



Prenatal Diagnosis and Treatment Center

First Trimester Screening for Down syndrome and Trisomy 18:

Maternal Serum Screening & Sonogram for Fetal Nuchal Translucency & Nasal Bone Assessment

Information for Physicians/Healthcare Professionals: First trimester screening is a means by which the predicted risk for having a child with Down syndrome, trisomy 13, or trisomy 18 can be adjusted using sonographic measurement of the fetal nuchal translucency, sonographic analysis of the fetal nasal bone, maternal serum hCG (human chorionic gonadotropin), and maternal serum PAPP-A (pregnancy associated plasma protein-A) between 11-1/7 and 13-6/7 weeks. This combined with maternal age is thought to have a detection rate of approximately 95% for Down syndrome and trisomies 13 & 18. The false positive rate is about 5-7%. For your information, a nuchal translucency sonogram performed without maternal hCG and PAPP-A will also modify the age-related risk, but provides a lower detection rate for Down syndrome and trisomies 13 & 18 (about 70%). Measurement of the fetal nuchal translucency and analysis of the fetal nasal bone must be done in accordance with very exacting, strict guidelines. Our center is certified by the Fetal Medicine Foundation, the nationally and internationally recognized certifying organization in London, England. In fact, we have been performing these scans for several years now for patients at increased risk for chromosomal abnormalities because of advanced maternal age or other factors.

Just as for second trimester maternal serum screening, we are now offering this test to pregnant women of any maternal age. Your patient will be scheduled for a sonogram and phlebotomy between 11-1/7 and 13-6/7 weeks at the Johns Hopkins Hospital Prenatal Diagnosis and Treatment Center. Consent for testing will be obtained from the patient by our office at the time of her appointment. The patient should have a full bladder for the sonogram. The blood specimen will be sent to NTD Laboratories and the results of the screening will be reported directly to your office by the laboratory within about one week of testing. Those patients identified at increased risk for fetal Down syndrome or trisomies 13 or 18 would then be offered genetic counseling and would have the option of chorionic villus sampling (CVS) or amniocentesis to analyze the fetal chromosomes. An increased nuchal translucency has also been associated with other fetal abnormalities, like congenital heart defects, diaphragmatic hernia and skeletal dysplasias. If an increased nuchal translucency is found, we will counsel your patient and explain her options for further evaluation during her visit.

An information sheet describing first trimester screening is provided for you to duplicate and give to your patients.

We are pleased to be offering this newly expanded service to you and your patients. If you are interested, please contact one of our genetic counselors at **(410) 955-3091**. We will help you obtain an NTD Laboratories physician code number so that the test results can be reported to your office. Please do not hesitate to contact one of our genetic counselors if you have any questions.