

Come and see what all of the buzz is about!

## Prenatal Testing

Before you decide whether you want to proceed with prenatal testing it is important to know what you would do with the information you would receive (the test result). For many parents, the process of prenatal testing with quell worries and anxiety about the baby's health, while for others it can cause worry and anxiety. You might want to consider whether a positive screen/test result would:

- Inform your decision to carry on with the pregnancy, or consider an interruption to the pregnancy
- Help you and your family prepare for a baby who may have special needs/considerations for labour, birth and postpartum
- Not influence your decisions, whatsoever



What does prenatal testing screen for?

Genetic Disorders: A genetic disorder is a disease caused by a change in the normal DNA sequence, and they can fall into two major categories:

**Aneuploidy** – any condition where there are three copies, instead of two, or one copy instead of two, of chromosomes. Most families are familiar with Down Syndrome, which is “Trisomy 21,” or three copies of chromosome 21. The most common chromosomal anomalies that prenatal screening tests for include Down Syndrome, Patau Syndrome, Edwards Syndrome, and Turner Syndrome.

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**Inherited Disorders** – can cause a number of conditions that result from mutations to the genes. In most cases both parents need to be carriers of the gene to have a child that is affected by the condition. Common inherited disorders include Sickle cell disease, Cystic Fibrosis, and Tay-Sachs disease.

## Screening versus Diagnosis

Prenatal screening tests ARE NOT diagnostic, and instead, the screening is an attempt to quantify (assign a number) to the risk of a person having a baby with a specific disorder.

**Trisomy 21** – The screening test has a cut-off of 1 in 350 for Trisomy 21. If your screen is “positive” then it is because the chance of your baby having Trisomy 21 was calculated to be greater than 1 in 350. If your screen is “negative,” then the chance of your baby having Trisomy 21 is less than 1 in 350, and may be reported as 1 in 17,000 (for example).

**Trisomy 18** – The test for Trisomy 18 has a cut-off of 1 in 200. If you got a “positive” screen result for Trisomy 18, it is because the chance of your baby having Trisomy 18 was calculated to be higher than 1 in 200.

## Understanding your chance:

If the Prenatal Screening Test is positive for Trisomy 21 and the number on your report is 1 in 100, then it means that out of the **100 people** who get **the same positive result**, 1 person will have a pregnancy with a baby who has Trisomy 21, while the other 99 people will not.



● Has trisomy 21 ● Does not have trisomy 21

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## What screening test is covered by Public Health: **First Trimester Screen**

Alberta Health Services finances the costs for all pregnant people in Alberta who have a valid health card to have Prenatal Genetic Screening with a blood test and ultrasound test called **The First Trimester Screen**. The results from the blood tests and measurements from the ultrasound scan (between the 11<sup>th</sup> week and before the 14<sup>th</sup> week of pregnancy) are put into an algorithm to tell you what the chances (or risk ratio) are that your baby might have the condition. It is 82% effective in detecting those who go on to have the condition, but this also means that people who screen negative have a 5-7% chance at having a baby who has the condition. If you have a history of genetic or family conditions, then your care provider may recommend an Invasive Prenatal Test to diagnose the condition.

## What screening test is not covered by Public Health: **Non-Invasive Prenatal Screening (NIPS)**

Privately, people can elect to pay for Cell-free DNA screening, and in Alberta, it is known as Non-Invasive Prenatal Screening “NIPS.” Anytime after 10-weeks of pregnancy a pregnant person can go into the lab and the blood collected will be tested for the conditions. NIPS offers more conditions to screen for than the First Trimester Screen and can tell the parents what the sex chromosomes are. Although NIPS is a screening test, because the test is not directly testing the baby, it is 99% accurate in determining whether the baby has the genetic/inheritable condition. Some people want to have the NIPS done as soon as they qualify (at 10 weeks of pregnancy), while others choose to use NIPS if they screened positive following the First Trimester Screen.

