CIHR IRSC

EVIDENCE IN THE MAKING

A BETTER PRENATAL TEST

Reducing Risk for Pregnant Women

THE CHALLENGE

Every year, 10,000 pregnant women undergo amniocentesis in Canada to test for conditions such as Down syndrome that are caused by extra or missing chromosome material, and 70 will lose healthy fetuses due to complications from this invasive procedure. With the recent discovery that fetal DNA is present in maternal blood, various researchers have developed blood tests as a non-invasive method for prenatal testing. While no test is perfect, published studies suggest that such blood tests could possibly screen out low-risk women from having to take an amniocentesis. However, studies done independently from the owners of the technology patents on these tests are needed. "The different approaches have never been compared head-to-head in the same sample of women to determine which are most effective," says Dr. François Rousseau of Laval University.

Research: As part of a four-year project funded by Genome Canada and CIHR, Dr. Rousseau and Dr. Sylvie Langlois of the University of British Columbia are co-leading a large pan-Canadian study involving 28 other investigators. Together, they will recruit 5,600 pregnant women to evaluate the effectiveness of different prenatal screening approaches, both in terms of results and value for money. In addition to conducting a real-life comparative study of the methods and computer-simulated economic analyses, the researchers will explore ethical, legal and social issues and lay the groundwork for eventual uptake of the "best" technique by health care professionals. "It's much less glamorous than discovering a gene or inventing a technology, but there's a lot of work to do before a potential health care application can be put into routine clinical use," says Dr. Rousseau. "We see the finish line, and that's what's exciting. This could really improve testing and lower risk for women."

Sources: Langlois, S., et al. "Current status in non-invasive prenatal detection of Down syndrome, trisomy 18, and trisomy 13 using cell-free DNA in maternal plasma," *Journal of Obstetrics and Gynaecology Canada* 35, 2 (2013): 177–81.

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Spring 2014, Volume 2, Issue 2, Insert 2

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