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### Prenatal screening test could limit need for amniocentesis

CALGARY — Local researchers are evaluating a new prenatal screening test that assesses fetal DNA present in the mother's blood to check for chromosomal disorders in the developing fetus.

It is the first time the test, known as non-invasive prenatal testing (NIPT), is being evaluated on a large scale in Canada.

The main benefit of NIPT is that it could substantially reduce the need for invasive tests such as amniocentesis, a diagnostic procedure that draws fluid from the amniotic sac surrounding the unborn baby. Although amniocentesis is a safe and well-established procedure, it is associated with a 0.5 to one per cent risk of miscarriage.

About 10,000 Canadian women undergo an amniocentesis each year as part of prenatal screening, with about 70 of those pregnancies being lost as a result of complications from the procedure. Researchers estimate NIPT screening, which studies have shown to be about 99 per cent accurate, could reduce the number of amniocentesis procedures in Canada each year to about 300.

NIPT is available through some U.S. companies on a patient-pay basis.

"While these companies have shown NIPT to be highly accurate, it has never been validated on a large scale outside of industry, nor has its role in the Canadian public health system been assessed," says Dr. JoAnn Johnson, an Alberta Health Services obstetrician, University of Calgary professor of obstetrics and gynecology, and the principal investigator in the Calgary arm of the study.

"This is a complex technology. We hope that Canadian data will help inform decision-making about how NIPT might be introduced here," says Dr. Johnson, who is also a member of the Alberta Children's Hospital Research Institute.

Calgary is one of five sites across the country involved in PEGASUS (Personalized Genomics for prenatal Aneuploidy Screening Using maternal blood), a \$10-million study funded by Genome Canada headed by Drs. Francois Rousseau (University of Laval) and Sylvie Langlois (University of British Columbia). The purpose of the study is both to develop NIPT technology in Canada and to determine the best way to integrate it into the Canadian health care setting.

Researchers plan to recruit approximately 5,000 women, 900 of them from Calgary. The study here is being delivered through a collaboration between AHS, the University of Calgary, EFW Radiology and Calgary Laboratory Services.

"Women can contact us directly if they're interested in learning more about the study," says study co-ordinator Priyana Sharma. "All that's required is that they undergo an ultrasound to confirm gestational age, and then provide a blood sample at about 10 weeks, which will be tested using the new technology. Then, when they come in for their first trimester screen, they'll be given the results of that test."

Women interested in participating in the study should phone 403-943-8382.

The costs of the new screening test are covered by the research study.

The current blood test that women receive in first trimester screening measures the levels of two substances found in the blood of all pregnant women, but which tend to be different in pregnancies with Down syndrome. That test, however, can't determine for certain if the baby has a chromosomal condition and only provides information needed to make an informed decision about additional testing.

A chromosomal condition means there are extra or missing chromosomes, or pieces of chromosomes have rearranged themselves. A baby's intellectual and physical development can be affected. Down syndrome is the most common chromosomal condition and becomes more common as a woman gets older.

Mazlina Navarre, who is expecting her second child in March, says she didn't hesitate to participate when she learned about the study.

"I'm 38, so I'm in a higher-risk category. When you're pregnant, you always want to know that everything is going well for the baby, so for us having the reassurance was very important. It alleviates a lot of anxiety."

Alberta Health Services is the provincial health authority responsible for planning and delivering health supports and services for more than four million adults and children living in Alberta. Its mission is to provide a patient-focused, quality health system that is accessible and sustainable for all Albertans.

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**B-roll is available at the ftp site below. One clip is of an amniocentesis procedure, the other is of study participant Mazlina Navarre having blood drawn and undergoing an ultrasound.**

URL: <ftp://208.118.126.84>

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