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Combined First Trimester Prenatal Screening Is a Better Test for Down syndrome

On October 9th, the *Wall Street Journal* carried an article, “A Better Test for Down Syndrome”. Down syndrome (DS) affects about 1 in 800 babies and is the most common chromosomal birth defect. The “new” combination of blood tests and ultrasound can detect fetal anomalies earlier and more accurately than other screening tests currently offered. The National Institute of Child Health and Development Study involving 8,215 women from a dozen US medical centers confirmed the findings already in practice in the U.S., Britain and 60 other countries. The study was published in the *New England Journal of Medicine* on October 9, 2003 (Vol. 349 15 p.1405-1413). It found that the first trimester method at 11 – 13 weeks detected 85% of the Down syndrome fetuses while comparatively second trimester screening typically only detects 60%.

Second trimester screening is currently the most common means to identify high risk women who would most benefit from a diagnostic test like amniocentesis, to determine whether a fetus has Down syndrome. Chorionic villi sampling (CVS) is not an option for the patient because second trimester testing is performed too late for this procedure, another benefit to performing first trimester screening. Dr. Mark Evans, president of the Fetal Medicine Foundation of America, said “the study will cause a shift from second trimester screening to this method.” The Wall Street article quotes Dr. Evans as saying, “In five years, I have no doubt that this will be the predominant form of diagnosis and screening.”

Earlier testing has lots of advantages including allowing expectant mothers and their physicians more options. As a result, studies have shown patients prefer first trimester screening. Dr. Ronald Wapner, lead researcher and Chairman of Obstetrics and Gynecology at Drexel University College of Medicine in Philadelphia, says “The absolute biggest advantage is this allows women to make private decisions” before they are visibly pregnant. Additional advantages to first trimester screening include 40% detection of fetal heart and many other types of birth defects.

The use of four indicators (maternal blood pregnancy associated plasma protein-A and free Beta hCG, ultrasonographic measurement of fetal nuchal translucency, and maternal age) significantly reduces the reliance on age as a predictor of Down syndrome risk “by giving each woman an individual risk assessment – based only in part on her age – the new test will allow many older women to avoid an unnecessary amniocentesis. “ Prenatal screening tests, however, are not a substitute for amniocentesis or chorionic villi sampling because women over the age of 35 are still at increased risk for a variety of chromosomal abnormalities, not just DS.

This early screening test began in the U.S. in 1995 and is available in private obstetric clinics and hospitals. Extensive training is necessary for the reading of the nuchal translucency measurement. To perform the test, the reader must use small calipers on the computer screen to measure the fluid on the back of the baby's neck. A small difference can signal a higher risk. Dr. Mark Evans and Dr. Ronald Wapner both agree standardization of the reading techniques is essential.

NT training is for physicians, sonographers and other healthcare providers is available through official non-profit courses offered by the Fetal Medicine Foundation of America. Contact GeneCare at (800) 277-4363 for further information. Official courses are listed at GeneCare.com. The first trimester "Ultrascreen" test is available at GeneCare Medical Genetics Center. For a list of trained obstetric clinics and hospitals, please call us or e-mail us at info@genecare.com.

Source Information:

Laura Johannes, A Better Test for Down Syndrome, Wall Street Journal, Health & Family, D3, October 9, 2003

First-Trimester Screening for Trisomies 21 and 18, The New England Journal of Medicine, Vol. 349: 1405-1413, 15, October 9, 2003

Similar References:

Genecare Medical Genetics Center: genecare.com

Fetal Medicine Foundation: fetalmedicine.org