First Trimester Screen (FTS)

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 in babies with Down syndrome.
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 for sure say 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.

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.

Maternal Serum Screen (MSS)

(Quad Screen)

This screen involves a blood test best performed after 15.0 weeks in pregnancy. The purpose of the screen is to estimate the risk of having a pregnancy with one of the following: Down syndrome, Trisomy 18 or Neural Tube Defects. It is only a screening test and further investigations are available if the test is abnormal.

***If you have already had a FTS in your current pregnancy, you would not be eligible for MSS*

18–20 Week Ultrasound

Even if you choose not to have prenatal genetic screening, an 18–20 week ultrasound is recommended for all pregnant women in Canada. Many major birth defects, like spina bifida, can be found with this ultrasound. It is considered safe for both the woman and baby.

Some women are told that an "ultrasound marker" has been detected. A "marker" is a slight difference in organ development. Markers are often seen in normal babies but are also seen in babies with chromosome conditions such as Down syndrome or trisomy 18. If a marker is found in your ultrasound, you will be given more information and may also receive a referral for prenatal genetic counseling or to a specialist in maternal fetal medicine.

Diagnostic Tests

Amniocentesis and chorionic villus sampling (CVS) testing are only offered to women who have pregnancies that are at increased risk for a chromosomal condition eg. positive FTS or ultrasound markers.

Amniocentesis and 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.
Although these procedures are invasive and have a small chance of miscarriage associated with them, most women don't have complications after the procedure and receive an accurate chromosome result.

What are birth defects?

A birth defect is a physical condition that a baby is born with. Examples of birth defects include spina 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.

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.

ESTIMATED RATES OF CHROMOSOME ABNORMALITIES

(In Live Births by Maternal Age at Expected Time of Birth)

Maternal Age At Birth	Down Syndrome	Chromosome Abnormality
20	1 in 1667	1 in 526
21	1 in 1667	1 in 526
22	1 in 1429	1 in 500
23	1 in 1429	1 in 500
24	1 in 1250	1 in 476
25	1 in 1250	1 in 476
26	1 in 1176	1 in 476
27	1 in 1111	1 in 455
28	1 in 1053	1 in 435
29	1 in 1000	1 in 417
30	1 in 952	1 in 384
31	1 in 909	1 in 384
32	1 in 769	1 in 323
33	1 in 625	1 in 286
34	1 in 500	1 in 238
35	1 in 385	1 in 192
36	1 in 294	1 in 156
37	1 in 227	1 in 127
38	1 in 175	1 in 102
39	1 in 137	1 in 83
40	1 in 106	1 in 66
41	1 in 82	1 in 53
42	1 in 64	1 in 42
43	1 in 50	1 in 33
44	1 in 38	1 in 26
45	1 in 30	1 in 21

WHO MAY BENEFIT FROM PRENATAL COUNSELLING:

- Women who have received results that have identified an increased risk to the pregnancy (e.g. positive prenatal screen or ultrasound marker)
- Women who have taken a medication or a drug during pregnancy and are concerned that it might cause a problem for the baby
- Couples who have had a previous child with a chromosome abnormality, such as Down syndrome.
- Women who are carriers or whose partners are carriers of a familial chromosome disorder.
- Couples who have a family history of spina bifida, meningomyelocele or anencephaly.
- A woman who is a carrier of a gene that causes a disorder in male children (e.g.hemophilia or Duchenne Muscular Dystrophy).
- Couples who are both known to carry a gene causing a recessive metabolic disorder for which a prenatal diagnostic test is available (example: Cystic Fibrosis). When there is doubt as to the possibility of increased risk, a genetic counselor is available for consultation.
- Couples from families in which other genetic diseases are known to occur and where direct or indirect examination of the gene is possible by molecular (DNA) means.

More Questions?

If you decide to go ahead with prenatal screening, your doctor will arrange it and provide you with the results.

But if you have more questions, feel free to contact a genetic counselor at:

Red Deer Outreach Genetics Clinic
(403) 314-5226/5228

Just Remember . . .

- **Most babies are born healthy**
- **Have prenatal screening is your choice**

Prenatal Screening Information Guide

Alberta Health Services