NOVA GROUP FOR WOMEN

FIRST TRIMESTER SCREENING FOR DOWN SYNDROME, TRISOMY 13 & 18

Maternal Serum Screening & Nuchal Translucency (NT) Ultrasound

What are Down Syndrome Trisomy 13 and 18.
Down Syndrome, Trisomy 13 and 18 are chromosomal disorders that cause mental retardation and birth defects. Babies with Down Syndrome have an extra chromosome #21 (Trisomy 21), which causes mental retardation and various medical problems involving the heart, digestive tract, and/or other organ systems. Trisomy 13 & 18 (having an extra chromosome 13 or 18 are more severe disorders, which cause profound mental retardation and severe birth defects in many organ systems. Few babies with Trisomy 13&18 survive more than a few months. Anyone can have a baby with Down Syndrome, Trisomy 13 or 18, however the chances increase with the mother's age.

What can first trimester screening tell me about my pregnancy?
First trimester screening is not a diagnostic test, which means it cannot tell you whether your baby has Down Syndrome, Trisomy 13 or 18. Instead, the screening provides a probability that the baby might have Down Syndrome, Trisomy 13 or 18. This probability, or chance, is based on three criteria: your age, information obtained on an ultrasound, and bloodwork. The screening results can either alert you and your doctor that your baby is at an increased risk for one of these chromosomal disorders or be reassuring that your baby is at a lower risk for these conditions.

How is First Trimester Screening performed?
This screening requires an ultrasound and maternal bloodwork performed between 11 4/7 – 13 6/7 weeks of pregnancy. The ultrasound will confirm how far along your pregnancy is. In addition, a measurement of the skin fold along the back of the baby's neck, called the nuchal fold, will be taken. 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), which are found in the blood of all pregnant women. In some pregnancies when the baby has Down Syndrome, Trisomy 13 or 18, there is extra fluid behind the baby's neck and the HCG and PAPP-A results are higher or lower than average. Combining your age-related risk with the NT measurement and bloodwork provides you with a risk for Down Syndrome and a risk for Trisomy 13&18.

How accurate is First Trimester Screen?
Because this is a screening test, a positive result (one that shows an increased risk) does not mean that your baby has a problem, only that further diagnostic tests are options for you to consider. Also, a negative or normal result (one that shows a decreased risk) does not mean that the baby will not have Down Syndrome, Trisomy 13 or 18. The first trimester screen detects about 85% of pregnancies in which the baby has Down Syndrome, Trisomy 13 or 18. A nuchal translucency ultrasound can be performed without measuring HCG and PAPP-A ; however, the detection rate of Down Syndrome, Trisomy 13 or 18 are reduced to about 70%. Finally, this screen is not designed to provide information about the possibility of other chromosome conditions, nor about many other genetic syndromes, genetic disorders, birth defects, or causes of mental retardation.

Should I still have a second trimester screen?
The AFP screen is performed between 15-20 weeks. The AFP and estriol (UE3) in the mother's blood identifies pregnancies at an increased risk for Open Neural Tube defects such as Spina Bifida, which first trimester screening does not include.

What is the cost of First Trimester Screening?
The screening involves a charge for the ultrasound examination and the blood test. These charges may not be covered by your insurance. You may want to check with your insurance company to see if your specific plan will cover these services. As with any service, if your insurance company requires pre-authorization, we will be happy to assist you.

How will I receive my First Trimester Screening results?
We will receive the results and will contact you with your results.

What if the screening shows an increased risk for one of the conditions?
If the screening results indicate that your baby is at an *increased* risk for either Down Syndrome or Trisomy 13 or 18, this does not mean that your baby necessarily has one of these conditions. We will go over your results and discuss additional testing options such as chorionic villus sampling (CVS), amniocentesis, and noninvasive chromosomal blood tests. CVS and amniocentesis are diagnostic tests that can tell you with greater than 99% accuracy whether or not a baby has any chromosome abnormality such as Down Syndrome, Trisomy 13 or 18. Also, extra fluid behind the baby's neck (a larger than expected nuchal translucency) is known to be associated with other birth defects like congenital heart defects and skeletal problems.

How do I schedule an appointment for First Trimester Screening?
We can give you more information about scheduling this screening procedure. You will be referred to either:

1. Perinatal Associates
   Antenatal Testing Ctr.
   3300 Gallows Road
   Falls Church, VA 22042
   (703) 776-6654

1. Genetics & IVF Institute
   3020 Javier Road
   Fairfax, VA 22031
   (703) 698- 7355

3. Greater Washington
   Maternal Fetal Medicine
   and Genetics
   3025 Hamaker Ct. #330
   Fairfax, VA 22031
   (571) 730-4587