EARLY RISK ASSESSMENT / FIRST LOOK

WHAT IS THE FIRST LOOK TEST?

The First Look test is a method of noninvasive screening performed early in pregnancy, between 12 and 13.6 weeks gestation, to look for problems in the developing fetus. Such problems can include birth defects, usually of the heart, chromosome abnormalities such as Down syndrome and, rarely, inherited diseases. This screening starts with an ultrasound measurement of the fluid at the nape of the fetal neck (the nuchal translucency). A maternal blood sample (for PAPP-A and total hCG) is also needed to specifically determine the chance a chromosome abnormality is present. A woman may want to consider this screening:

- if she is at increased risk for a child with a birth defect because of (1) a family member with a birth defect (herself, father of baby, or prior child), (2) she has particular medical conditions (such as diabetes) or (3) she has exposures to medications suspected of causing congenital malformations;
- if she is at increased risk for fetal chromosome abnormalities due to (1) maternal age > 35 years, or (2) a prior pregnancy with a chromosome abnormality;
- if she would like information early in her pregnancy about her risk of a chromosome abnormality.

HOW LONG DOES IT TAKE TO GET RESULTS?

In 1-3% of women, the ultrasound alone will indicate an increased risk. As risk assessment for chromosomal abnormalities also requires a blood sample, these final results will be sent to your doctor in approximately one week.

WHAT IF MY RESULTS INDICATE AN INCREASED RISK?

Remember that as a screening tool, the goal of the First Look test is to identify those women whose pregnancy is at increased risk for a birth defect, chromosome abnormality or inherited disease. If your risk for a birth defect is increased, further imaging studies will be recommended. If your risk for a chromosome abnormality is increased, this will involve directly testing the fetal chromosomes by either free fetal DNA testing (a more accurate screening blood test), chorionic villus sampling (at 11-14 weeks) or amniocentesis (>15 weeks). Most of the time, direct testing will reveal normal chromosomes. This does not mean that the screening process is a failure or someone has made a mistake. As opposed to CVS or amniocentesis, the screening process does not give a specific diagnosis, only an estimate of risk. Only CVS or amniocentesis- not even the free fetal DNA blood test- can diagnose a chromosome abnormality; however either procedure has a small (1% or less) risk of miscarriage. The decision to undertake further testing by CVS or amniocentesis is individual. You will be given a risk (the chance the baby has a chromosome problem) based on your ultrasound and your blood test. You can decide whether the risk is large enough to warrant CVS or amniocentesis. If requested, you can meet with a genetic counselor to assist with any questions.