ACOG Says All Pregnancies Should Be Evaluated for Down's Syndrome

WASHINGTON, Jan. 2, 2007 -- All pregnant women regardless of age should be offered screening for Down's syndrome before the 20th week of pregnancy, according to new recommendations from the American College of Obstetricians and Gynecologists (ACOG).

A maternal age of 35 should no longer be used as the primary benchmark to determine who is offered screening or the option of counseling and diagnostic testing with amniocentesis or chorionic villus sampling (CVS), said Deborah Driscoll, M.D., of the University of Pennsylvania, who is vice chair of ACOG's Committee on Practice Bulletins-Obstetrics.

The guidelines, developed with ACOG's Committee on Genetics and the Society for Maternal-Fetal Medicine, also advise that all pregnant women, regardless of age, should have the option of diagnostic testing.

A woman's decision to have amniocentesis or CVS screening, ACOG said, should be based on many factors, such as a family or personal history of birth defects, the risk of a chromosome abnormality in the fetus, or, on the other hand, the risk of pregnancy loss from an invasive procedure.

In describing the ultrasonographic and serum markers for selected aneuploidy screening in pregnancy, the researchers said that an increase in "nuchal translucency" (fluid collection at the back of the fetal neck) is now widely recognized as an early presenting feature of a broad range of fetal chromosomal, genetic and structural abnormalities.

However, considerable variability in the detection rates for Down's syndrome among the studies in the early and mid-1990s limited the practical use of the test. A significant advance in first-trimester screening came with the addition of serum analytes, free or total β-hCG, and pregnancy-associated plasma protein A (PAPP-A).

However, the ACOG researchers noted, "not all strategies for first-trimester screening include nuchal translucency measurement because this screening approach is not available in all regions due to the need for specialized training to obtain it."

The goal of the screening techniques, they said, is to offer tests with high detection rates and low false-positive rates that also provide patients with diagnostic testing options if the screening test indicates a risk. To this end, ACOG offered the recommendations on the basis of "good and consistent scientific evidence." They included:

- First-trimester screening using both nuchal translucency measurement and biochemical markers is an effective screening test for Down’s syndrome in the general population. At the same false-positive rates, this screening strategy results in a higher Down's syndrome detection rate than does the second-trimester maternal serum triple screen and is comparable to the quadruple screen.
- Measurement of nuchal translucency alone is less effective for first-trimester screening than is the combined test (nuchal translucency measurement and biochemical markers).
- Women found to have an increased risk of aneuploidy with first-trimester screening should be offered genetic counseling and the option of CVS or second-trimester amniocentesis.

Primary source: Obstetrics and Gynecology: Bahado-Singh R, Driscoll, D., et al "Practice Bulletin #77 Screening for Fetal Chromosomal Abnormalities" *Obstet Gynecol* 2007; 109:217-227

- Specific training, standardization, use of appropriate ultrasound equipment and ongoing quality assessment are important to achieve optimal nuchal translucency measurement for Down's syndrome risk assessment. This procedure should be limited to centers and in dividuals meeting these criteria.
- Neural-tube defect screening should be offered in the second trimester to women who elect only first trimester screening for aneuploidy.

Other recommendations were based on "limited or inconsistent scientific evidence." They included:

- Screening and invasive diagnostic testing for aneuploidy should be available to all women who present for prenatal care before 20 weeks of gestation regardless of maternal age. Women should be counseled regarding the differences between screening and invasive diagnostic testing.
- Integrated first- and second-trimester screening is more sensitive with lower false-positive rates than first-trimester screening alone.
- Serum integrated screening is a useful option in pregnancies where nuchal translucency measurement is not available or cannot be obtained.
- An abnormal finding on second-trimester ultrasound examination identifying a major congenital anomaly significantly increases the risk of aneuploidy and warrants further counseling and the offer of a diagnostic procedure.
- Patients who have a fetal nuchal translucency measurement of 3.5 mm or higher in the first trimester, despite a negative aneuploidy screen, or normal fetal chromosomes, should be offered a targetedultrasound examination, fetal echocardiogram or both.
- Down syndrome risk assessment in multiple gestation using first- or second-trimester serum analytes is less accurate than in singleton pregnancies.
- First-trimester nuchal translucency screening for Down's syndrome is feasible in twin or triplet gestation but has lower sensitivity than first-trimester screening in singleton pregnancies.

Along with the new guidelines, ACOG suggested screening strategies that clinicians might use to meet the needs of individual patients, such as subsequent evaluation after first-trimester screening. ACOG reviewed the advantages and disadvantages of each screening test and some of the factors that determine which screening test should be offered, such as gestational age at first prenatal visit, number of fetuses, previous obstetrical and family history, and the availability of various screening tests.

Ideally all patients should be offered screening before 20 weeks gestation, but it is not practical to have women choose from among the large array of screening strategies. Before deciding which method or methods to use, clinicians are advised to determine which tests are available in their area and which will meet the needs of their patients.

The guidelines also recommended using nuchal translucency along with biochemical markers, and that the optimal time for the ultrasound test appears to be between 12 and 13 weeks of gestation. Training is required to learn the standardized techniques for this measurement, ACOG emphasized.

WASHINGTON IMAGING SERVICES

1135 116th Ave, Suite 260, Bellevue, WA 98004
Phone: 425-688-0100, Fax: 425-454-8911
E-mail: info@washingtonimaging.com
For More Information: www.washingtonimaging.com