What are chromosome conditions?

A chromosome condition occurs when there are extra or missing chromosomes, or a rearrangement of pieces of chromosomes. Intellectual and physical development can be affected as a result of this chromosome difference. Down syndrome, also known as trisomy 21, 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 difficulties and can also have a number of health problems. It is hard to predict how much people with Down syndrome will be affected with intellectual and health challenges. Down syndrome can occur in pregnancies of women of any age; however, this condition 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 spina bifida, heart defects and cleft lip. All women regardless of their age and family or previous pregnancy history have a 2-3% chance of having a baby born with a birth defect. The 18-20 week ultrasound that is offered to all women is used to evaluate the baby's growth and development and screen for birth defects. Further information about the 18-20 week ultrasound is available on the program's website.

More Questions?

We are here as a resource for you. We encourage you to ask questions during your appointment. You may also contact the Program Manager or nurse at the Maternal Fetal Medicine Centre with your questions.

Just Remember

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

FTS provided through the Southern Alberta Centre for Maternal Medicine:

North Location:
Suite 100 - 3280 Hospital Drive NW Calgary AB.

South Location:
10333 Southport Rd. SW Calgary AB.

Appointments: (403) 289-9269
Program Manager: (403) 943-8382

EARLY PRENATAL RISK ASSESSMENT PROGRAM

health information

FIRST TRIMESTER COMBINED SCREENING (FTS)

EARLY PRENATAL RISK ASSESSMENT PROGRAM

www.earlyriskassessment.com

"Provided through EFW Radiology, Specialists in Diagnostic Imaging at the Southern Alberta Centre for Maternal Medicine"

EFW Radiology Specialists in Diagnostic Imaging

calgary health region

Calgary Laboratory Services

The Early Prenatal Risk Assessment Program is a multidisciplinary collaborative of the Calgary Health Region, EFW Radiology, Specialists in Diagnostic Imaging, and Calgary Laboratory Services.

First Trimester Combined Screen

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.

With FTS, two measurements are used, together with your age, to estimate your chance of having a baby with Down syndrome, trisomy 18, or trisomy 13. The first is the nuchal translucency (NT) measurement taken by ultrasound, and the second is a blood test.

The Nuchal Translucency (NT) Measurement

The NT measurement involves a special ultrasound done between 11 weeks and 13 weeks 6 days of pregnancy. A measurement of the fluid at the back of the baby's neck (called nuchal translucency) is taken. All babies will have some fluid here. The fluid is more likely to be increased in pregnancies with Down syndrome.

12 week fetus with normal NT

The NT ultrasound also:

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

The Maternal Blood Test

The maternal blood test measures two substances (PAPP-A and free Beta-hCG), which are normally found in the blood of all pregnant women. In pregnancies with Down syndrome, the levels of free Beta-hCG and PAPP-A tend to be out of the expected normal range.

FTS has an 85–90% detection rate. This means that 85–90% of pregnancies with Down syndrome, trisomy 18, or trisomy 13 will be identified at increased risk with FTS.

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. In the FTS report, this is called the "adjusted risk".

What happens after FTS?

Your FTS result helps your doctor decide if you should be offered more prenatal testing (such as amniocentesis). Most women receive reassuring results and are not offered further testing, other than the 18 week ultrasound that is routinely offered to all women. Women who receive a risk result that places them at an increased risk for Down syndrome, trisomy 18, or trisomy 13 are offered the option of having a diagnostic test such as amniocentesis. Regardless of a woman's age or FTS results, the decision to have a diagnostic test is a personal one.

Diagnostic Tests

Amniocentesis and chorionic villus sampling (CVS) are diagnostic tests. Samples collected from these tests contain cells from the baby. The laboratory looks at the chromosomes within these cells in detail to determine whether or not the baby has a chromosome condition. These procedures are not routinely offered to all women, as they place the pregnancy at an increased risk for miscarriage. Most women undergoing an amniocentesis or chorionic villus sampling do not have complications following the procedure and receive reassuring chromosome results.

Women over 35 years are routinely offered amniocentesis. Many use their FTS results to help them make a decision about diagnostic testing.

Benefits of FTS

- Early, more accurate screening gives peace of mind to many women.
- The FTS personal risk estimate can be used to assist women in making a more informed choice about diagnostic testing.
- It is possible to detect certain major birth defects at the time of the NT scan.

Limitations of FTS

- About 1 in 20 women will receive an increased-risk result. It is normal to be worried when you hear you are at increased risk. 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.