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PUBLIC

PRENATAL SCREENING
NON-INVASIVE PRENATAL TESTING (NIPT)

What is prenatal screening?

Prenatal screening is about checking the health and development of your baby before it is born. Prenatal screening cannot tell your for certain if your baby does or does not have a condition.

This resource is about one kind of prenatal screening called Non-Invasive Prenatal Testing (NIPT).

What does NIPT look for?

NIPT looks at DNA in your blood. While most (90%) of the DNA in your blood is yours, some (~10%) comes from the pregnancy (specifically, the placenta).

NIPT will report on the chance that your baby has a specific genetic condition: Down syndrome (trisomy 21), trisomy 18, trisomy 13. Every pregnant person has a chance to have a baby with one of these conditions, and that chance increases with the age of the person when the baby is born. Nobody can cause or prevent these conditions; they occur by chance.

NIPT can also report on the sex chromosomes (male or female). Sometimes NIPT will report that there might be an extra or missing sex chromosome.

More on the conditions screened for:

Down syndrome, trisomy 18, trisomy 13 are genetic conditions where there is *extra* genetic information. The genetic library is a collection of recipes needed for healthy growth and development. Having extra (or missing) genetic information affects health and development. This can show up as intellectual disability from mild to severe, a greater chance for health conditions, or differences at birth that could require surgery or other medical intervention. Down syndrome occurs in 1 in 780 births. Trisomy 13 and 18 are more rare. Trisomy 18 occurs in 1 in 6,000 births and trisomy 13 occurs in 1 in 10,000 -1 in 25,000 births.

There are several conditions with sex chromosome differences where there could be extra or missing chromosome(s) (the X and/or Y chromosome). These are more common and happen in about 1 in 500 individuals. There is a lot of variation between the different conditions depending on which chromosome is extra or missing.

Can NIPT tell me if my baby will be healthy?

No

There is no test to guarantee a healthy baby. NIPT will only look for the chance the baby has one of the specific genetic conditions screened for.

Do I have to have NIPT or any prenatal screening?

No

It is your choice whether or not to have prenatal screening.

Can NIPT hurt me or my baby?

No

NIPT is a blood test. There is no harm to you or the baby.

When can I have NIPT?

The blood can be drawn anytime after 10 weeks' gestation. Results are available in 7-10 business days..

Can NIPT tell me any other information?

Because NIPT testing is looking at DNA from both the pregnancy and from you, a result could reveal information about your DNA or health. In rare cases, an appointment with a genetics specialist would be offered.

More details about NIPT and these genetic conditions can be found here

www.geneticseducation.ca >Public resources

What will the results say?

The results will most often say that there is a *very low* chance for the baby to have one of these conditions (less than 0.01% or 1 in 10,000).

Sometimes the results will say there is a *very high* chance the baby has one of the conditions (higher than 99%).

Sometimes the report will say *no result* which means that the laboratory was not able to get a clear result.

Remember NIPT is a screening test. It cannot tell you for sure if you baby has the condition. Only a diagnostic test can.

Diagnostic tests are amniocentesis or chorionic villus sampling, performed by a maternal-fetal medicine specialist. These tests do have a small risk to the pregnancy.

A second trimester ultrasound to look at the baby's growth and development is recommended for all pregnancies. Additional testing could also be considered in some cases (e.g. a birth difference on ultrasound, a family history of a condition).

Your healthcare practitioner will discuss all of your options and support any choice you make.

How good is NIPT?

NIPT will identify almost all pregnancies where the baby has Down syndrome, trisomy 18, or trisomy 13. It is the best prenatal screening test to identify these conditions.

Is prenatal screening right for me?

Some questions to consider before NIPT:

- Do I want to know if my baby has one of these conditions before birth?
- What would the results mean for me in this pregnancy?
- Would I consider ending a pregnancy that had one of these conditions?
- Would it be important for me to plan, identify support, and get prepared for a baby that has one of these conditions, before the baby is born?
- Would I have a diagnostic test? Would I fly/travel to another city to have a diagnostic test?

  A diagnostic test the only way to know *for sure* if the baby has the condition before birth (i.e. amniocentesis or chorionic villus sampling). These tests have a small risk to lose the pregnancy (miscarriage).