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PRENATAL TESTING OPTIONS

As part of your obstetrical care, our office offers several prenatal tests to identify whether a pregnancy may be at risk for various congenital or hereditary problems. This literature is for informational purposes only.

Ultrasound

Ultrasound imaging involves exposing part of the body to high-frequency sound waves to produce pictures of the inside of the body. Because the ultrasound images are captured in real-time, they can show the structure and movement of the body's internal organs, as well as blood flowing through the blood vessels. Ultrasound during pregnancy can be used for various reasons:

- To establish the presence of a living embryo/fetus
- To estimate the age of the pregnancy
- To screen for congenital abnormalities
- To evaluate the position of the baby
- To evaluate the position of the placenta
- To determine if there are multiple fetuses
- To determine the amount of amniotic fluid around the baby
- To check for the opening or shortening of the cervix

Ultrasound may not be able to detect fetal anomalies with 100% accuracy. In order to diagnose a condition or completely rule it out, a more invasive test such as Chorionic Villus Sampling (CVS) or Amniocentesis must be performed.

Ultrascreen

FIRST TRIMESTER SCREENING FOR DOWN SYNDROME AND TRISOMY 18

- What are Down Syndrome and Trisomy 18
    - Down Syndrome (Trisomy 21) and Trisomy 18 are chromosomal disorders that cause mental retardation and birth defects. Infants with Down Syndrome have an extra chromosome #21 which causes mental retardation and various medical problems involving the heart, digestive tract, and/or other organ systems.
    - Trisomy 18 (extra chromosome # 18) is a more severe disorder which causes profound mental retardation and severe birth defects in many organ systems. Few infants with Trisomy 18 survive more than a few months. The risk of having a baby with chromosomal abnormality increases with age, although a young woman is at risk at well.
- What can Ultrascreen tell me about my pregnancy?
    - Ultrascreen in **not** a diagnostic test, which means it cannot tell you whether your baby has Down Syndrome or Trisomy 18. Instead, the screening provides a probability, or risk, which is based on three criteria: your age, information obtained on ultrasound and blood work. The screening results can either alert you and your healthcare provider that your baby is at an increased risk for one of these chromosome disorders or reassure you that your baby is at a lower risk for these conditions.

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- How is Ultrascreen performed?
    - The screening requires an ultrasound and maternal blood work performed between 11 and 13 weeks of pregnancy. During the ultrasound, the skin fold along the back of the baby's neck, called the nuchal translucency, will be measured. A maternal blood sample is used to analyze two chemicals called free beta-human chorionic gonadotropin (hCG) and pregnancy associated plasma protein-A (PAPP-A) circulating throughout your bloodstream. If there is extra fluid behind the baby's neck and the hCG and PAPP-A levels are abnormal, the baby is at increased risk of having Down Syndrome or Trisomy 18. This test combines your age-related risk with the NT measurement and blood work to determine your individual risk.
- How accurate is Ultrascreen?
    - Because this is a screening test, a positive test result shows an increased risk but does not mean that your baby has a problem. On the other hand, a result that shows a decreased risk cannot entirely exclude the possibility that the baby is affected by one of these conditions. The only way to be completely sure that the baby is not affected is to get a diagnostic test such as an amniocentesis. Ultrascreen detects about 85% of pregnancies in which the baby has Down syndrome or Trisomy 18. If the nuchal translucency is measured without the blood work, the detection rate decreases to 75%. Additionally, this screening test is not designed to provide information about the possibility of other chromosomal defects, or causes of mental retardation.

Should I still have the second trimester serum screening (Quad Screen)?

- The Quad Screen is performed between 16 and 20 weeks of pregnancy. It measures four chemicals: AFP, hCG, estriol, and Inhibin-A in the mother's blood. Like the first trimester screening, results from a second trimester quad screen can be used to statistically adjust a woman's age-related risk for Down Syndrome and Trisomy 18. In addition, the AFP portion can identify an increased risk for open neural tube defects such as spina bifida, which is not included in the first trimester screen. The quad screen detects about 80% of pregnancies in which the baby has Down Syndrome an 75-80% of pregnancies affected by spina bifida.