For those who decide not to proceed with any prenatal screening tests, then many conditions and disorders might appear at the Anatomy Screen Ultrasound Scan between 18-20 weeks of pregnancy. The detailed ultrasound between 18-20 weeks of pregnancy looks at baby’s features inside (organ systems) and out (limbs, fingers, and toes to name a few) and can often give families more information about whether their baby is healthy, or not, and whether baby has a particular condition/anomaly. The ultrasound **alone** cannot guarantee a healthy baby.

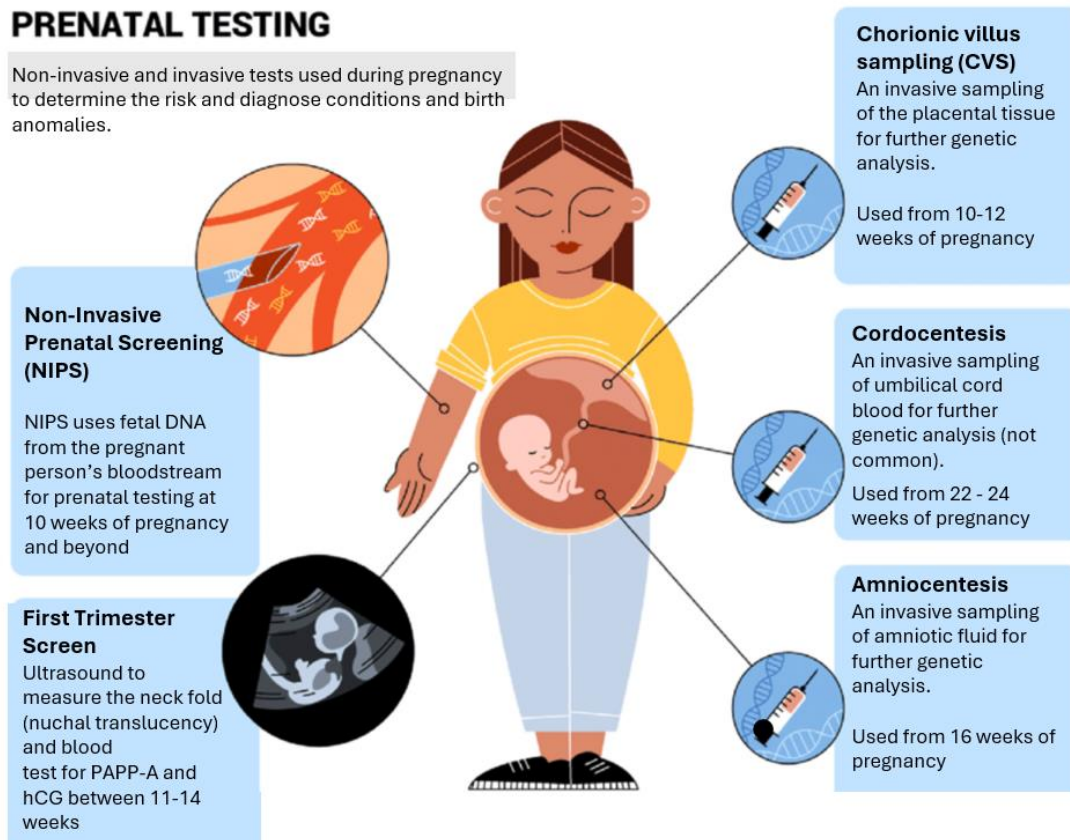
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## Invasive Prenatal Diagnostic Testing: Chorionic Villus Sampling and Amniocentesis

Chorionic Villus Sampling and Amniocentesis are less used with the availability of NIPS. Pregnant people who have rare and less common genetic disorders may decide to undergo either of these diagnostic tests to determine the health of the baby. You can ask your midwifery team if your family history warrants the need for invasive prenatal diagnostic testing (most people won't require this).

### PRENATAL TESTING

Non-invasive and invasive tests used during pregnancy to determine the risk and diagnose conditions and birth anomalies.



Alberta Health Services. 2021. Prenatal screening and testing <https://myhealth.alberta.ca/genetics/prenatal-screening-and-testing>

Alberta Health Services. 2021. Amniocentesis. <https://myhealth.alberta.ca/Health/Pages/conditions.aspx?hwid=hw1810>