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First Trimester Screening

Every pregnancy, regardless of a woman's age, has a chance to have a chromosome condition. The most common chromosome condition is Down syndrome, though there are other chromosome conditions that can be more severe. First Trimester Screening is a combination of blood testing and particular ultrasound measurements available to women who would like to learn more about their chances to have a pregnancy with one of these conditions. All genetic testing is optional and this information is intended to help you understand your options. Keep in mind, no test or combination of tests can screen for all genetic conditions and birth defects.

What is First Trimester Screening and when is it done?

First Trimester Screening, also called FTS, is a testing option performed during pregnancy that screens for Down syndrome (also called trisomy 21), trisomy 18, and trisomy 13. FTS does not screen for all genetic conditions. Using a combination of factors including the age of the mother, ultrasound measurements, and a sample of mother's blood (which is analyzed for naturally occurring proteins that are made by the pregnancy), FTS provides a risk estimate for your pregnancy to have these three chromosome conditions. FTS can be performed between the beginning of the 11th and end of the 13th week of pregnancy and it takes about 1 week to get the results back

How accurate is First Trimester Screening?

FTS is only a screening test, it is not 100% accurate. It is not considered a diagnostic test (see What are my other options?). The detection rate is laboratory-dependent but is typically between 80-90% accurate and has a false positive rate of approximately 5%.

What are the possible results?

Negative/Low Risk: A negative or low risk result means that it is very unlikely the pregnancy has one of the conditions FTS screens for. It is again important to remember that this test is not diagnostic and a relatively low chance still remains for the pregnancy to have one of the conditions.

Positive/High Risk: A positive or high risk result means the pregnancy is at increased risk for a particular condition. Only diagnostic testing can provide a definite answer. Genetic counseling is recommended to discuss positive/high risk results along with further testing options.

False positive and false negative results are possible with FTS, therefore a result may be negative/low risk and a baby be born with one of these conditions, or may be positive/high risk and the baby not have that condition.

Can anyone have the First Trimester Screen?

Yes, anyone whose pregnancy measures 11 weeks to 13 weeks and 6 days may have the first trimester screen. If a woman is at an increased risk for their pregnancy to have one of these chromosome conditions, such as women who are 35 years or older at the time of delivery, who had a previous pregnancy or child with a chromosome condition, or when there are concerns on ultrasound in their current pregnancy, should consider speaking with a genetic counselor to discuss additional testing options.

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What are my other options?

Ultrasound: screens for birth defects and markers for genetic conditions. Approximately 50% of pregnancies with Down syndrome and 90% of pregnancies with trisomy 18 and trisomy 13 have findings on ultrasound that raise concern. Thus, a normal ultrasound can be reassuring and lower the risk for these conditions, but it cannot rule out all birth defects or genetic conditions.

Second Trimester Screening: screens for Down syndrome, trisomy 18, and open neural tube defects (called spina bifida). These tests provide an estimate of risk, or chance, that the pregnancy has one of these conditions; however, they are not diagnostic. Depending on the test, approximately 75-80% of pregnancies with these conditions will screen positive and about 5% of pregnancies will receive a false positive result.

Non-invasive Prenatal Testing: also called NIPT, is a blood test performed during pregnancy that screens for Down syndrome, trisomy 18, and trisomy 13. Additionally, NIPT screens for conditions caused by extra or missing sex chromosomes. The detection rate is laboratory-dependent but is typically between 90-99% for high-risk, singleton (only one baby) pregnancies with false positive rates of less than 1%.

Diagnostic Testing: provides a "yes" or "no" answer regarding chromosome conditions during the pregnancy. There are two options. Both diagnostic procedures are performed by an experienced obstetrician under ultrasound guidance; however, both options carry a risk for pregnancy complications and/or miscarriage, less than 1%.

- Chorionic villus sampling (CVS): procedure between 10-14 weeks of pregnancy that samples a small piece of the developing placenta to evaluate the pregnancy's chromosomes.
- Amniocentesis: procedure between 16-20 weeks of pregnancy that samples a small amount of amniotic fluid to evaluate the pregnancy's chromosomes.

How do I know if FTS is right for me?

Talking to a genetic counselor may be beneficial in helping you decide on a personal screening plan. Genetic counselors are healthcare professionals trained to discuss the various risk factors during pregnancy and review the risks, limitations, and benefits of screening and diagnostic testing, as well as what it may mean for a family if test results are concerning for a condition. It is also important to remember that while most babies are born without complications, there is always a 3-5% risk for birth defects in any pregnancy.

If you have additional questions about these testing options and would like to speak to a genetic counselor, please inform the front desk and this will be arranged for you.

Kindest regards,

The Prenatal Genetic Counselors and Care Team