

# PRENATAL SCREENING



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It is your choice whether or not to have prenatal screening.

## What is prenatal screening?

All pregnant women have a very small chance of having a baby with a *chromosomal anomaly* or a *neural tube defect*. A mother can have tests done while she is pregnant to assess the risk that her baby has one of these conditions. It is important to remember that most babies are born healthy.

**What is a chromosomal anomaly?** Every cell in your body has genetic material (DNA). Chromosomes are the structures that hold your DNA. Some people are born with too few or too many chromosomes in each cell, or there is something wrong with one of their chromosomes. The most common anomaly is Down syndrome, when a person has an extra copy of the 21<sup>st</sup> chromosome; people with Down syndrome usually have mild to moderate intellectual delay and are more likely to have some health problems. The second most common anomaly is an extra copy of chromosome 18; babies with 'trisomy 18' have serious mental and physical disabilities and usually do not survive very long.



**What is a neural tube defect?** This is a problem with the brain or spinal cord that a baby develops very early in pregnancy. An example is spina bifida, when the bones protecting the spinal cord are not fully formed. Spina bifida causes physical disabilities and sometimes mental disabilities.

## How does prenatal screening work?

If you choose to have prenatal screening, data from your ultrasounds and samples of your blood will be tested for signs of certain conditions.

The results are not a diagnosis. They tell you a likelihood that your baby has a condition. For example, possible results might be a *1-in-100* or a *1-in-400* chance that your baby has Down syndrome. Your doctor, nurse or midwife will discuss the results with you.

## What do the results mean?

A 'negative' screen means that your risk is not over a certain threshold: it is unlikely that your baby has one of the conditions that you were tested for. More diagnostic testing is not recommended.

A 'positive' screen means that your risk is over a certain threshold – there is a chance (usually small) that your baby has one of the conditions you were tested for. More diagnostic testing may be offered to you. These additional tests, such as *amniocentesis*, can give you a better idea of whether your baby has one of these conditions. You and your health-care provider can use this information to make decisions about your pregnancy and help you prepare for the birth of your child.

However, these additional tests come with risks, such as a 1 in 200 chance of miscarriage. Not all women choose to have additional tests; the choice is yours.

Prenatal screening tells you the chance that your baby *might* have a condition. It does not tell you for sure. Most women who have a 'positive' screen result will not have a child with a condition. Only further diagnostic testing can tell you for sure. For example, a 1 in 100 risk means a 99 in 100 chance of having a baby without the condition.

## Is all prenatal screening the same?

No. There are different types of prenatal screening. Which type you are offered will depend on how far into your pregnancy you are when you first see your caregiver. The earlier you see your caregiver, the more options you will have. It will also depend on your age, your medical history, and if you are carrying more than one baby.

If you choose to have prenatal screening, you will have at least one of the following tests:

- A nuchal translucency measurement taken during an ultrasound between 11 and 13 weeks
- A blood sample taken between 10 and 13 weeks of pregnancy
- A blood sample taken between 15 and 20 weeks of pregnancy

## What are common types of prenatal screening?

Each province in Canada offers different types of prenatal screening tests. Talk to your doctor, nurse or midwife about which prenatal screening options are available and which one is right for you.

If you live in a rural or remote area, you may have to travel to get a nuchal translucency measurement.

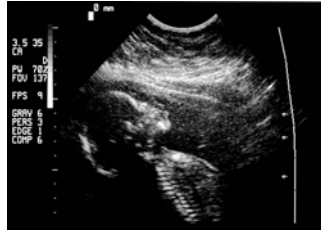
**First trimester combined screening (FTS):** Requires a nuchal translucency measurement and a blood sample taken between 10 and 13 weeks of pregnancy. This type of screening can identify your baby's risk for a chromosomal anomaly.

**Second trimester quad screen:** Requires a blood sample taken between 15 and 20 weeks of pregnancy. This can identify your baby's risk for a chromosomal anomaly or neural tube defect.

**Serum integrated prenatal screen (SIPS):** Requires two blood tests, taken between 10 and 13 weeks and 15 and 20 weeks of pregnancy, to identify your baby's risk for a chromosomal anomaly or a neural tube defect.

**Integrated prenatal screen (IPS):** Requires a nuchal translucency measurement as well as first-trimester and second-trimester blood tests to identify the risk of a chromosomal anomaly or neural tube defect.

**Routine ultrasounds:** Even if you choose *not* to have prenatal screening, ultrasounds are a normal and important part of pregnancy. For example, they are used to confirm your due date, see if your baby is growing well, and find out if you are carrying more than one child. Sometimes, a physical abnormality – such as a cleft lip or a heart defect – can also be seen during an ultrasound. Many women have at least two ultrasounds: a dating ultrasound in the first trimester (often between 11 and 13 weeks of pregnancy), and another done between 18 and 22 weeks of pregnancy.



## What is nuchal translucency?

This is a measurement of the thickness of tissue in your baby's neck. This measurement is usually taken during an ultrasound between 11 and 13 weeks.

## Do I have to get prenatal screening?

Prenatal screening is always *your* choice. You might want to consider your answers to the following questions:

- If you had a 'positive' screen, would you want to have further testing done?
- Would you want to know if your baby had a chromosomal anomaly or neural tube defect?
- If the diagnostic testing showed that your baby had a serious condition, would knowing help you make decisions?
  - Would you continue the pregnancy? Would you end the pregnancy? Would you give the baby up for adoption?
  - Would you want this information to help you prepare for the birth of a baby who might need special care?
- If your screening results are positive, how will this information affect your feelings throughout your pregnancy? Would it cause you too much worry?

Your health-care provider will be there to support you in making decisions.

## Does my age affect my baby's risk?

Some problems such as chromosomal anomalies become more likely the older a pregnant woman is. Maternal age is one factor used to calculate your baby's risk, and a screening result is more likely to be positive with increasing maternal age.

## Further resources

- Available at [www.sogc.org](http://www.sogc.org) from the Society of Obstetricians and Gynaecologists of Canada:
  - The committee opinion, *Counselling Considerations for Prenatal Genetic Screening*
  - The book, *Healthy Beginnings: Giving your baby the best start from preconception to birth*
  - The brochure, *Age and fertility*

For more information on conditions:

- March of Dimes at [www.marchofdimes.ca](http://www.marchofdimes.ca)
- The Canadian Down Syndrome Society at [www.cdss.ca](http://www.cdss.ca)