Prenatal Genetic Screening

Making decisions

This pamphlet explains common types of prenatal genetic screening available in Ontario. It does not replace talking to your health-care practitioner about what screening means and making decisions that are right for you.

PRENATAL SCREENING
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Key facts

As you read this pamphlet, it's important to understand prenatal genetic screening can only let you know if the baby's chance of having certain genetic conditions is higher or lower, and you can then consider further tests that will say for sure. Prenatal genetic screening does not tell you if your baby definitely has a genetic condition.

- Prenatal genetic screening tests focus on the chance of your baby having trisomy 18 (Edwards syndrome) or trisomy 21 (Down syndrome), two genetic conditions that cause intellectual disability and health challenges.
- Being screened presents no risk to the pregnancy because it's done by ultrasound and blood work.
- Prenatal genetic screening is available to all pregnant people in Ontario; it's your choice whether you have it or not.
- Any person can have a baby with trisomy 18 or trisomy 21 (even if no one else in their family has). The chance of it happening increases as the pregnant person gets older.

What are trisomy 18 and trisomy 21?

Trisomy 18 and trisomy 21 are genetic variations that usually happen in the egg or the sperm before the baby is conceived. In most cases of trisomy 18, a baby has three copies of chromosome 18 instead of two. In trisomy 21, the extra copy is of chromosome 21.

Trisomy 18 (Edwards syndrome)

Trisomy 18 affects how different parts of the body develop and many babies with trisomy 18 die before birth; most that are born die within a year. Those born alive usually have severe health and intellectual problems. A small number of babies with trisomy 18 live past their first year of life and need a lot of care.

Trisomy 21 (Down syndrome)

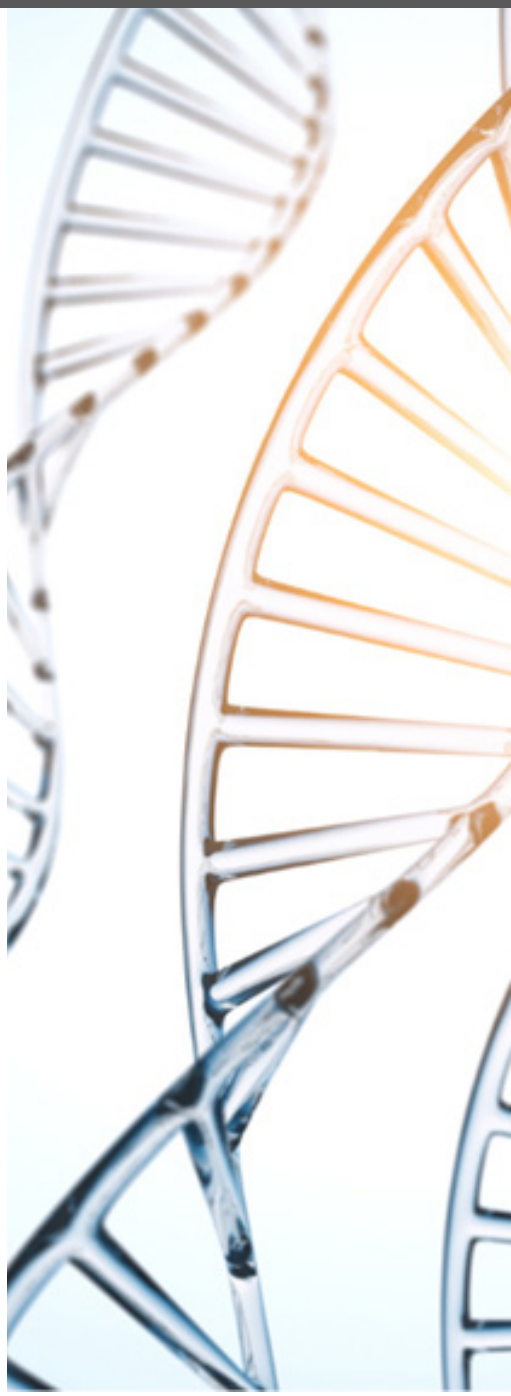
Everyone with trisomy 21 is unique and has their own strengths and challenges, but they all have some degree of intellectual disability, which means most will take longer to learn. As they get older, people with trisomy 21 may need support doing personal care and daily tasks. The level of support needed varies from person to person.

People with trisomy 21 are more likely to have certain health issues, including:

- heart defects
- eyesight and hearing problems
- problems with feeding and digestion
- sleep issues
- Alzheimer's disease later in life

Some people with trisomy 21 do not have any of these issues, while others have several. There is treatment and support to help people with trisomy 21, but there is no cure.

People with trisomy 21 usually live into their 60s and most are living more independently now than ever before. Many adults with trisomy 21 lead fulfilling lives and can participate in school, work and community life.



Is prenatal genetic screening right for me?

Deciding whether to have prenatal genetic screening is up to you and there is no right or wrong choice. Thinking about these questions might help you make a decision.

Do you want to know if there is an increased chance of your baby having a genetic condition that could affect their health and development?

