EMORY UNIVERSITY SCHOOL OF MEDICINE

Department of Human Genetics
Division of Medical Genetics
www.genetics.emory.edu

First & Second Trimester Screening Tests

There are maternal serum screening tests now available in both the first and second trimesters to determine if pregnant women may have an increased risk for certain fetal chromosome abnormalities such as Down syndrome and trisomy 18. The information below describes first trimester screening.

The first trimester screen is relatively new and consists of:

- a maternal blood test measuring two analytes: free beta-hCG (human chorionic gonadotropin) and PAPP-A (pregnancy associated plasma protein A), ***and***
- a fetal ultrasound measurement of an area on the back of the fetal neck termed nuchal translucency (NT). This screening combination is thought to detect approximately 80-90% of Down syndrome and trisomy 18 pregnancies and is performed between the 10th and 13th weeks of pregnancy.

"Nuchal Translucency" or NT refers to a measurement taken on an ultrasound of a pregnancy, usually between 10 and 13 weeks gestation. During fetal development, a layer of fluid on the back of the body normally exists between the skin and underlying tissue. In certain disorders, such as chromosome abnormalities (e.g., Down syndrome), heart defects, and others, there is more fluid in this layer. This layer is translucent on ultrasound and its thickness is measured at the level of the fetal neck. Approximately 5% of normal pregnancies have increased nuchal translucency, so this measurement alone *cannot* determine whether a problem definitely exists or not.

Not all physicians use both criteria above, and *they are not 100% accurate* on their own. It is important to discuss with your physician which types of first trimester screening criteria are being utilized, along with their accuracy. **Second trimester MSAFP screening for open-neural tube defects is recommended for women who have the first trimester serum screen and nuchal translucency.**

Second trimester maternal serum screening for open-neural tube defects, Down syndrome, and trisomy 18 is considered standard of care at this time by the American College of Obstetrics & Gynecology (ACOG). It is performed by measuring maternal serum alpha-fetoprotein (MSAFP), human chorionic gonadotropin (hCG), and estriol (uE3) with or without dimeric inhibin A (DIA) between 15 and 20 weeks of pregnancy. This test can detect:

- - 85% of pregnancies with open neural tube defects (e.g. spina bifida, anencephaly)
- -70% of pregnancies with Down syndrome (85% in women over 35)
- -approximately 70% of pregnancies with trisomy 18.

Both types of serum screening have little value in detecting most other chromosome abnormalities. If the values from the maternal serum screening test indicated an increased risk for Down syndrome or trisomy 18, additional testing would be necessary to diagnose either condition. Since women who are 35 years of age and over have a higher risk for a fetus with a chromosome condition based on their age, they should **not** depend completely on serum screening (either first or second trimester) for detection of chromosome conditions in their pregnancies.