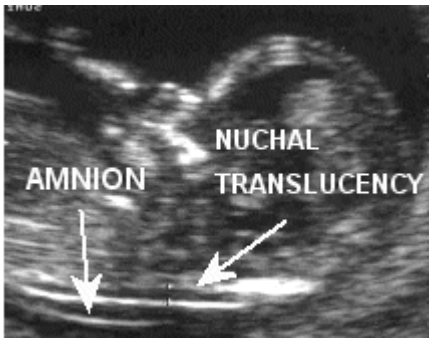
San Gabriel Valley Perinatal Newsletter

March 1, 2006 Vol.4, No. 1

First Trimester Screening

Currently, women in California are offered a blood test known as the triple marker test or expanded AFP at about 16 weeks into their pregnancy to screen for neural tube defects, Down syndrome, and trisomy 18. If the test is positive the woman may then undergo amniocentesis to confirm the diagnosis. However, screening for Down syndrome and trisomy 18 may be performed **as early as 11-13 weeks** from the first day of the last period using a combination of ultrasound and maternal blood testing called first trimester screening. **First trimester screening is offered by San Gabriel Valley Perinatal Medical Group.**

The first trimester screen uses ultrasound to measure the thin layer of fluid beneath the fetal skin in the area of the fetal neck called the nuchal translucency (NT). In addition, maternal blood is measured for levels of a protein called pregnancy-associated plasma protein A (PAPP-A) and a hormone known as human chorionic gonadotropin (HCG). In fetuses with Down syndrome, the nuchal translucency measurement is increased, HCG is increased and PAPP-A is decreased [1].



The sonogram above shows a fetus with a normal nuchal translucency. Increased nuchal translucency has been associated with several birth defects including Down syndrome.

How Accurate Is First Trimester Screening?

First trimester screening can detect up to 85% of babies with Down syndrome and trisomy 18. Up to 15% of Down syndrome and trisomy 18 cases will not be detected. The test incorrectly reports an increased risk for Down syndrome (a false positive result) approximately 5 % of the time [2-4] .

What Do the Results Mean?

The first trimester screen is not diagnostic. The results are an estimate of the risk for Down syndrome or trisomy 18 in a particular fetus.

A positive test result indicates an increased risk for Down syndrome above that of a 35 year old at the same gestational age (1/240-1/260). A positive result for trisomy 18 indicates a risk of 1/150 (0.67%) or greater.

If the results are positive the genetic counselor will explain the results, and discuss options of further testing such as chorionic villus sampling.

If the results are negative, then maternal serum AFP screening for neural tube defects at 16 weeks and a high-resolution ultrasound at 18 to 20 weeks are recommended.

Women over age 34 or those with additional risk factors may still choose to have chorionic villus sampling or amniocentesis to test for other abnormalities.

Health Insurance Coverage

The cost of screening includes genetic counseling, ultrasound, and a laboratory fees.
Patients wishing to have first trimester screening should call their insurance carriers to verify coverage for the first trimester screen.

Current procedure terminology codes (CPT codes) for first trimester screening:

Counseling: CPT code 99243
Ultrasound: CPT code 76801 (for a singleton pregnancy)
Laboratories: CPT codes: 84702 AND 84163).

The procedures are covered by most insurance carriers. Patients also have the option to self pay for first trimester screening.

REFERENCES

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