For Parents: Non-invasive Prenatal Testing (NIPT) Factsheet

What is NIPT?
Non-invasive prenatal testing (NIPT) is a new way to *screen* your pregnancy to see if your baby has an increased chance of having a few specific chromosome disorders. The chromosome disorders are Down syndrome, trisomy 18 and trisomy 13. In any pregnancy, a small amount of the developing baby's genetic information (DNA) is in the mother's blood. This test is done by looking at the baby's DNA in the mother's blood. The purpose of NIPT is to find babies with a high chance of having the specific disorders mentioned above (mainly Down syndrome).

What information can NIPT provide?
NIPT can find about 98-99% of developing babies with Down syndrome. It is sometimes used to tell if a baby has trisomy 18 or trisomy 13; however, NIPT is not as good at finding trisomy 18 or 13 compared to looking for Down syndrome. NIPT is not perfect. There is a small chance (<2%) that NIPT will say that a baby *does* have Down syndrome when, in fact, it does not (i.e. false positive). Similarly, there is a small chance that NIPT will say that a baby does *not* have Down syndrome when, in fact, it does (i.e. false negative).

How and when is NIPT done?
NIPT is done by taking a blood sample from the mother early in pregnancy, usually around 10 weeks of pregnancy. An ultrasound is needed before having NIPT to date the pregnancy accurately and to be sure there is only one baby. An ultrasound is not needed at the time the blood test is taken.

Are there any risks of NIPT to the baby?
NIPT is a blood test done on the mother. It does not hurt the developing baby and does not increase the chance of a miscarriage. This is different from other types of testing such as CVS (chorionic villus sampling) and amniocentesis.

Does NIPT replace CVS/amniocentesis?
No. At the present time, NIPT is a *screening test,* meaning that it cannot tell for certain if your baby has Down syndrome. If NIPT detects Down syndrome in a baby, more testing is needed to see if the baby really has the chromosome change. This is done by tests called chorionic villus sampling (CVS) or amniocentesis. CVS and amniocentesis tell for sure if the baby has Down syndrome, trisomy 18 or trisomy 13. Both amniocentesis and CVS have a small chance of miscarriage.

Who is NIPT for?
Currently, NIPT has mostly been studied in women who have a higher chance of Down syndrome. It is not known how accurate this test will be in women who have a low chance of their baby having Down syndrome. Studies are being done currently to find that out. Also, it cannot be used in women with multiple babies (e.g. twins) and cannot be used in pregnancies using an egg donor. NIPT can be used if you've had a previous pregnancy because the baby's DNA is gone from the mother's blood just hours after the baby is born.

How is NIPT different from the currently available prenatal screening?
Current prenatal screening tests involve one or two blood samples and usually an ultrasound to measure the thickness of the back of the baby's neck (nuchal translucency). Depending on the type of screen, information about the *chance* of the baby being born with Down syndrome, trisomy 18 and sometimes trisomy 13 and open neural tube defects (ONTDs) such as spina bifida, is provided. Current prenatal screening tests are not as accurate as NIPT for Down syndrome. For example, of every 100 pregnancies where the baby has Down syndrome, current screening tests can identify 80-90 of the affected pregnancies. NIPT can find 98-99 of the pregnancies where the baby really has Down syndrome. NIPT does not look for ONTDs.

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Is NIPT available in Ontario?
Since NIPT is a new test, it is not widely available in Ontario and is presently not covered by OHIP. Women who want NIPT must pay for the test and pay to have their blood sent to the laboratory which is in the United States of America for testing. Women who want the test must find a physician or health centre offering NIPT. The Prenatal Screening Subcommittee is currently working in its advisory capacity to recommend NIPT as part of an updated provincial prenatal screening program. Until then, the availability of and use of NIPT will be inconsistent throughout Ontario. A list of providers is currently not available. The Prenatal Screening Committee will do its best to inform patients and providers of updates regarding NIPT in Ontario.

What is the Prenatal Screening Subcommittee recommending?
At the present time, the Prenatal Screening Subcommittee is recommending that all women considering having prenatal screening for Down syndrome use the tests available in Ontario, which are covered by OHIP. If the prenatal screening test result is screen positive or if a woman has a higher chance of having a baby with a specific chromosome problem, NIPT may be considered. The woman would have to be willing to pay for NIPT and see a physician or centre offering the test. If the NIPT test results say it is likely the baby has a chromosome problem, the recommendation is that the results should be confirmed by a diagnostic test, specifically CVS or amniocentesis. Until scientific studies are completed, NIPT is not being recommended as a first line screening test for women at low risk of having a baby with Down syndrome, trisomy 18 or 13.