



**CHU de Québec Collaborates with PerkinElmer on a study funded by  
Genome Canada, Génome Québec, CIHR and Genome Alberta  
using DNA sequencing alone or in a contingent model with first trimester  
serum markers as non-invasive option to determine risk of Down Syndrome**

**Montréal, November 4, 2013** – A team of clinical researchers led by CHU de Québec has received CAN\$10.5 million from Génome Québec, Genome Canada, CIHR, Genome Alberta and other partners to recruit 5,600 pregnant women in Canada to study the potential of using DNA sequencing alone or in a contingent model with first trimester serum markers as a non-invasive method for detecting birth defects. The four marker first trimester serum quadruple screen, 1T-Quad (PAPP-A, Free beta hCG, AFP and PIGF) provided by [PerkinElmer](#) will be used by researchers to validate its use in a contingent model where women identified by the 1T-Quad screen as having an increased risk of fetal aneuploidy undergo analysis of cell-free circulating fetal DNA from a standard blood draw to better estimate their risk of Down syndrome or other trisomy.

Each year 450,000 Canadian women become pregnant and participate in population-based pre-natal screening. With current approaches, up to 10,000 women undergo amniocentesis, an invasive procedure that carries a risk of pregnancy loss. Using the 1T-Quad screen followed by DNA sequencing of cell free DNA in women found at higher risk on serum screening, researchers aim to increase the detection rate of Down syndrome while reducing the use of amniocentesis to a smaller subset of women, as well as making best use of scarce health care resources.

“Recent data suggests that using the 1T-Quad screen to contingently select 10-20% of women at highest risk for non-invasive fetal DNA testing provides a novel way to integrate genomic testing into clinical usage that is both cost-effective and associated with excellent performance” said Dr. Jo-Ann Johnson, one of the principal investigators of the project, from University of Calgary.

Furthermore, Dr. François Rousseau, the project Leader from Université Laval, added that “We are fortunate that PerkinElmer has agreed to join this initiative and offer to evaluate a potentially very effective test for first trimester screening that combines both biochemical blood tests and genomic-based non-invasive assays, with a short turn-around time, which is key in the context of an ongoing pregnancy.”

“This technology used in Dr. Rousseau’s project demonstrates the importance of partnerships between the public and private sectors, particularly in the area of research into personalized medicine. Researchers have to carry out their work in order to meet the patient’s needs, in this case by developing more efficient prenatal screening tests for women’s health,” explains Marc LePage, President and CEO of Génome Québec.

“Genome Canada is pleased to support Dr. Rousseau’s project and commends the partnership with PerkinElmer. This project, which is one of seventeen large-scale genomics and personalized health projects we are supporting across Canada, has the potential to transform the way health care is delivered for Canadians, including improvements in clinical practice, better treatment and outcomes for patients and a more efficient, cost-effective health care system,” said Pierre Meulien, President and CEO of Genome Canada.

“The outcomes of Dr. Rousseau’s project will provide doctors and patients with meaningful, clinic-ready tools that are necessary to make clear and informed decisions”, said Dr. Alain Beaudet, President of the Canadian Institutes of Health Research. “We are at a threshold in research, where advancements in genetic discovery will provide the opportunity to deliver a more stratified approach to diagnosis and treatment. As this continues, partnerships at all levels are ever more important”.

“We are proud that our researchers have taken the leadership for this important translational research project in the field of personalized genomics,” said CHU de Québec’s Research Director, Dr. Serge Rivest. “This shows the strength of the CHU de Québec Research Center’s expertise in evidence-based laboratory medicine, in pregnancy related research and in personalized medicine.”

“This area of research is vital in continuing to advance access to reliable first trimester screening in support of healthy pregnancies for women across Canada,” said Yvonne Parker, Vice President, Maternal and Fetal Health, PerkinElmer, “The IT QUAD as a front-line screen has the potential to cost-effectively, identify a subset of women, who would benefit from the new non-invasive approach. “

### **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](http://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, innovation-driven 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 <http://www.genomecanada.ca>

### **About the Canadian Institutes of Health Research**

The [Canadian Institutes of Health Research](http://www.cihr.gc.ca) (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 health care system for Canadians. Composed of 13 Institutes, CIHR provides leadership and support to more than 14,100 health researchers and trainees across Canada.

### **About Genome Alberta**

Genome Alberta is a publicly funded not-for-profit corporation that initiates, funds, and manages genomics research and partnerships. We strive to be a leading source of information and administration related to genomics, proteomics, bioinformatics and bioethics research in Alberta. Visit us at [genomealberta.ca](http://genomealberta.ca) or check out news for the 'omics' generation at [GenOmicsNews.ca](http://GenOmicsNews.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](http://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](http://www.ulaval.ca)

### **About PerkinElmer, Inc.**

PerkinElmer, Inc. is a global leader focused on improving the health and safety of people and the environment. The company reported revenue of approximately \$2.1 billion in 2012, has about 7,500 employees serving customers in more than 150 countries, and is a component of the S&P 500 Index. Additional information is available through 1-877-PKI-NYSE, or at [www.perkinelmer.com](http://www.perkinelmer.com). Join the conversation and follow us on twitter at [www.twitter.com/perkinelmernews](http://www.twitter.com/perkinelmernews)

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