



Genome Québec



Genome Canada



Ariosa Diagnostics, CHU de Québec and Genome Canada to Launch Large-Scale Cost-Effectiveness Study of Non-Invasive Prenatal Testing in Canada

Québec, Canada, August 12, 2013 – A team of researchers led by CHU de Québec and Université Laval, has received CAN\$10.5 million from Genome Canada, the Canadian Institutes of Health Research (CIHR), Génome Québec and other partners to conduct a large-scale comparative effectiveness study on non-invasive prenatal screening techniques. Dr. François Rousseau, professor at the Faculty of Medicine of Université Laval and researcher at the CHU de Québec, and his team will examine current prenatal screening practices for chromosomal conditions, such as Down syndrome, among pregnant women in Canada in order to improve screening approaches and avoid unnecessary procedures.

Each year in Canada, about 450,000 women become pregnant and are offered Down syndrome prenatal screening using biochemical and ultrasound markers. The vast majority will be negative or low risk. However, for positive or high-risk results, those pregnant women are referred to amniocentesis, which is an invasive procedure done to confirm the screening result. Approximately 5% of all biochemical screening results are falsely positive, attributing to unnecessary invasive procedures that pose a 1 in 300 risk for miscarriage.

“Génome Québec is a strong supporter of public-private partnerships, such as the one developed between Ariosa Diagnostics and Dr. François Rousseau’s team. The PEGASUS: PErsonalized Genomics for prenatal Aneuploidy Screening USing maternal blood project, made possible through such partnerships in personalized medicine, will help meet women’s healthcare needs by giving them access to safer prenatal screening tests”, explains Marc LePage, Génome Québec’s President and CEO.

The research project aims at independently comparing the performances of different such approaches that involve various combinations of the available tools for screening of chromosomal conditions, as well as to evaluate the cost-effectiveness, the ethical and social aspects of this new technology and to identify and adapt the best implementation tools for users in the health care system. The researchers will recruit 5,600 pregnant women (3,600 at high-risk of trisomy conditions and 2,000 at low-risk). Samples from these women will be tested in parallel using different screening approaches that involve genomic-based NIPT, but also existing or new biochemical and ultrasound screening tools. The samples will be analyzed without knowledge of the true status of the pregnancy outcome. This will provide a comprehensive evaluation of the most efficient ways to improve the prenatal screening techniques widely used today.

Ariosa Diagnostics is one of the major commercial partners for this project and will provide testing for part of this large sample that will include mainly low-risk women. The investigators have chosen the Harmony™ Prenatal Test as the commercially available NIPT assay to be included as one of the various screening tests that will be compared.

Dr. Rousseau, Leader of the Project, mentions: "We are happy that Ariosa Diagnostics has joined this effort and provided a significant contribution to this Project. Ariosa's Harmony test is the most affordable commercial NIPT available, and it was chosen by the research team for this study because it is the most likely to be used in the Canadian context."

Dr. Thomas Musci, VP of Clinical and Medical Affairs at Ariosa remarked that “Ariosa, as a core corporate value, is committed to furthering rigorous clinical and scientific study of cell-free DNA technology and the Harmony Prenatal Test in a variety of clinical settings and in a number of different countries. We are extremely pleased to support this collaboration with Genome Canada and fully support the goals of their research efforts.”

Dr. Serge Rivest, Director of the Research Centre at the CHU de Québec, stated that "The leadership of the researchers from CHU de Québec Research Center was key in putting this exceptional academic team together to tackle one of the hottest clinical application of next-generation sequencing in the context of a population-based screening program."

Université Laval's Vice-Rector, Research and Innovation, Sophie D'Amours, points out the exceptional quality of the research conducted by Dr. Rousseau and his team as well as its concrete impact on prenatal care: "The implementation of new screening techniques resulting from their work will significantly contribute to women's well-being in terms of effectiveness, safety and non-invasiveness."

About Génome Québec

Since May 2000, Génome Québec has been the driving force behind the development of genomics in Québec. By supporting nearly 80 projects and 800 researchers and managing the operations of the McGill University and Génome Québec Innovation Centre, Génome Québec is helping to accelerate the discovery of new applications for genomics in strategic areas, such as human health, forestry and the environment.

The funds invested by Génome Québec are provided by the Ministry of Higher Education, Research, Science and Technology, the Government of Canada, through Genome Canada, and private partners.

For more information, visit www.genomequebec.com.

About Genome Canada

Genome Canada is a not-for-profit organization that invests in genomics research to generate economic and social benefits for Canadians. Genome Canada builds bridges between government, academia and industry to forge a genomics-based public-private innovation enterprise focused on key life science sectors. We develop these partnerships to invest in and manage large-scale research and translate discoveries into commercial opportunities, new technologies, applications and solutions. For more information, visit www.genomecanada.ca

About CIHR

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.

www.cihr-irsc.gc.ca

About CHU de Québec

Consist of the CHUL, L'Hôtel-Dieu de Québec and hospitals l'Enfant-Jésus, Saint-François d'Assise and du Saint-Sacrement, the CHU de Québec is the most important health care institution in the province of Québec and one of the biggest in Canada. The CHU de Québec offers general and specialized care, but mostly ultra-specialized care throughout Eastern Quebec, which represents nearly two million persons. In partnership with Université Laval and focused on the future, the CHU de Québec also has core missions in teaching, in research in many fields of excellence and in the evaluation of health technologies and professional practices. Are needed to accomplish those missions 14,000 employees, 1,700 doctors, dentists and pharmacists and also 500 researchers. For more information, visit www2.chudequebec.ca

About Université Laval

Located in Quebec's historic capital, a World Heritage City, Université Laval is the first French-language university in North America. It is one of Canada's leading research universities, ranking 7th among the country's 94 university-level institutions in terms of research funding with \$300 million devoted to research last year. Université Laval's 1,400 professors-researchers share their knowledge with 48,000 students, 10,000 of whom are enrolled in graduate-level programs. www.ulaval.ca

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