

First Trimester Screening and Genetic Counseling

By Karen Stebner, C.G.C. and Philip Buchanan, PhD

Only 11 weeks pregnant, Laura excitedly views her baby for the first time by ultrasound. The sonographer employs a standard method for birth defect's screening, and measures a nuchal translucency of 5 mm. Laura sees the concerned look on the sonographer's face and anxiously asks, "Is there something wrong with my baby?" Her obstetrician explains that an increased nuchal translucency may be an indication of a cardiac defect, skeletal abnormality, or chromosome abnormality. Laura's heart sinks.

First trimester screening by sonographic measurement of nuchal translucency (NT), the echo-free space at the back of the fetal neck, in conjunction with biochemical analysis of two proteins in the mother's blood, freeBeta hCG and PAPP-A, detects 91 percent of fetuses with Down syndrome and Trisomy 18. Genetic screening is offered to all low-risk women to identify those women at increased risk for an abnormality. If a woman's screening test shows an increased risk, she is offered prenatal diagnosis. Several diagnostic tests are available including chorionic villus sampling (CVS) at 10-13 weeks, early amniocentesis at 11-14 weeks, and traditional amniocentesis at 15 or more weeks of gestation. These tests are diagnostic for chromosome abnormalities and safe.

Laura's biochemical screening test shows an increased risk for Down syndrome. She meets with a genetic counselor, who explains the implications of her screening tests. After discussion and careful consideration, Laura decides to proceed with amniocentesis to determine if the baby has Down syndrome.

"Laura, I have the results of your amniocentesis," the counselor says, facing Laura in her office. "I'm sorry; the test shows conclusively that your baby has Down syndrome." Laura cries openly, simultaneously experiencing grief and relief from uncertainty. After an emotional discussion with the genetic counselor regarding whether or not to continue the pregnancy, Laura and her husband decide to continue. At 20 weeks gestation, Laura spontaneously miscarries her baby.

Genetic counseling throughout the various stages of genetic screening and diagnosis is imperative and beneficial. A genetic counselor's role is to educate the patient about her medical care and aid in the emotional process that is an integral part of testing. The genetic counselor provides informational and emotional support. After diagnosis of an abnormality in one pregnancy, patients are counseled about the risk for recurrence and are offered prenatal diagnosis in subsequent pregnancies.

A year after the loss of her baby, Laura decides to become pregnant again. She has an early amniocentesis so she can attain results as early in the pregnancy as possible. The test shows normal chromosomes, and Laura gives birth to a healthy baby girl.

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