



THE UNIVERSITY OF BRITISH COLUMBIA

Department of Medical Genetics

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Research study: Personalized Genomics for prenatal Aneuploidy Screening Using maternal blood: Towards First Tier Screening and beyond.

What is the Study?

Researchers are looking for safer and more effective ways to test for Down syndrome and other chromosomal abnormalities during pregnancy. This is a study being conducted in multiple centers in British Columbia and Quebec. Dr. Sylvie Langlois is involved in the study by recruiting pregnant patients who will agree to donate a sample of blood for the study, provide information about their pregnancy and their newborn baby and complete on line questionnaires related to their experience with screening. Pregnant women are being invited to consider participating in this research study but your participation is totally voluntary.

What Are You Testing in My Blood?

We are testing DNA. DNA makes up the genes in your body. As your baby develops, a very small amount of its DNA naturally gets into your bloodstream and can be measured there. Testing this DNA using new technologies has allowed the development of a new test to detect Down syndrome. This test is known as NIPT for Non Invasive Prenatal Test.

Why do you Need My Help?

We are asking you because you have chosen to have screening for Down syndrome. We want to find out how NIPT as a screening test for Down syndrome and other chromosomal abnormalities compares to the standard screening approach offered to all pregnant women in British Columbia.

What Does the Study Involve?

If you are interested in participating, details of the study will be provided to you. In summary, participating in the study involves: 1. being selected on a random basis to undergo either the standard screening for Down syndrome OR NIPT (One-third of participants will be randomly assigned to the standard screening group; two-thirds to the NIPT group); 2. give a sample of your blood for the clinical test and research; 3. remain in touch with the study team throughout your pregnancy to provide information about your pregnancy and newborn baby; 4. complete on line questionnaires three times during your pregnancy; and 5. in the event your NIPT result is positive, you may be asked to allow sampling of your placenta after delivery.

Will I Get the Results?

Your midwife or doctor who looks after your pregnancy will receive the results of the test you were selected for, either the standard screen or NIPT, and he/she will share those results with you.

How Will This Study Help Me?

This study is not designed to help women who participate in the study. However, if you are selected for receiving NIPT, you may benefit from receiving earlier results. Furthermore, we hope that the study will help you and many other women in the future. The information from the study will help health care decision makers make decisions about the value of using NIPT for prenatal screening for Down syndrome in Canada.

Will I be compensated for participating in this study?

No. You will not be compensated for participating in this study.

What should I do if I am interested in participating in this study?

If you are interested in participating in the study, you can contact the study team by email at pegasus@cw.bc.ca, or phone at 604-875-3600 ext. 6789. Please provide your contact information (both phone and email) so we can reply with more details about the study.