Canada. It offers general and specialized care, but also superspecialized services to the residents of Eastern Québec, a population of nearly two million people. Working closely with the Université Laval, the CHU de Québec is a forward-looking institution that also fulfils a teaching and research mission in many specialized niches of excellence and in the evaluation of technologies and health intervention modes. It carries out this mission with the support of nearly 14,000 employees, 1,700 doctors, dentists and pharmacists and 550 researchers. To learn more about the CHU de Québec, go to www.chudequebec.ca

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