First-Trimester Screening

Palmetto Health USC
OBSTETRICS AND GYNECOLOGY
GENETIC COUNSELING

First-trimester screening is an optional screening test to determine if your baby has an increased chance for Down syndrome, Trisomy 18 or Trisomy 13. These conditions occur when an extra chromosome is present in every cell of a baby's body. Babies with Down syndrome have common physical features with mild to moderate intellectual disability and may also have other birth defects such as heart problems. Trisomy 18 and Trisomy 13 are less common than Down syndrome, but they are serious conditions that include birth defects, severe disability, and shortened lifespan.

How does it work?

This screening test involves a blood draw and an ultrasound exam to estimate the chance of a pregnancy having Down syndrome, Trisomy 18 or Trisomy 13.

A blood sample is drawn between 9 and 13 weeks after the last menstrual period. Two or three proteins normally found in every pregnancy (hCG, PAPP-A, sometimes AFP) are measured from the patient's blood sample, and the levels of these proteins are used to predict the chances for the conditions.

A certified sonographer performs an ultrasound between 11 and 13 weeks that measures the nuchal translucency (NT). An NT is a fluid-filled space in the back of the baby's neck that is normal; however, babies with chromosome conditions usually have an NT that is larger than average. An increased NT suggests a higher chance for a chromosome condition in the pregnancy or, in some cases, other birth defects.

If only the blood screen is performed, 60% to 70% of pregnancies with these conditions will be detected. If both the blood sample and the nuchal translucency are measured, 90% of pregnancies with Down syndrome and 95% of pregnancies with Trisomy 18 or Trisomy 13 will be detected. Nuchal translucency measurement, with or without the blood sample, may be useful in cases of twins or triplet pregnancies although the detection rates are somewhat lower.

What does it mean to be screen positive?

If the screening test shows an increased chance for Down syndrome, Trisomy 18 or Trisomy 13, the result is interpreted as "screen positive." A positive screen is not diagnostic, but suggests that there is an increased chance, and follow up testing is offered to rule out or diagnose the condition. There are three options for follow up testing including chorionic villus sampling (CVS), amniocentesis, and/or non-invasive prenatal screening (NIPS). Genetic counseling is helpful to discuss the options for further testing/screening in the pregnancy including their benefits, limitations and risks.

What other tests are suggested for women who have increased nuchal translucency?

If CVS, amniocentesis or NIPS studies are normal, sometimes the explanation for the increased measurement can be due to a fetal heart defect or other birth defect. Detailed ultrasound including a fetal echocardiogram can be done later in pregnancy (usually between 20 and 24 weeks) to help determine if there is a heart defect or other birth defect.

What does it mean to be screen negative?

If first-trimester screening does not show an increased risk, the result is interpreted as "screen negative." Patients will still be offered the option of a separate screening for neural tube defects (NTDs) in the second trimester (ideally between 15 and 18 weeks). NTDs are birth defects that occur when the spinal cord does not close properly. MSAFP, a protein made by the fetus that is present in maternal blood, may be found at higher levels when an NTD is present in the baby. Women with a high level of MSAFP will be offered follow up testing for NTD. Also, most women will be offered a detailed ultrasound in the second trimester (20 to 22 weeks) to check the growth and development of the baby.

Who should have the testing?

First-trimester screening is offered to all women who do not have a family history of one of these conditions. Women over 35 years of age or with a family history of chromosome conditions are usually offered genetic counseling and additional testing options, such as CVS, amniocentesis, and/or non-invasive prenatal screening (NIPS).

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