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FIRST TRIMESTER GENETIC SCREENING

Recent technology has made it possible to identify fetuses at increased risk for Trisomy 21 (Down syndrome) and Trisomy 18 *without risk* to the mother or fetus. First Trimester Screening is a combination of maternal blood testing for two pregnancy proteins, "Beta hCG and PAPP-A" and an ultrasound measurement of the fluid around the neck of the fetus called "nuchal translucency." This test is performed between 11-13 weeks gestation, and results are available in 4-5 days, making first trimester diagnosis possible in many cases.

If the Screen identifies a fetus at increased risk, additional testing such as chorionic villus sampling (CVS) or amniocentesis can be performed to determine whether the problem is actually present. The detection rate for Down syndrome risk is 91%, and 97% for Trisomy 18. An enlarged nuchal translucency can also be indicative of heart defects and certain other anomalies, but only amniocentesis and CVS are capable of rendering a definitive diagnosis of chromosomal abnormalities. There is, however, some risk involved in either of these procedures.

Prior to the advent of the First Trimester Screening, the earliest available genetic screening was the AFP (alpha-fetoprotein), which is performed between 13-22 weeks. Because the AFP has a high number of false positives, hundreds of unnecessary amniocentesis are performed every year. According to Saleen Chenevert, M.S., Certified Genetics Counselor at North Texas Perinatal Associates in Dallas, Texas, "most offices would have to perform 42 amniocentesis/CVS procedures before finding one child with a chromosome abnormality. Now, with First Trimester Screening, which has better detection and lower false positives, the average office has to do 14 procedures to identify one abnormality." As the availability of First Trimester Screening becomes more widespread, AFP may be used less and amniocentesis used only when increased risk is indicated.

Ms. Chenevert stresses that First Trimester Screening does not replace amniocentesis and CVS. These tissue tests are still used when the Screen shows an increased risk for Trisomy 18 or Down syndrome. Amniocentesis or CVS is also important for the patient who needs to know absolutely whether or not the fetus carries *any* chromosomal abnormalities.

The American College of Obstetrics and Gynecology recommends that First Trimester Screening be offered to all pregnant women, regardless of age. However, because it requires very specialized training for the physician to become certified in performing this test, only a few centers in the country are able to offer it at this time. Providers support the findings of the Screen with counseling by certified genetics counselors who can help patients understand the risks and issues related to evaluation for birth defects and chromosomal abnormalities.

As fertility nurses, we should also be aware that ACOG strongly encourages patients who have undergone any type of assisted reproduction to avail themselves of genetic counseling and screening because of the increased incidence of birth defects noted among ART patients. The January 2005 *Journal of Ob/Gyn Management* cites a study by Schieve and colleagues in Western Australia which found a 9% risk in birth defects among infants conceived through IVF and IVF with ICSI, in contrast to a 4% risk among infants conceived with spontaneous conception. The reason for this increase is unclear at this time, but offering genetic screening would appear prudent in any case.

While First Trimester Screening cannot absolutely rule out birth defects, it can certainly provide reassurance for patients and reduce the need for unnecessary testing. It can also make earlier diagnosis possible for patients who are in need of this information.

For centers offering this test in your area, you may contact GeneCare Medical Genetics Center in Chapel Hill, North Carolina at 1-800-277-4363. Fax 1-919-967-9519.

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