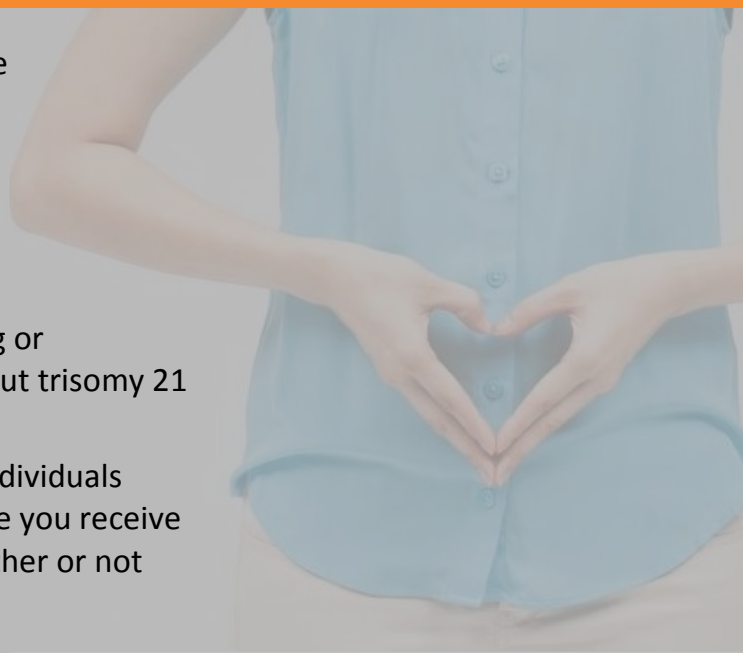
Ontario

PRENATAL GENETIC SCREENING



- Prenatal genetic screening is a way for you to determine the chance your baby may or may not have trisomy 21 or trisomy 18.
- This screening poses no risk to the pregnancy as it involves ultrasound and blood work.
- Prenatal genetic screening is not diagnostic. Only diagnostic testing, such as chorionic villus sampling 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 it is optional. The routine pregnancy care you receive from your healthcare provider will not be affected whether or not you choose to have this screening.

What Do We Screen for?

Traditional prenatal genetic screening involves screening for two common chromosome differences:

- trisomy 21 (Down syndrome)
- trisomy 18 (Edward 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 healthcare 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 healthcare 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.

NT Ultrasound

- Nuchal translucency (NT) ultrasound can be done in the first trimester, between 11 weeks 2 days and 13 weeks 3 days.
- 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.
- Can be considered even if you decline prenatal genetic screening.

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- Read more about chromosome differences and available tests
- Speak to a Genetic Counsellor

www.prenatalscreeningontario.ca

1-833-351-6490

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Prenatal Genetic Screening at a Glance

Traditional prenatal genetic screening is funded by the Ministry of Health for all pregnant individuals in Ontario. How far along you are in the pregnancy, how many babies you are carrying, and availability in your area will determine which of the two tests is possible for you:

- enhanced First Trimester Screen (eFTS) or
- Maternal Serum Screen (MSS)

Non-invasive Prenatal Testing (NIPT) is a highly effective screen and it is funded by the Ministry of Health in certain situations. NIPT can be paid for out-of-pocket (self-pay) by those who do not meet any of the criteria for funding, which are listed on our website.

	eFTS	MSS	NIPT
When in pregnancy is it done?	11 weeks and 2 days to 13 weeks and 3 days	14 weeks 0 days to 20 weeks and 6 days	9 weeks or later
What does it include?	♦ maternal blood work ♦ NT ultrasound	♦ maternal blood work	♦ maternal blood work
What does it screen for?	♦ trisomy 21 ♦ trisomy 18	♦ trisomy 21 ♦ trisomy 18	♦ trisomy 21 ♦ trisomy 18 ♦ trisomy 13 ♦ sex chromosome differences ♦ Microdeletions*
Detection rate (trisomy 21)	*Meaning, how many pregnancies where the baby really DOES have trisomy 21 will be flagged as screen positive (or high risk) by this test?*		
	89%	81%	More than 99%
False positive rate (trisomy 21)	*Meaning, how many pregnancies will this test flag as screen positive (or high risk) but the baby does NOT really have trisomy 21?*		
	7%	5%	0.1%
Detection rate (trisomy 18)	*Meaning, how many pregnancies where the baby really DOES have trisomy 18 will be flagged as screen positive (or high risk) by this test?*		
	78%	60%	95%
False positive rate (trisomy 18)	*Meaning, how many pregnancies will this test flag as screen positive (or high risk) but the baby does NOT really have trisomy 18?*		
	0.2%	0.5%	Less than 0.1%

*The current guidelines in Ontario do not currently support the use of NIPT to screen for microdeletion syndromes, such as 22q11.2 deletion syndrome.

NOTES:

1. The detection rates and false positive rates for the screening tests were obtained from Ontario data.
2. eFTS may be less available during the COVID-19 pandemic. In response to this, a temporary change has been made to MSS screening cut-off to make the trisomy 21 detection rate similar to that of eFTS.

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