Combined First Trimester Screening for Down Syndrome and Trisomies 18 and 13

Maternal Serum Screening & Nuchal Translucency/Nasal Bone Sonogram

1. **What are Down syndrome, trisomy 13 and trisomy 18?**
   Down syndrome and trisomies 18 and 13 are chromosomal disorders. Babies with Down syndrome have an extra 21st chromosome (trisomy 21), which causes intellectual disability and may have various medical problems involving the heart, digestive tract and/or other organ systems. Trisomy 18 (having an extra 18th chromosome) and trisomy 13 (having an extra 13th chromosome) are more severe disorders which cause profound intellectual disability and birth defects in many organ systems. Few babies with trisomies 13 or 18 survive more than a few months. Anyone can have a baby with these chromosome abnormalities, however, the chance increases with the mother's age.

2. **What can Combined First Trimester Screening tell me about my pregnancy?**
   First trimester screening is <u>not</u> a diagnostic test, which means it cannot tell you whether your baby has Down syndrome, trisomy 13 or trisomy 18. Instead, the screening provides the probability that the baby might have Down syndrome, trisomy 13 or trisomy 18. This probability, or chance, is based on three criteria: your age, information obtained on a sonogram (ultrasound) and blood work. The screening results can either alert you and your doctor that your baby is at an increased risk for one of these chromosome disorders or be reassuring that your baby is at a lower risk for these conditions.

3. **How is Combined First Trimester Screening performed?**
   This screening includes a sonogram and maternal blood work performed between 11-4/7 and 13-6/7 weeks of pregnancy. The sonogram will confirm how far along your pregnancy is. In addition, a measurement of the fluid underneath the skin along the back of the baby's neck, called the nuchal translucency (NT), will be taken. The sonogram will also try to determine if your baby's nasal bone is able to be seen. A maternal blood sample will be drawn to analyze three chemicals called free beta-human chorionic gonadotropin (hCG), pregnancy associated plasma protein-A (PAPP-A) and alpha-fetoprotein (AFP), which are found in the blood of all pregnant women. In some pregnancies when the baby has Down syndrome, trisomy 13 or trisomy 18, there is extra fluid behind the baby's neck and/or the hCG, PAPP-A and AFP results are higher or lower than average. While a baby's nasal bone may be absent in some pregnancies with a chromosome abnormality, most babies with this finding are normal. Combining your age-related risk with the NT measurement, nasal bone data and blood work provides you with one risk figure for Down syndrome and one risk figure for trisomy 13 or trisomy 18. Your obstetrician will receive the results of your screening results from NTD Laboratories within approximately one week.

4. **How accurate is Combined First Trimester Screening?**
   Because this is a screening test, a positive result (showing an increased risk) does *not* mean that your baby necessarily 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 necessarily mean that the baby will not have a chromosome abnormality. The first trimester screen's detection rate is approximately 96% for pregnancies in which the baby has Down syndrome, and is somewhat lower for pregnancies with trisomy 13 or trisomy 18. A nuchal translucency sonogram can be performed without the bloodwork; however, the detection rate is reduced to about 70%. This screen is not designed to provide information about the possibility of other chromosome conditions, but does have limited utility for some other genetic syndromes, genetic disorders and birth defects.

5. **Should I still have second trimester screening?**
   The second trimester maternal serum screening test, also known as the "quad screen," is performed between 16 and 20 weeks and measures four chemicals in the mother's blood. Like first trimester screening, results from a second trimester "quad screen" are used to statistically adjust a woman's age-related risk for Down syndrome and trisomy 18 (not trisomy 13). It is not recommended that quad screening be performed if combined first trimester screening has already been performed. The AFP portion of the screen in the second trimester can help identify pregnancies with open neural tube defects such as spina bifida, however, a second trimester sonogram is a more sensitive way to detect open spina bifida.

6. **How is Combined First Trimester Screening billed?**
   The screening involves a charge for the ultrasound examination and a charge for the blood work. The blood work will be performed by NTD Laboratories who will bill your insurance $160; you may be responsible for a co-pay. The ultrasound examination will be billed by Johns Hopkins; this charge varies over time. As with any service, if your insurance company requires pre-authorization, you are responsible for obtaining it from your OB or primary care physician prior to the appointment.

7. **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 18 or 13, this does NOT mean that your baby necessarily *has* one of these conditions. A genetic counselor is available to go over your result and to discuss additional screening and testing options such as cell free DNA screening, 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 chromosome abnormality. 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.

8. **How do I schedule an appointment for Combined First Trimester Screening?**
   You can call the Prenatal Genetics Service at (410)-955-3091 to arrange an appointment or if you have questions regarding your screening and testing options.