



November 4, 2013

## **PerkinElmer Collaborates with CHU de Québec on a Study Funded by Genome Canada, Génome Québec, CIHR and Genome Alberta**

*The study will evaluate DNA sequencing alone or in a contingent model with first trimester serum markers as a non-invasive option to determine risk of Down Syndrome*

**WHAT:** [PerkinElmer, Inc.](#), a global leader focused on improving the health and safety of people and the environment, is providing a four marker first trimester serum quadruple [screen](#) for a study to evaluate the effectiveness of a non-invasive prenatal screen to decrease the rate of higher risk invasive procedures. The study is led by [CHU de Québec](#) and will involve 5,600 pregnant women identified with 1T-Quad (PAPP-A, Free beta hCG, AFP and PIGF) as having an increased risk of fetal aneuploidy. The team of clinical researchers will use the screen 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.

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.

"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 1T 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."

"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 collaborations 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 collaboration 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, collaborations 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."

The study is funded by [Genome Canada](#), [Genome Québec](#), [The Canadian Institutes of Health Research \(CIHR\)](#) and [Genome Alberta](#). Researchers from [Université Laval](#) will be contributing to this research study.

#### **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<sup>®</sup>, or at [www.perkinelmer.com](http://www.perkinelmer.com).

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Media Contact:

Jen Reid Edelman (on behalf of PerkinElmer)

Phone: 404-460-8587

Email: [jennifer.reid@edelman.com](mailto:jennifer.reid@edelman.com)