

August 1, 2000

AVAILABLE THROUGH GENECARE MEDICAL GENETICS CENTER
First Trimester Prenatal Diagnosis

Standardized first trimester screening in the US has been provided for over five years with over 30,000 patients screened. Results of 10,200 patients were published in the August 1st issue of *Obstetrics and Gynecology* (Vol. 96:No.2; 207-13, August 2000). The established standardization for clinical centers in the US follows the exact guidelines of Dr. Kypros Nicolaides at the Fetal Medicine Foundation in Kings College, London. Only FMF certified sonographers' NT (nuchal translucency) measurements are combined with freeBeta/PAPP-A lab testing to provide a patient-specific combined report for singleton and multiple pregnancies.

The uniform cut-offs within the clinical program in the US are week specific and are the same as Dr. Nicolaides. As a result, the laboratory analysis and the NT calculation are performed by the same standards in London and in the US. The combined testing has improved detection to 91% for both Down syndrome and trisomy 18, with false positives less than 5%. NT training courses are provided in the US in combination with the Fetal Medicine Foundation. The follow up film certification is provided through Naomi Green at Cedar Sinai Hospital in Los Angeles. The course and film certificates are from Dr. Nicolaides. NT medians are monitored quarterly which is critical to maintaining precise NT measurements. Training schedules at Cedar Sinai Hospital, GeneCare, and NTD Laboratories are available through those Centers.

The first trimester screening by prospectively documenting freeBeta/PAPP-A prospective screening for over 10,200 patients and clinical program for NT/freeBeta/PAPP-A in the US meets the requirements of ACOG for clinical use. NT in this large US study with standardized QC. Because the FASTER trial (a new NIH study) has not taken the established approach to training, QC, laboratory and is not reporting results until after 16 weeks, this trial will not impact first trimester screening programs following the FMF's internationally documented protocol.

First screening is now available in most states in the US. Insurers have been very positive about paying for first trimester screening. If you have questions about interpretation of reports, call your local center or our Center for references and consultations. Genetic counselors are also encouraged to attend the NT courses because they are critical for program development, patient counseling, program QC and outcomes analysis.

Philip D. Buchanan, PhD, FACMG
Director, GeneCare Medical Genetics Center