

Reproductive Genetics Institute

First Trimester Screening Pamphlet

An earlier non - invasive way to screen for Down syndrome in your pregnancy

What is the first trimester screen?

First trimester screening (FTS) is a recent advancement in prenatal care. Through the combination of a special ultrasound examination and a simple blood test, it is possible to identify pregnancies that have an increased risk of having a baby with a chromosome abnormality (such as Down syndrome), as well as other birth defects during the first trimester of pregnancy.

What does FTS involve?

1. Ultrasound

Either transvaginal or transabdominal ultrasound may be performed. This form of testing is noninvasive and runs no risk of harming the fetus. During the exam, the gestational age is estimated (first trimester ultrasound is the best method for this), and a measurement is taken of the “skin” thickness in the neck area (nuchal fold). In 80% of fetuses with chromosome abnormalities, the nuchal thickness is increased. Of the participants, it is expected that 4% will have a nuchal thickness measurement which indicates an increased risk of a fetus with Down syndrome or another chromosome problem, or possibly a heart problem.

2. Blood screen

A simple blood test measures three chemicals found in the mother’s circulation: Pregnancy Associated Plasma Protein A (PAPP-A), free beta hCG, and alpha-fetoprotein (AFP). The levels of these chemicals may indicate an increased or decreased risk of the fetus having chromosome abnormalities. Approximately 75% of pregnancies with chromosome problems are detected by this procedure.

Studies show that 5% of patients having this test have abnormal results indicating increased risk of chromosome abnormalities in the fetus.

By combining the results of the ultrasound and blood test, along with the age, race, and weight of the mother, a number can be generated which represents the risk of the pregnancy being affected by Down syndrome or another chromosome problem. This combined risk estimate is more powerful than either test used separately.

How accurate is this screening test?

Research has shown that, together, the ultrasound and blood screen identify 87% of fetuses with chromosome abnormalities.

Why should I consider having FTS?

FTS is beneficial for the following reasons:

1. Earlier detection of an increased risk in your pregnancy of Down syndrome, other chromosome abnormalities, or birth defects can make first trimester prenatal testing a valuable option. Chorionic villus sampling (CVS) can be performed to confirm whether the fetus has a chromosome abnormality. If results of this test indicate that the fetus has a chromosome abnormality, there is more time for counseling and decision making, often prior to when a woman is “showing.” In addition, there are more options regarding pregnancy termination.
2. First trimester ultrasound is the most accurate way of dating your pregnancy. This information can be invaluable in pregnancy management.
3. First trimester ultrasound can identify if you have a multiple gestation and determine whether they are monozygotic (identical) or dizygotic (fraternal) twins. This can aid in the obstetrical management of the pregnancy.
4. FTS can detect a higher percentage of fetuses with Down syndrome than the currently available second trimester screening test (MSAFP, triple screen, or quad screen tests).
5. FTS allows for early detection of many severe fetal malformations of chromosomal and non-chromosomal origin.

What are the risks of FTS?

FTS involves minimal risk. Patients receiving abnormal FTS results will be offered prenatal diagnostic testing to determine if a chromosome problem is present. These procedures, called CVS and amniocentesis, each carry a risk of miscarriage. As with any test, if a patient receives an FTS that shows abnormal results, her anxiety about the pregnancy may increase.

Who is eligible for FTS?

Patients must be between 10 ½ and 13 weeks, six days of pregnancy to have FTS performed. The **optimal** time to have tests done simultaneously is between 10 ½ and 12 weeks of pregnancy. **Women ages 35 and over should be offered diagnostic testing based on age-related risk alone.**

When are results available?

Results will be available in less than one week and will be reported to both the patient and the referring physician. If the results reveal an increased risk for Down syndrome or another chromosome abnormality, a session with a genetic counselor or physician will be offered, during which diagnostic options can be explained. Both CVS (performed between 10-13 weeks or pregnancy) and amniocentesis (performed at 16+ weeks) can accurately diagnose any chromosome abnormalities in the pregnancy.

A summary of the advantages of the first trimester screen:

1. Earlier detection of an increased risk for carrying a child with Down syndrome or other chromosome problem which allows more time for decision making, including the option of first trimester prenatal diagnosis.
2. Better detection rate (87% versus 60-80%) than the MSAFP (triple/quad screen) for Down syndrome and other chromosome problems.
3. Possible identification of heart defects associated with abnormal test results through further testing.
4. If the results show an increased risk, several testing options exist.
5. Like the MSAFP (triple/quad screen), this test is noninvasive and safe to the fetus.

For more information, or to schedule an appointment, please call the Reproductive Genetics Institute at (773) 472-4900.