

**GENETIC SCREENING**

Your midwife will offer you testing in early pregnancy designed to screen for genetic disorders, specifically Down syndrome (also called Trisomy 21), Trisomy 18 and Open Neural Tube Defects.

When you have your first clinical visit with your midwife, you can determine if you have any risk factors that increase the chance of having a child with any of these disorders. If you do have any risk factors or are over age 35, you will be offered genetic counseling. Some of the factors that increase the chance of having a baby with the disorders are; family history of genetic or inherited disorders, environmental exposure to toxins, previous child with chromosomal disorders or anomalies, ethnicity or recurrent miscarriages. In every pregnancy, there is a base-line risk of having a baby with a condition not detected by prenatal testing of 2-3%.

These tests are screen tests only and do not give a definite result, rather they will provide you with an odds ratio for having a baby with these conditions.

This means that you will be provided with either a “screen positive” (higher than expected chance the fetus may have a condition), or a “screen negative” (lower than expected chance the fetus may have a condition).

It is important to understand that if your test results show a “screen positive” then you will be offered more definite testing, like amniocentesis or Chorionic Villus Sampling (CVS). Both of these tests increase the risk of miscarriage. You will be offered genetic counseling before you make a decision about proceeding with these tests.

*False Positive*: results of the tests show a positive screen but after further testing or at delivery it is determined that the baby does **not** have a disorder

Down syndrome: All people with Down syndrome have mental disabilities. They are also more likely to have physical disabilities. Each person born with Down syndrome has a different degree of mental and physical disability and there is no way of telling that before the delivery. There is no cure for Down syndrome but there are many support groups available. There is a greater chance of having a baby with Down syndrome as maternal age increases. Without any risk factors, someone age 20 has a 1:1650 chance of having a baby with Down syndrome, someone age 35, 1:375.

Trisomy 18: Babies with trisomy 18 have serious mental disabilities and many physical disabilities. Many pregnancies with trisomy 18 will miscarry. Most babies born with trisomy 18 do not survive the first few months of life. The chance of having a baby with Trisomy 18 increases with maternal age.

Open Neural Tube Defects (ONTD): When the brain or spinal cord of the fetus does not form properly ONTD can occur. An open neural tube defect involving the spinal cord is called spina bifida. Spina Bifida causes physical disabilities such as having difficulty walking or controlling the bladder and bowel. These babies may have mental disabilities. There is no cure but there is help for many of the physical disabilities. An open neural tube defect involving the brain is called anencephaly. A baby with anencephaly will die before or shortly after birth. In Ontario ONTD affects about 1:2000 pregnancies and age is not a factor in its occurrence.

You may want to consider the following before deciding whether to take the testing.

* Do you want to know the chance of your having a baby with Down syndrome, Trisomy 18 or ONTD?
* Would you have further, riskier testing if your test result was “screen positive”?
* What will you do if further testing shows that you have an affected fetus?
* How will the test results make you feel about your pregnancy?
* How would your life be affected if you had a child with mental or physical disabilities?

The following chart illustrates the testing available to you. The standard of care in this community is to offer integrated prenatal screening. If your pregnancy is farther along than the recommended gestational age, the other tests will be offered.

Advantages to testing: results may be reassuring

Disadvantages to testing: waiting for results can be stressful, results may cause worry and tests do not detect every kind of condition

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| **TEST NAME** | **WHEN** | **DETECTION RATE for Down syndrome** | **FALSE POSITIVE RATE** |
| Integrated Prenatal Screening | At 11-13 weeks   * blood test * ultrasound\*   At 15-20 weeks   * 2nd blood test | 85-90%  no result until after second test | 2-4% |
| Serum Integrated Prenatal Screening  (SIPS) | At 11-13 weeks   * blood test   At 15-20 weeks   * second blood test | 80-90%  no result until after second test | 2-7% |
| First Trimester Screening | At 11-13 weeks   * blood test * ultrasound | 80-85%  earlier result | 3-9% |
| Quadruple Screening | At 15-20 weeks   * blood test | 75-80% | 5-7% |
| Triple Maternal Serum Screening | At 15-20 weeks   * blood test | 71% | 7% |

These tests are screening for Down syndrome, Trisomy 18 and Open neural Tube Defects

\*ultrasound to detect Nuchal Translucency; the thickness of the fluid filled space at the back of the baby’s neck