- What if the screening shows an increased risk for one or the conditions?
    - If the screening risks indicate that your baby is at an increased risk for either Down Syndrome or Trisomy 18, this does not mean that your baby definitely has one of these conditions. A genetic counselor is available to go over your result and to discuss additional testing options such as chorionic villus sampling (CVS) and amniocentesis. CVS and amniocentesis are diagnostic tests that can tell you with greater than 99% accuracy whether or not a baby has a chromosomal abnormality. Additionally, extra fluid behind the baby's neck (abnormally large nuchal translucency) is known to be associated with other birth defects like congenital heart defects and skeletal problems.

Non-Invasive Prenatal Test (NIPT)

Non-invasive prenatal testing (NIPT) uses a blood sample from a pregnant woman to analyze DNA from the baby that is circulating in the mother's blood. NIIPT screens for certain chromosome conditions including Down syndrome, trisomy

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18, trisomy 13 and monosomy x. It can also determine fetal sex. NIPT is recommended for pregnant women who have a higher chance for chromosome abnormalities because of their age, family history or other abnormal screening tests such as ultrasound or blood tests. The American College of Obstetricians and Gynecologists recommends these women be offered a NIPT like The Panorama Test. The Panorama test can detect most cases of extra or missing chromosomes tested but it cannot detect all of them. Studies have shown that the Panorama test can detect >99% of babies with Down syndrome, trisomy 18 and trisomy 13 and approximately 92% of babies with monosomy X. The Panorama test can be performed as early as 9 weeks of pregnancy. Result may not always be reported and will require the test to be repeated. The chance of not obtaining results can be decreased if a cheek swab from the father is also collected. If you have a high risk result, your midwife will suggest genetic counseling. You will also be provided with information about diagnostic testing, such as amniocentesis or CVS.

Quad Screen

The Quad Screen is a screening test done between 15 and 20 weeks gestation. This test measures the levels of Alpha Fetoprotein (AFP), Human Chorionic Gonadotropin (HCG), Estriol and Inhibin-A in the maternal blood. This test may identify infants at risk for Down Syndrome (DS) or open neural tube defects (openings to the brain and/or spinal cord). This test can detect about 80% of infants affected by DS, about 75-80% of pregnancies affected by spina bifida (opening to the spinal cord), an nearly 95% of those affected by anencephaly (a fatal condition in which the baby's brain and skull are severely underdeveloped). It is important to remember that this is only a screening test and the only way to know if an infant is affected by any of these disorders is by getting an amniocentesis.

Chorionic Villus Sampling (CVS)

CVS is a procedure in which a thin tube is guided through the cervix or a needle is inserted into the uterus to obtain a sample of part of the placenta called chorionic villi. Because these wispy projections have the same genetic makeup as your baby, this test can detect various genetic and chromosomal disorders. This test is usually performed between 10 and 12 weeks of pregnancy and carries around the same risk of miscarriage as amniocentesis (1/200). Final results are usually available in 7-10 days, although, unofficial results are often ready in 2-3 days. Having a diagnostic test in the first trimester allows the woman to make choices about her pregnancy if she receives an abnormal test results.

Amniocentesis

Amniocentesis is a procedure in which a small amount of amniotic fluid is removed by inserting a small needle through a pregnant women's abdomen to obtain a sample for genetic studies. This procedure is usually done between 15 and 20 weeks gestation and uses ultrasound guidance to prevent any harm to the baby. The fluid will be sent to the lab for chromosomal analysis and AFP levels. The conclusive results are generally ready in 2 weeks. The risk of miscarriage due to amniocentesis is often cited to be between 1/400 and 1/200 cases. However, there are some studies which cite the risk to be much lower. This test does not guarantee the birth of a completely healthy baby. There are certain congenital or hereditary abnormalities that cannot be detected by amniocentesis.