LEGACY LABORATORY SERVICES

UNDERSTANDING FIRST-TRIMESTER SCREENING

What is First-trimester screening?

FTS is designed to identify pregnancies at higher than background risk for Down syndrome and trisomy 18 in the first trimester. The test does not diagnose either condition, but provides a risk estimate.

Who may choose to be screened?

Women between 11 and 14 weeks pregnant from the first day of their last menstrual period may choose to be screened. This is an optional test.

What does the screening involve?

The test involves a combination of a special ultrasound examination and a blood test. The ultrasound examination measures a space found at the back of the fetal neck called the nuchal translucency (NT). A small amount of blood is drawn to measure levels of two proteins, Beta-hCG (the pregnancy hormone) and PAPP-A (Pregnancy associated plasma protein-A). These proteins are normally found in the blood of all pregnant women. This screen is safe for the mother and the fetus.

What is Down syndrome and trisomy 18?

These problems are caused by the fetus having an extra chromosome in each cell. The extra chromosome, which contains hundreds of genes, causes birth defects and mental retardation. The common forms of Down syndrome (trisomy 21) and trisomy 18 are usually not passed along in families. Any woman could have a baby with a chromosomal abnormality. The risk of these types of problems increases with maternal age.

When will I get results? How are they reported?

Final results are reported as “positive” (when your risk is determined to be higher than expected) or “negative” (when your risk is determined to be lower than expected). For Down syndrome, the standard of comparison is the risk of a woman who is 35 years old. Results will be sent to your doctor. Your doctor or genetic counselor will contact you to discuss the result of the screen and options for further testing, if necessary.

What do I need to do if my results are negative?

A negative screen means that your risk for Down syndrome or trisomy 18 is less than the screening cut-off. FTS can never eliminate the chance for a chromosome problem. Down

syndrome and trisomy 18 make up about half of all chromosome abnormalities. This screening estimates the risk for Down syndrome and trisomy 18, but not for the other half of chromosome abnormalities or other genetic disorders. This screening can detect about 80-90 percent of Down syndrome pregnancies. There is no screening test that will detect 100 percent of Down syndrome pregnancies.

What if my results are positive?

A positive screen means that you have an increased chance to have a baby with Down syndrome or trisomy 18. It does not mean a chromosome abnormality has been diagnosed. In many cases, the baby does not have Down syndrome or trisomy 18. Your doctor or genetics counselor will contact you with the results of the screen and options for further testing.

What tests are available for confirmation if my screen is positive?

If you have a positive screen, diagnostic testing is available by either chorionic villus sampling (CVS) (until 14 weeks) or amniocentesis (after 15 weeks). Your doctor or genetic counselor can discuss these options with you in greater detail.

How does my age affect the options available to me?

Traditionally the American College of Obstetricians and Gynecologists (ACG) has recommended that all women who will be 35 or older at delivery be offered CVS or amniocentesis for prenatal chromosome testing. Most pregnancies with Down syndrome, however, occur in women who are younger than 35. Recently, ACPG amended its recommendations to say that screening and diagnostic tests should be available to all women regardless of age, and that women be counseled regarding the differences between the two types of testing. While screening tests are good options for all women, some families may feel that a diagnostic test is more appropriate for them. FTS is not a substitute for diagnostic testing because screening 1) does not give a diagnosis, 2) estimates a risk for only two chromosome abnormalities and misses others that you may be at risk for, and 3) may falsely reassure women who still have an increased risk or chromosome abnormalities. We recommend that all women age 35 or older meet with their doctor or a genetic counselor to discuss risks and options due to the higher background risk for problems. If you are younger than 35 and considering a diagnostic test or have questions, please speak with your doctor or a genetic counselor.

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What if I have a family history of Down syndrome or other chromosomal genetic abnormalities?

FTS should not replace diagnostic testing in these situations because it may provide false reassurance. Parents/couples with a family history of any genetic disease (including Down syndrome) should discuss diagnostic testing with their doctor and/or genetic counselor.

What else does a first trimester screen look for?

If the nuchal translucency is larger than expected, there is a higher chance of other problems, such as heart defects. If the nuchal translucency is larger than expected, you should meet with your doctor or a genetic counselor.

What is the accuracy of the test?

First-trimester screening has about 80-90 percent detection for Down syndrome. Detection for trisomy 18 is approximately 80% percent.

Can all birth defects be detected by this or any other screening?

No. No prenatal test can guarantee a healthy child.

Will my insurance pay for FTS?

Most insurance companies cover the cost of FTS. If you have any questions about whether your insurance will cover testing, please contact your insurance company.

If you have additional questions, please contact your doctor.