Milton Centre For Women's Health

Prenatal Genetic Testing

What is Prenatal Genetic Testing?

Prenatal genetic testing consist of a blood test and ultra sound available to all pregnant individuals to test for the chance that your baby is born with one of the following health conditions:

Trisomy 18, Trisomy 21, neural tube defects, Trisomy 13, and sex chromosome differences

Although most babies are born healthy, all individuals have a chance of having a baby with a health condition even if they and their families are healthy.

What Tests are Available?

Multiple Marker Screening: is funded by the Ministry of Health for all pregnant individuals in Ontario.

Enhanced First Trimester Screening (eFTS)
Occurs in the first trimester of pregnancy (about 3 months pregnant) includes the **nuchal translucency (NT) ultrasound** and a **blood test**. This screening can indicate the chance for having a baby with Trisomy 21 (Down Syndrome) and Trisomy 18 (Edwards Syndrome).

Second Trimester Screening (STS) occurs when you did not have access to eFTS. This occurs in your second trimester (about 5 months pregnant). This involves a **blood test** which screens for Trisomy 21 (Down Syndrome), Trisomy 18 (Edwards Syndrome) and neural tube defects.

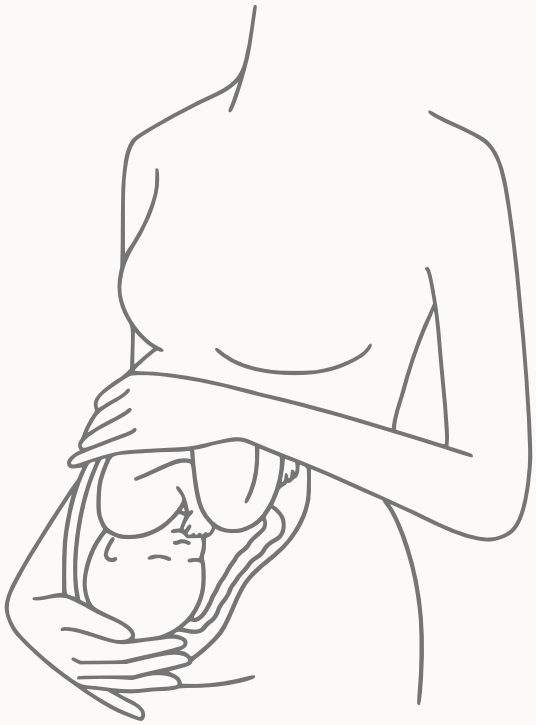
Non-Invasive Prenatal Testing: this is the most accurate prenatal testing and can occur after 9-10 weeks of pregnancy. The screening is funded by the Ministry of Health in certain situations and if funding is not approved this test must be paid for out-of-pocket. This screening involves a **blood test** which screens for Trisomy 21, 18, 13 and sex chromosome differences.
Some NIPT labs are able to screen for other genetic conditions, such as microdeletions. However, it is important to keep in mind that the accuracy of these results is difficult to determine due to limited evidence and the rare nature of the conditions.
This testing cannot be done when there are more than 2 babies or if one twin was lost during pregnancy ("vanishing" twin) as genetic material from the miscarried twin can be present in the blood and can affect the results.

Diagnostic Tests
This testing is more invasive and occurs when there is an increased risk that the baby may be born with a genetic or chromosomal difference.

Chronic Villus Sampling (CVS): This is a prenatal test that involves sampling tissue from the placenta to test for differences in chromosomes and genetic problems. The placenta is a structure in the uterus that provides nutrients and blood to the baby. This procedure takes place between week 10 & 12 of pregnancy. This test does not test for neural tube defects. This procedure can occur in 2 ways:
Transcervical: Where a catheter is inserted through the cervix into the placenta to obtain the tissue sample
Transabdominal: Where a needle is inserted through the abdomen and uterus into the placenta to obtain the tissue sample

Amniocentesis: This is a procedure where a small amount of amniotic fluid is take to test for genetic disorders and open neural tube defects.
This testing is completed between 15-20 weeks of pregnancy, when it is indicated that the baby may be at risk for birth defects.
The risks of the procedure include: cramping, bleeding from injection site or vagina, miscarriage or preterm labor. *However, miscarriage occurs in in less than 1% of individuals after having this procedure which is only slightly higher than the normal risk of miscarriage during pregnancy.

What Conditions are Screened for?



Trisomy 21 (Down Syndrome)

Trisomy 21 is a condition resulting in an extra copy of chromosome 21. This alters a person's development, it affects each person with Down syndrome's development differently. There is a greater chance that people with this condition have problems with their heart, stomach, bowel, thyroid, vision and hearing. However, treatment for these conditions are available and many people with Down syndrome live long, healthy and productive lives.

Trisomy 21 occurs in about 1 in 800 babies. Although this condition can occur at any age of the pregnant individual, the chances increase with age.

Trisomy 21 is detected in eFTS, STS & NIPT.