Many parents, but not all, want to know the chance of the baby having trisomy 18 or 21. Some want to know if there's a higher chance of the baby having a genetic condition so that they can decide what to do next, while others hope for the reassurance of being told there's a low chance. Some are worried they'll get a false positive, where the screening says the baby has an increased chance of a genetic condition but in fact they don't. That happens more often with two of the screens described here (enhanced First Trimester Screening and Second Trimester Screening).

Would you have further diagnostic testing if your screening results show there is a higher chance for trisomy 18 or 21?

There's a small risk the tests that diagnose trisomy 18 and 21 (chorionic villus sampling and amniocentesis) will cause miscarriage, but they're the only way to be sure if the baby has either condition.

What are your thoughts about continuing or ending a pregnancy with trisomy 18 or 21?

Your decision needs to be right for you, depending on your life situation, values and beliefs. Speaking with health-care practitioners, family, friends or counsellors and learning about further tests might help you make your choice.

Would knowing about one of these genetic conditions help you prepare for it?

Some people who continue their pregnancy find it helpful to have the extra time to learn more about trisomy 18 or 21, and what local resources are available or perhaps to make an adoption plan for the baby.

What could the result mean for your pregnancy care?

Prenatal genetic screening can lead to more prenatal care and monitoring. Even if you don't have a follow-up diagnostic test but there are signs of a genetic condition or other concerns, your care may change.

What are my options?

If you decide to have prenatal genetic screening for trisomy 18 and 21, your health-care practitioner can help organize it for you.

Available screening tests

01 Enhanced First Trimester Screening (eFTS)

This screen is paid for by the Ontario Health Insurance Plan (OHIP) from 11 weeks and 2 days to 13 weeks and 3 days of pregnancy. It involves a special ultrasound called the 11-14 week (nuchal translucency) ultrasound and a blood test and finds most pregnancies with trisomy 18 and 21, but not all.

02 Second Trimester Screening (STS)

Also paid for by OHIP, STS is an option if eFTS is not available. STS is done by a blood test from 14 weeks to 20 weeks and 6 days of pregnancy. Like eFTS, it finds most pregnancies with trisomy 18 and 21, but not all.

03 Non-Invasive Prenatal Testing (NIPT)

Also a blood test, NIPT can be done any time after 9 or 10 weeks of pregnancy, depending on the laboratory. It can find more pregnancies with trisomy 18 and 21 than the above screens. NIPT also screens for another genetic condition called trisomy 13.

NIPT is OHIP-funded if you meet one of the funding criteria on our website, which include:

- You will be age 40 or older at the time of birth.
- You had a past pregnancy with trisomy 18, 21, or 13.
- You are pregnant with twins.

If you do not meet the criteria but still want NIPT, you can pay for it yourself. Your health-care practitioner will still need to organize it for you.

If you choose NIPT, you don't need enhanced first or second trimester screening, but it's still recommended you have a 11-14 week ultrasound (without the blood test) for more information about the health of the baby.

What do my results mean?

Enhanced First Trimester Screening (eFTS) and Second Trimester Screening (STS) Results

The results will be sent to your health-care practitioner about 5 business days after your blood test. The results will tell you if there is a higher chance ("screen positive") or lower chance ("screen negative") of the baby having trisomy 18 or 21.

Screen negative

Most people get a negative result, showing the baby has a lower chance of having trisomy 18 or 21. A negative result does not mean there is no chance that the baby has one of these conditions, just that it is unlikely. The way your health-care practitioner looks after your pregnancy will probably not change.

Screen positive

A positive result means there's a higher chance of the baby having trisomy 18 or 21, not that they will for sure. In fact, most people with this result do not actually have a baby with one of these conditions. To be more certain, you might have another screening test, the more accurate NIPT, which is paid for by OHIP if either eFTS or STS have come back positive.

Another option is to have diagnostic testing, either chorionic villus sampling (CVS) which tests a sample of placental tissue, or amniocentesis, which tests amniotic fluid. Either will show for certain if your baby has a chromosome change (but either brings a small risk of miscarriage). You can also choose to not have any more testing.

Non-Invasive Prenatal Testing (NIPT) Results

It takes about 10 business days to get NIPT results, which are reported as “high risk” or “low risk”. Like the other two screens, the results cannot say for certain whether the baby has a genetic condition, but it is more accurate. Most people who get a “high risk” result are carrying a baby with trisomy 18, 21, or 13, but you would still need CVS or amniocentesis to know for sure. You may want to talk to a genetics specialist to discuss your “high risk” result and options for more testing.

No result

Sometimes, NIPT fails to give a result. This rarely happens with the other two screens. When the NIPT fails, this does not mean the baby has a genetic condition. You may have the option to repeat the NIPT. Other options include having one of the other screens, ultrasounds or diagnostic testing (CVS or amniocentesis).

Note: the terms “screen positive”, “screen negative”, “high risk” and “low risk” are the ones used in reports from Ontario laboratories.

Have more questions? We are here to help.

Visit our website to read more about prenatal screening.

Contact our certified genetic counsellors by phone or email.

Contact information

PrenatalScreeningOntario.ca

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