

INTEGRATED PRENATAL SCREENING (IPS)

NORTHWESTERN
ONTARIO REGIONAL
GENETICS
PROGRAM



If you are pregnant, you can choose to have prenatal screening to find out your chances of having a baby with specific problems. In Ontario, pregnant women can have Maternal Serum Screening (MSS) to see if they have a lower or higher chance of having a baby with *Down syndrome, open neural tube defects or trisomy 18*. A newer prenatal screening test called Integrated Prenatal Screening (IPS) has been developed. This looks for the same problems as MSS but it is a better screen for Down syndrome. You should discuss screening options with your healthcare provider early on in your pregnancy (preferably by the 10th week of your pregnancy) before deciding whether you wish to be screened and which test you would prefer.

WHAT PROBLEMS ARE BEING SCREENED?

DOWN SYNDROME

People with Down syndrome have physical and mental disabilities. They are more likely to have medical problems such as heart problems. Each person with Down syndrome is different. There is no way to predict how serious any of the disabilities will be. There is no cure for Down syndrome. Any pregnant woman can have a baby with Down syndrome. However, the chance increases as the woman's age increases.

OPEN NEURAL TUBE DEFECTS

Open neural tube defects happen when the brain or spine does not develop properly. There are two kinds of open neural tube defects: spina bifida and anencephaly.

Spina bifida is an opening in the bones around the spinal cord. Usually the opening is not covered by skin. Spina bifida causes physical problems and sometimes mental disabilities. There is no way to predict how serious these physical problems will be or if the person will have mental disabilities. Spina bifida can usually be repaired, but there is no cure for the damage that occurs while the baby is in the womb.

When a baby has anencephaly, the brain and skull do not develop completely. A baby with anencephaly will die shortly after birth.

Between one and two babies out of 1,000 babies are born with a neural tube defect. The chance does not depend on the age of the mother. There are some maternal conditions, such as diabetes or a family history of neural tube defects, which may affect your risks for neural tube defects. We encourage you to discuss this with your healthcare provider.

TRISOMY 18

Trisomy 18 causes very serious physical and mental disabilities. Most infants with trisomy 18 die by one year of age. There is no cure for trisomy 18. Trisomy 18 is a very rare problem. The chance of trisomy 18 increases with a woman's age.

HOW DOES IPS WORK?

IPS is performed in two parts. The first part is done between 11 and 13 weeks + 6 days of pregnancy and involves an ultrasound and a blood sample. The second part is done between 15 and 18 weeks + 6 days

and involves a blood sample only.

The IPS result is calculated using both sets of information, and estimates your chance of having a baby with Down syndrome, open neural tube defects and trisomy 18.

WHY IS INTEGRATED PRENATAL SCREENING (IPS) BETTER THAN MATERNAL SERUM SCREENING (MSS)?

IPS will detect more pregnancies with Down syndrome than MSS. The biggest concern with MSS is that 8 out of 100 women who have MSS are called "screen positive". However, the chance that their baby actually has Down syndrome is small. With IPS only 3 out of 100 women are called "screen positive" for Down syndrome.

With MSS and IPS, 2 out of 100 women are called "screen positive" for open neural tube defects.

Very few women are called "screen positive" for trisomy 18.

WHAT DOES IPS INVOLVE?

Part 1: An ultrasound is done between 11 weeks and 13 weeks 6 days of pregnancy. This is done to date the pregnancy and to measure the nuchal translucency. The nuchal translucency (NT) is a measurement of the thickness of the skin at the back of the neck of the developing baby. This measurement must be performed by a certified ultrasound provider. We do offer this service in Thunder Bay.

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Following the ultrasound, a sample of your blood is taken to measure the level of a protein called PAPP-A.

Part 2: The second blood sample is taken between 15 and 18 weeks + 6 days of pregnancy. The levels of alpha-fetoprotein (AFP), estriol (uE3) and chorionic gonadotrophin (hCG) are measured in this sample.

Result: The levels of PAPP-A, AFP, uE3 and hCG in your blood are used, together with the nuchal translucency measurement and your age to determine your chance of having a baby with Down syndrome. The level of AFP is used to see if there is an increased risk of an open neural tube defect. The levels of AFP, estriol and hCG are used to see if there is an increased risk for trisomy 18. It is only after the second blood test that a report will be sent to your healthcare provider. All samples are sent and tested at North York General Hospital Laboratories in Toronto.

WHAT DOES A SCREEN POSITIVE RESULT MEAN?

A screen positive result means that the *chance* is higher than usual that your baby might have Down syndrome, an open neural tube defect or trisomy 18. **Most women with a screen positive result have healthy babies.** Only further tests can show if a baby has one of these problems.

Making a decision about whether to have further testing can be difficult. Speak to your healthcare provider about further testing. For additional information you can ask for a referral for genetic counselling.

WHAT ARE THE TESTING OPTIONS?

If IPS shows a “screen positive” for **Down syndrome** or **trisomy 18**, you will be offered amniocentesis. During amniocentesis, a small amount of fluid surrounding the baby is taken and tested for Down syndrome or trisomy 18. Amniocentesis can cause a miscarriage in about one out of every 200 women who have the test.

If IPS shows a “screen positive” for an **open neural tube defect**, you will be offered another ultrasound where the head and the back of the baby will be examined. Occasionally, an amniocentesis is offered and the fluid is tested for open neural tube defects.

Most times the results of the ultrasound or amniocentesis will show that your baby **does not** have Down syndrome, an open neural tube defect or trisomy 18, but occasionally a prenatal screening test may lead to finding another type of problem for your baby.

Other tests may be suggested after further discussion with your healthcare provider or genetic nurse.

WHAT DOES “SCREEN NEGATIVE” RESULT MEAN?

If the levels of the protein in the blood and the nuchal translucency are in the usual range, then the result is called “screen negative” and further testing is not offered. About 95 women out of 100 will have a screen negative result. Although most women with a screen negative result will have normal babies, IPS can miss a case of Down syndrome, an open neural tube defect or trisomy 18.

WHAT HAPPENS IF I DO NOT HAVE THE SECOND BLOOD TEST?

If you do not have the second blood test, North York General Hospital Laboratories will not be able to report a result for your IPS screening. You will receive a result based on the first blood test and the ultrasound alone.

This is a **less effective** screen for Down syndrome. Also, it will not detect open neural tube defects or trisomy 18 so it is important that both parts of the screen are completed to receive a more thorough result.

It is important to be aware that every pregnant couple in the general population has a 2-3% risk of having a baby with a birth defect. While normal results of prenatal testing are reassuring, they do not guarantee the birth of a healthy baby.