Trisomy 18 (Edwards Syndrome)

Caused by an extra copy of chromosome 18, babies with this condition have 3 copies of this chromosome rather than the typical 2. Babies with this condition often do not make it to term or result in miscarraige or stillbirth. When babies with this condition survive they often live with severe intellectual disability and health challenges, often living shorter lives as many babies do not live past their first year of life, and require a high amount of caregiving.

Trisomy 18 occurs in about 1 in 6000-8000 of babies born. Although the condition can occur at any age of the pregnant individual, the chances increase with age. This condition occurs more frequently than this, however, many of these babies do not make it to birth.

Trisomy 18 can be detected through all prenatal screening tests, ultrasound (through detection of anatomical differences common in babies with this condition) and diagnostic testing (diagnostic testing occurs when a previous test shows a positive or high risk result).

Open Neural Tube Defects

This condition occurs when the brain or spinal cord does not form properly.
When involving the spinal cord this condition is called **Spina bifida**. Spina Bifida causes physical disability can cause one to have difficulty walking and controlling their bladder and bowel, in addition people may also have intellectual disabilities as well. Treatment is available for the physical disabilities associated with this condition. When the neural tube defect involves the brain this is called **anencephaly**. A baby with anencephaly will be born still born or will die shortly after birth.

Neural Tube Defects occur in approximately 1 in 2000 babies. The chances of this condition occurring is not influenced by the pregnant individuals age.

Trisomy 13

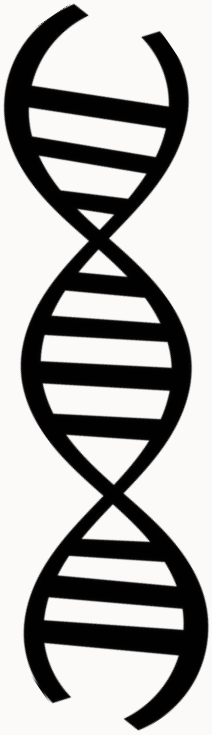
Trisomy 13 is caused by an extra copy of chromosome 13, leading to babies with this condition having 3 copies of this chromosome instead of 2. This condition disrupts typical development and results in severe intellectual disability and health challenges and requires a high amount of caregiving. Most pregnancies do not make it to birth and result in miscarriages or stillbirth.

Trisomy 13 occurs in about 1 in 6000-29000 babies born. Although the chances of a baby being born with this condition can occur at any age of the pregnant individual, the chances increase with age. This condition occurs more frequently than this, although, many of these babies do not make it to birth.

This condition can be detected through prenatal screening (NIPT) and a chromosome difference can be predicted due to anatomical differences through the NT ultrasound during eFTS. However, it is not directly screened for in eFTS or STS through the blood test. It is also detected in diagnostic testing.

Sex Chromosome Differences

Sex chromosomes are the the 23rd pair of chromosomes which differ based on sex. Males typically have one X and one Y chromosome and females typically have two X chromosomes. Sometimes people are born with an extra or missing sex chromosome(s). Some people with this condition do not experience any developmental challenges or health concerns, where others may have differences in height, issues during puberty, heart or other organ problems and/or differences in development, such as speech or learning difficulties. Implications on health are particularly impacted by Turner syndrome.



Turner Syndrome (45,X) is a chromosome difference that occurs in females where they have only one X chromosome. It occurs in about 1 in 2500 babies and in 1-1.5% of pregnancies as many babies with this condition do not make it to birth. Development can affect each baby differently. Some common issues include heart defects, short stature, delayed puberty, infertility and learning disabilities.

Sex chromosomes differences happens in about 1 in 500-1000 babies however it may be more common as some people may have a sex chromosome difference and not be aware of it.

Sex chromosome differences can be detected during pregnancy through NIPT and diagnostic testing.

What Happens After the Screening?

If the results show a **positive screen** this does not mean that your baby has a health condition for sure. Your health care provider will discuss the results with you and more testing can be performed. You may also be referred to a genetic counselor.

Most often the results show a **negative screen**. This means that your chances of having a baby with a health condition that is tested for is very low. This does not mean your chances are 0%. Keep in mind, eFTS is accurate 72% of the time and NIPT is accurate 99% of the time.

Trying to Decide if Prenatal Screening is Right for You?

Many people have difficulty deciding whether or not to have prenatal screening. It can be helpful to ask yourself some questions to help you come to the best decision for you and your pregnancy. Prenatal screening is a personal decision, no one can tell you if prenatal screening is right for you, as there is no right answer.

Some questions you may ask yourself are;

- How important is it for you to know the chance that your baby may have a chromosome difference that may affect their health and development?
- Would knowing about a chromosome difference be helpful for you to know before your baby's birth?
- What are your thoughts around ending your pregnancy if your baby has a chromosome difference?
- How would knowing versus not knowing affect you emotionally throughout your pregnancy?

Some people would prefer to wait until their baby is born and others would like to know ahead of time to prepare for a child that may need special care.
In addition, your health care provider may be able to recommend changes to how your pregnancy is looked after or change your birth plan.