Translucency/1st Screen Testing

What is Combined Screening ?

Combined screening is a new prenatal test for Down syndrome and other birth defects in pregnancy. Because it is a screening test, it cannot tell for certain if a baby has Down syndrome. However, the test can identify those women who have an increased risk, so that they can be offered further testing, such as chorionic villus sampling (CVS) or amniocentesis.

Does Combined Screening Detect All Pregnancies with Down Syndrome?

No. Combined screening can detect 85 percent to 90 percent of Down syndrome fetuses. This means that about 15 percent of Down syndrome fetuses will not be identified. Only diagnostic testing, such as CVS or amniocentesis, can detect *all* cases of Down syndrome and other chromosome problems.

Does Combined Screening Identify Other Birth Defects?

Combined screening also can identify about 90 percent of fetuses with a very serious chromosome abnormality known as trisomy 18. However, unlike the expanded alpha-fetoprotein (AFP) test, which is performed later in pregnancy, combined screening cannot detect birth defects such as spina bifida. Women who have combined screening should consider screening for spina bifida with either a second trimester blood test or ultrasound.

How Is Combined Screening Performed?

In combined screening, a sample of a woman's blood is taken between 9 and 14 weeks of pregnancy. Between 111/2 and 14 weeks, an ultrasound examination is performed to measure the amount of fluid at the back of the baby's neck, known as the nuchal translucency. The results of the blood test, the nuchal translucency (NT) measurement, and the motherôs age are then combined to determine the risk for Down syndrome.

When Are the Results Available?

Many women choose to have the blood test done first, and schedule the NT ultrasound examination for about one week later. That way, combined screening results can be determined as soon as the NT ultrasound is completed, and explained to you in person while you are in the Prenatal Diagnosis Center. If you have the NT ultrasound and blood test on the same day, the results will be called to you about one week later.

What If My Combined Screening Test Indicates a High Risk?

If the results are positive, indicating an increased risk of Down syndrome, this does not mean the baby definitely has a problem. However, it does mean that the risk is high enough that you might wish to consider definitive testing with either CVS or amniocentesis. Our genetic counselors and doctors are available to answer questions you may have about what the results mean, as well as the options available for further testing.