

## Is family history a factor?

No. In fact, most of these conditions occur in families with no history of babies born with these disorders.

## What does the blood test measure?

The blood test looks at a number of natural substances in the mother's blood that are made by the baby and the placenta. (The placenta is the organ in the uterus that nourishes and preserves the fetus through the umbilical cord). These substances are seen in all pregnant women.

By taking a sample of your blood, the level of each of these substances can be measured.

If the levels are what is expected, your test is called "screen negative".

If the levels are higher or lower than expected, or occur in certain patterns, your test is called "screen positive". A screen positive result does not mean your baby has one of these conditions. It just means you are eligible for more testing.

Most pregnant women who get a "screen positive" result go on to have a healthy baby. A screen positive result may be due to confusion about how far along you are in the pregnancy. Sometimes confirming how far along you are is the first step after a screen positive result.

## Whom do I see if I get a screen positive result?

Following a screen positive test result, your healthcare provider might speak to you about having an ultrasound, or meeting with a genetic counsellor.

Genetic counsellors are specially trained healthcare professionals. They can help you fully understand your results, talk with you about further testing options and help you decide if more testing is right for you.

## What further testing options will be offered?

The first step is to check the baby's age with ultrasound, if this has not already been done. A correct estimate of how far along you are in the pregnancy is needed to get an accurate prenatal screening result.

The follow-up testing options will depend on the condition that received a screen positive result. The tests may include ultrasound, diagnostic testing like amniocentesis (which carries a small increased risk of miscarriage), non-invasive prenatal screening (NIPS), or possibly further blood tests.

**NOTE:** The additional tests most often will find that your baby does not have Down syndrome, trisomy 18, or a neural tube defect.

## What if my baby really does have a problem?

If that is the case, your genetic counsellor will explain the diagnosis to you and talk with you about your choices. These choices may include continuing the pregnancy, adoption, or having a termination of pregnancy (abortion).

Making a decision is not easy and you will be provided with lots of support and resources. Unfortunately, many of the conditions these tests screen for do not have a cure.



# Manitoba Prenatal Genetic Screening Program

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## What is prenatal genetic screening?

Prenatal genetic screening is a blood test (sometimes combined with a special ultrasound) available to all pregnant women in Manitoba as part of their prenatal care. The test can tell you about your risk of having a baby with certain conditions such as Down syndrome or spina bifida. It cannot tell you for sure if your baby has one of the conditions being screened for – it only tells you what the risks are.

## Is prenatal genetic screening optional?

Yes. The test will only be done if you want it. Some people find having this information is reassuring while others do not want it. Your decision about having this test will not affect the care you receive during your pregnancy.

## What does the prenatal genetic screening test for?

It screens for chromosome conditions, and neural tube defects.

### Chromosomal conditions

Chromosomes are the carriers of genetic information. Two common chromosomal conditions are:

#### Down syndrome (trisomy 21)

This is a common cause of intellectual (learning) disabilities. Children with Down syndrome have a unique facial appearance and may have certain birth defects, including heart problems. This condition occurs in about 1 in 700 babies.

#### Trisomy 18

Babies with trisomy 18 have severe mental and physical disabilities. Many pregnancies with trisomy 18 will miscarry. Most babies born with trisomy 18 do not survive past the first few months of life. This condition occurs in about 1 in 4,000 babies.

#### Neural tube defects

These are birth defects where the spinal cord or brain fails to form properly. This condition occurs in about 1 in 1,000 babies, and the mother's age is not a major risk factor.

#### Spina bifida

Spina bifida is a birth defect involving the spinal cord. This condition can cause problems with bladder and bowel control, or difficulties with walking, due to abnormal spinal cord development.

#### Anencephaly

Anencephaly is a defect involving the brain. A baby with anencephaly will be stillborn or die shortly after birth.

## What if I test positive (increased risk) for any of these conditions?

Specific diagnostic testing options will be offered to women whose prenatal genetic screening shows an increased risk for any of these conditions.

## How is the genetic screening done?

Your healthcare provider will give you a requisition for a blood sample to be taken from your arm between 15 and 20 weeks of pregnancy. The ideal time is around 16 weeks of pregnancy.

In some instances, a special ultrasound, called a nuchal translucency (NT) ultrasound, will also be completed between approximately 11 and 13 weeks 6 days of pregnancy.

Nuchal translucency (NT) is a special measurement of a thin layer of fluid that is normally found under the skin behind the baby's neck. The measurement of the NT will be combined with your blood test result. Not all women are eligible (suitable) for this measurement. Your healthcare provider can tell you if you are eligible.

**NOTE:** Having an accurate due date, and performing the test at the right time, is important to ensure the test results are read correctly.

## How will I get my result?

Approximately one week after your blood is drawn, your results will be sent to your healthcare provider. They will explain the results to you and arrange any follow-up appointments, if necessary.

## Where can I get more information?

Manitoba Prenatal Genetic Screening Program  
Health Sciences Centre Winnipeg  
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