GENETIC SCREENING

Midwifery Group of Ottawa

Genetic prenatal screening is a way to determine the degree of chance your baby may or may not have certain genetic conditions called trisomies (like Down Syndrome). Anyone can have a pregnancy with a trisomy regardless of their family history. This chance increases with age. The screening has no risks for your baby. The goal is to inform you of your likelihood of having an affected baby. Screening is not the same as diagnostic testing which can tell you whether or not your baby actually has a condition.

In a low-risk pregnancy, the **enhanced first trimester screen (eFTS)** is often the first step. It can tell you your risk of having a baby with Trisomy 21 (Down Syndrome) or 18 (Edwards Syndrome). It involves an ultrasound and blood work done in early pregnancy. The results take a few weeks. When a result is negative, the risk of having an affected baby is considered low. Where a person screens 'positive', or at higher risk, follow-up tests are offered. This screen is covered by OHIP.

When people are too late for eFTS, but are interested in genetic screening, people can do **maternal serum screening (MSS)**. This is also a screening tool, and not diagnostic.

For people 40 years old or more at their estimated due date, or for people who meet certain criteria, the Ministry of Health funds a highly effective screen called **non-invasive prenatal screen (NIPT)**. Only blood work is required, it can be done as early as 9 weeks of pregnancy, and it screens for Trisomy 21, 18, 13 (Patau Syndrome) and sex chromosome differences. Anyone who does not meet the funding criteria can still do NIPT by opting to pay out-of-pocket. A doctor must fill in the requisition for this test as midwives are not able to order it.

Pursuing genetic screening or testing is a personal choice. Some parents do not feel this information would make a difference in their choices regarding continuing a pregnancy. Your decision will not affect how you are treated by your midwife.

For more detailed information about prenatal genetic screening, please go to **prenatalscreeningontario.ca**.

Enhanced First Trimester Screen (eFTS)

If you choose eFTS, you should schedule a nuchal translucency (NT) **ultrasound and a blood test** between 11+2 and 13+3 weeks of pregnancy. The blood test must be done after the NT ultrasound, but it is NOT required that it be done on the same day. The blood test can be performed anytime up to 13+3 weeks.

The NT measurement, your age, race, weight, and a few other variables, along with the measurement of different protein levels in your blood will be used to calculate your chance of having a baby with trisomy 21 (Down syndrome) or trisomy 18.

You must have a requisition from your care provider, either in hand or faxed directly to the ultrasound clinic/laboratory in order for them to do the test. The results are available within one to two weeks.