JOHNS HOPKINS
MEDICINE

FIRST TRIMESTER SCREENING FOR DOWN SYNDROME AND TRISOMIES 13 & 18

Maternal Serum Screening & Nuchal Translucency/Nasal Bone Sonogram

- **What are Down syndrome, trisomy 13, and trisomy 18?**

Down syndrome and trisomies 13 & 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 18 (having an extra chromosome #18) and trisomy 13 (having an extra chromosome #13) are more severe disorders which cause profound mental retardation and severe 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.

- **What can 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 a 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 bloodwork. 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.

- **How is First Trimester Screening performed?**

This screening includes a sonogram and maternal blood work performed between 11 4/7 – 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 determine if your baby's nasal bone is present or absent. 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 trisomy 18, there is extra fluid behind the baby's neck and/or the hCG and PAPP-A results are higher or lower than average. Additionally, a baby's nasal bone may be absent in some pregnancies with a chromosome abnormality. 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 your screening results from NTD Laboratories within approximately one week.

- **How accurate is First Trimester Screening?**

Because this is a screening test, a positive result (showing 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 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 higher for pregnancies with trisomy 13 or trisomy 18. A nuchal translucency sonogram can be performed without measuring hCG and PAPP-A; however, the detection rate is 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 second trimester screening?**

The second trimester maternal serum screening test, also known as the "triple screen" or "quad screen", is performed between 16-20 weeks. Both of these screens measure chemicals in the mother's blood. Like the first trimester screening, results from a second trimester "triple screen" or "quad screen" can be used to statistically adjust a women's age-related risk for Down syndrome and trisomy 18 (not trisomy 13). In addition, the AFP portion of the screen can identify pregnancies at an increased risk for open neural tube defects such as spina bifida, which first trimester screening does not include. While these screens are certainly an additional screening option after having first trimester screening, it is unclear how to interpret results of the second test in light of the first. Currently most laboratories do not combine results from first trimester and second trimester screening into one overall adjusted risk figure.

- **How is 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.

- **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. 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 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.

- **How do I schedule an appointment for First Trimester Screening?**

You can call the Prenatal Genetic Counseling office at (410)-955-3091 to arrange an appointment or if you have questions regarding your two screening options. One option, called "instant risk assessment", involves collecting your own blood at least 7 days prior to your sonogram appointment, which allows your results to be reported to you on the day of your sonogram. The second option involves the blood draw and sonogram on the same day, and your results will be reported back to your obstetrician within one week of your appointment with us.