

# PRENATAL GENETIC SCREENING

Information for Your Pregnancy



## KEY FACTS

- Prenatal genetic screening is a way for you to find out the chance that your baby has or does not have trisomy 21 or trisomy 18.
- This screening poses no risk to the pregnancy since it involves ultrasound and blood work.
- Prenatal genetic screening is not diagnostic. Only diagnostic testing, such as chorionic villus sampling (CVS) or amniocentesis, can give you a “yes” or “no” answer about trisomy 21 and trisomy 18 during the pregnancy.
- Prenatal genetic screening is available to all pregnant individuals in Ontario, and is optional. The routine pregnancy care you receive from your health-care provider will not be affected whether or not you choose to have this screening.

## WHAT DO WE SCREEN FOR?

Prenatal genetic screening involves screening for at least two common chromosome differences:

- trisomy 21 (Down syndrome)
- trisomy 18 (Edwards syndrome)

Anyone may have a baby with trisomy 21 or trisomy 18, regardless of their family history. This chance increases with the age of the mother (or the age of egg donor).

## IS PRENATAL GENETIC SCREENING RIGHT FOR ME?

Would knowing whether there is a higher or lower chance to have a baby with trisomy 21 or trisomy 18 be helpful to you during the pregnancy? Some people would prefer to wait for this information until the baby is born. Others would want to know if there is a chromosome difference to help them prepare for having a child that may require special care. When there is a chromosome difference, the health-care provider may recommend changes to how the pregnancy is looked after, or the birth plan. Some individuals would consider interrupting the pregnancy if the result is confirmed by further diagnostic testing.

You may wish to discuss the decision with your health-care provider if you have further questions. The choice whether to have prenatal genetic screening is personal. If you decide you do not want these tests, you can still have ultrasounds in the pregnancy.

## HOW DO WE SCREEN?

If you have not yet done a prenatal genetic screen, you can choose one of the following tests available in Ontario:

### enhanced First Trimester Screening (eFTS) / Maternal Serum Screening (MSS)

Both tests are covered by Ontario Health Insurance Plan (OHIP). How far along you are in the pregnancy and ultrasound availability in your area will determine which of these two tests is possible for you.

### Non-Invasive Prenatal Testing (NIPT)

NIPT is the most accurate prenatal genetic screening test, and is OHIP-funded in certain situations. You can choose to pay for NIPT out-of-pocket (private-pay) if you do not meet any of the [funding criteria](#) on our website.

### LEARN MORE

- Visit our [website](#) to read more about chromosome differences and available tests.
- Reach out to one of our Genetic Counsellors by phone or email.



Address: [www.prenatalscreeningontario.ca](http://www.prenatalscreeningontario.ca)



Phone: 1-833-351-6490



Email: [ps@bornontario.ca](mailto:ps@bornontario.ca)



PRENATAL SCREENING  
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### What is the Nuchal Translucency (NT) Ultrasound?

- Typically done as part of eFTS, from 11 weeks and 2 days to 13 weeks and 3 days of pregnancy.
- Measures the fluid-filled pocket at the back of the neck of the developing baby.
- Offers valuable information about the pregnancy, beyond screening for trisomy 21 and trisomy 18.
- You can consider this ultrasound in the first trimester even if you are choosing not to have eFTS.

Meaning, how many pregnancies where the baby really does have trisomy 21 will be flagged as "screen positive" (or "high risk") by this test?

Meaning, how many pregnancies will this test flag as "screen positive" (or "high risk") but the baby does not really have trisomy 21?

Meaning, how many pregnancies where the baby really does have trisomy 18 will be flagged as "screen positive" (or "high risk") by this test?

Meaning, how many pregnancies will this test flag as "screen positive" (or "high risk") but the baby does not really have trisomy 18?

Below are questions that you and your health-care provider can consider when choosing a test.

QUESTIONS TO CONSIDER	eFTS	MSS	NIPT
When in pregnancy is it done?	11 weeks and 2 days to 13 weeks and 3 days	14 weeks and 0 days to 20 weeks and 6 days	9-10 weeks or later
What does it include?	<ul style="list-style-type: none"> <li>• blood work</li> <li>• NT ultrasound</li> </ul>	<ul style="list-style-type: none"> <li>• blood work</li> </ul>	<ul style="list-style-type: none"> <li>• blood work</li> </ul>
What does it screen for?	<ul style="list-style-type: none"> <li>• trisomy 21</li> <li>• trisomy 18</li> </ul>	<ul style="list-style-type: none"> <li>• trisomy 21</li> <li>• trisomy 18</li> </ul>	<ul style="list-style-type: none"> <li>• trisomy 21</li> <li>• trisomy 18</li> <li>• trisomy 13</li> <li>• sex chromosome differences</li> <li>• microdeletion syndromes<sup>1</sup></li> </ul>
Can it be done if pregnant with more than one baby?	No	No	Yes, it can be done if you are pregnant with one baby or twins
What is the detection rate for trisomy 21? <sup>2</sup>	89%	79%	More than 99%
What is the false positive rate for trisomy 21?	6%	6%	Less than 0.1%
What is the detection rate for trisomy 18?	85%	39-89%	96%
What is the false positive rate for trisomy 18?	Less than 1%	Less than 1%	Less than 0.1%

1 The current Canadian guidelines do not currently recommend the use of NIPT to screen for microdeletion syndromes.  
 2 Detection rates and false positive rates were obtained from Ontario pregnancies (except twins) with a due date between Sept. 2016 and March 2021.