What are chromosome conditions?

A chromosome condition is when there are extra or missing chromosomes, or pieces of chromosomes re-arrange themselves. This can affect both intellectual and physical development.

Down syndrome is the most common chromosome condition. People with Down syndrome have 47 instead of 46 chromosomes, having an extra chromosome 21.

People with Down syndrome have some learning issues and can also have health problems. It is hard to know how much Down syndrome will affect someone. It can happen in pregnancies of women of any age; however, it becomes more common as a woman gets older.

What are birth defects?

A birth defect is a physical condition that a baby is born with. Examples of birth defects include spinal bifida, heart defects, and cleft lip.

All women, no matter their age, their pregnancy history, or family history, have a 2% to 3% chance of having a baby born with a birth defect.

The 18–20 week ultrasound all women are offered checks a baby's growth and development and look for (screen) for birth defects. Go to our website for more information about the ultrasound.

More Questions?

We are here as a resource for you. Please feel free to ask questions during your appointment. You may also contact the Program Coordinator or speak with a nurse at your appointment if you have any questions.

Just Remember...

- Most babies are born healthy.
- Having prenatal testing is your choice.

Contact Us

ERA Program Coordinator: 403-943-8382
ERA Program Secretary: 403-943-8379

Visit www.earlyriskassessment.com to learn more about FTS, prenatal diagnostic testing, and other prenatal testing options.

EARLY PRENATAL RISK ASSESSMENT PROGRAM

health information

FIRST TRIMESTER COMBINED SCREENING (FTS)

Alberta Health Services

First Trimester Combined Screening

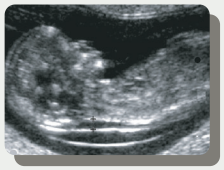
FTS is a screening test for chromosome conditions and birth defects. It is available to all women between 11 weeks and 13 weeks 6 days of pregnancy.

Two measurements, together with your age, are used to estimate your chance of having a baby with Down syndrome (trisomy 21), trisomy 18, or trisomy 13.

The first measurement is the nuchal translucency (NT) measurement (taken by ultrasound) and the second is a blood test.

The NT Measurement

The ultrasound to measure the NT is done between 11 weeks and 13 weeks 6 days of pregnancy. All babies have some fluid at the back of their neck. This amount of fluid is measured. More fluid is often found babies with Down syndrome.



12 week fetus with normal NT

The NT ultrasound also:

- confirms that your baby is alive
- confirms your dates
- diagnoses a multiple pregnancy
- checks for birth defects that can be seen at this stage

The Maternal Blood Test

This blood test measures two substances (PAPP-A and free Beta-hCG) that are normally found in the blood of all pregnant women. These levels tend to be higher in pregnancies with Down syndrome.

Between 85–90% of pregnancies with Down syndrome, trisomy 18, or trisomy 13 are identified by FTS (called the detection rate).

What type of results should I expect from FTS?

You will be given a "personal risk estimate" that tells you how likely it is that your pregnancy will be affected with Down syndrome, trisomy 13, or trisomy 18. The FTS report calls it the "adjusted risk".

What happens after FTS?

It's important to remember that FTS won't tell you for sure if your baby has a chromosomal condition or health concern. Instead, it tells you the chance that your baby could be affected. Your FTS result will give you the information you need to make an informed decision about having more testing.

18–20 Week Ultrasound

No matter what type of prenatal genetic screening you have, an 18–20 week ultrasound is recommended for **all pregnant women** in Canada. Many major birth defects, like spinal bifida, can be found with this ultrasound.

Diagnostic Tests

Amniocentesis and chorionic villus sampling (CVS) are diagnostic tests, meaning the test can diagnose a chromosome condition. Samples collected from these tests contain cells from the baby. The laboratory looks at the chromosomes within these cells to learn whether or not the baby has a chromosome condition.

Amniocentesis and CVS testing are only offered to women who have pregnancies that are at increased risk for a chromosomal condition. Although these procedures are invasive, most women don't have complications after the procedure and receive an accurate chromosome result.

What are the benefits of FTS?

- Early, more accurate screening may give you peace of mind.
- The FTS personal risk estimate can be used to help you make a more informed choice about diagnostic testing.
- It is possible to find certain major birth defects during the NT scan.

Limitations of FTS

- About 1 in 20 women will be told they are at an increased risk or have a "positive screen". It's normal to be worried when you hear that; however, most women with an increased-risk result do have healthy babies.
- An increased-risk result does not mean that the baby has a chromosome condition.
- A reduced-risk result does not guarantee a healthy baby.