Down syndrome, also known as trisomy 21, is caused by an error in cell division early in a pregnancy that results in a fetus having an extra chromosome (47 instead of 46). One in every 800 to 1,000 live births is a child with Down syndrome, the most common cause of mental retardation. Eighty percent of children born with Down syndrome are born to women younger than 35 years old. However, the incidence of births of children with Down syndrome increases with the age of the mother.

Like Down syndrome, trisomy 18 results from an extra chromosome. But, it is not as common as Down syndrome, occurring in roughly one in 6,000 live births. Only half of babies with trisomy 18 live longer than a week and 90 percent do not live beyond 1 year old. They typically have birth defects, especially congenital heart disease, and developmental disabilities.

This normal NT noninvasive ultrasound reveals normal fetal neck thickness in the first trimester. Increased fetal neck thickness is a sign of possible chromosomal abnormalities.

+ indicates nuchal measurement
Early Screening in Pregnancy (ESP)

Cleveland Clinic Maternal-Fetal Medicine at Hillcrest Hospital offers pregnant women a new, Early Screening in Pregnancy (ESP) program that helps detect chromosomal abnormalities, such as Down syndrome, earlier, with greater accuracy and less invasively than ever before possible. Preliminary findings suggest that ESP also helps identify obstetrical risk and poor outcomes.

ESP employs Nuchal translucency (NT), a sophisticated ultrasound that measures the thickness of the fetal neck. Increased thickness may indicate a higher risk for chromosomal abnormalities and certain birth defects.

This risk assessment poses no danger to the fetus and offers detection rates higher than traditional testing. Maternal-fetal medicine specialists at Cleveland Clinic use the test results and medical history to predict the occurrence of Down syndrome and certain other chromosomal (genetic) abnormalities in a fetus.

Cleveland Clinic maternal-fetal medicine specialists trained and certified in the NT measurement procedure conduct the tests and provide follow-up counseling, ensuring both quality and reliability.

Improved Accuracy, Earlier Detection

Until now, the best noninvasive assessment of the risk for chromosomal abnormalities was the age of the mother combined with quad screening. This screening was performed at 15 to 20 weeks of gestation, well into the second trimester. Our new tests may be performed earlier – at 11 or 12 weeks into the pregnancy, allowing a woman more time to plan.

With the new early screening tests, detection rates are as high as 95 percent, with a false-positive rate of less than 5 percent. Previously, blood testing detected abnormalities only 65 to 81 percent of the time with a false-positive rate of 5 to 8 percent. (False-positive means a normal fetus was incorrectly identified as abnormal.)

The blood tests used in the first trimester measure PAPP-A (pregnancy-associated plasma protein A) and beta hCG (human chorionic gonadotropin), proteins made by the fetus and placenta. The amount of these proteins may be abnormal when a fetus is at risk for Down syndrome. Combined early testing reduces the need for further assessments, such as chorionic villus sampling or amniocentesis, which are invasive and carry a small risk to the fetus. Perhaps the most significant advantages of early screening are the elimination of the risk of pregnancy loss caused by these tests, and earlier reassurance.

Early screening also helps determine accurate due dates and detect multiple fetuses. Early screening also can be an indicator of low birth weight, heart abnormalities, premature rupture of membranes, pre-eclampsia and other complications as well.

Early Screening Program Options

The Cleveland Clinic Early Screening in Pregnancy Program offers testing options that best suit each woman’s particular needs. Options range from first- or second-trimester screening with nuchal translucency or a combination of both, maternal blood testing alone, ultrasound alone and several other choices. Each option has known success and false-positive rates that we can discuss with you.

When an ESP procedure yields an abnormal measurement (a positive result), we may recommend diagnostic testing (chorionic villus sampling or amniocentesis) to determine if a fetus is affected. Most women who have a positive screening will still have a normal baby.

Is Early Screening Right for You?

ESP is optional. Early noninvasive screening can provide useful information that will help you plan for the rest of your pregnancy and prepare for the future. The decision requires careful thought and a thorough understanding of your options. Please don’t hesitate to ask us any questions.

Screening Process

The ESP program offered at Hillcrest Hospital includes testing, results, counseling and follow-up testing (including chorionic villus sampling and amniocentesis). Testing and counseling may take up to 45 minutes. We will call you with your results in four days.

Costs/Insurance Coverage

Screening in the first trimester is so new, and some insurers may not yet cover testing or counseling. We recommend that you check with your insurance provider. Please call us with any questions.