

## No more triple screens!

*By Philip Buchanan, PhD, FACMG*

Because of our commitment to providing the best chromosome and open neural tube screening possible, GeneCare offers tests different from the customary screening tests. Our options for testing have shifted from the second trimester into the first trimester. Our first trimester Ultrascreen combines an ultrasound examination and measurement of two biochemical markers found in the blood of all pregnant women, freeBeta and PAPP-A. This 1<sup>st</sup> trimester screening makes up the majority of testing performed in Europe and GeneCare has made it available here in the US.

In the early days of screening, only AFP was measured to screen for Down syndrome and ONTDs. Even after other chemicals, such as hCG, were discovered to be better markers for Down syndrome than AFP alone, many labs continued to use AFP only screening for years, even with its paltry 30% detection rate. Over the years, hCG and uE3 have been added to develop the 2<sup>nd</sup> trimester triple screen and improve the detection rate for Down syndrome to 60%. Later on, the freeBeta form of hCG was discovered to be the best known chemical predictor of Down syndrome. The double screen method of combining 2<sup>nd</sup> trimester levels of freeBeta and AFP improves sensitivity for Down syndrome to 80%.

In the US, we have moved away from the 2<sup>nd</sup> trimester triple screen to 1<sup>st</sup> trimester Ultrascreen, which provides improved screening with earlier results for our patients. The 1<sup>st</sup> trimester test has a higher Down syndrome detection rate (90%) than the triple screen (60%) and double screen (80%). 1<sup>st</sup> trimester screening provides other advantages over the triple screen as well, such as:

Ultrascreen has a reduced false positive rate; only half as many women are reported to be at increased risk of having a baby with Down syndrome.

The amount of expensive follow-up testing needed, such as CVS and amniocentesis, is also decreased.

1<sup>st</sup> trimester screening allows women to receive test results earlier (11-13 weeks gestation) and plan accordingly.

Ultrascreen's 1<sup>st</sup> trimester trisomy 18 detection rate (>90%) is dramatically increased over the triple screen rate of 60%.

Since 1<sup>st</sup> trimester Ultrascreen is more accurate, it is not necessary to use 3 and sometimes even 4 2<sup>nd</sup> trimester markers like the triple and quad screens.

With GeneCare's second trimester AFP only test, the detection rate for ONTDs, such as spina bifida, is 98% vs. the triple screen=80%. This is achieved by complex Bayesian analysis, which estimates a patient-specific risk instead of just using AFP MoM value cut-offs.

Ultrascreen uses a finger stick to collect blood onto filter paper. The chemicals are fixed and thus stabilized against degradation, which yields more accurate results than traditional blood tubes. In addition, a minimal amount of phlebotomy skill is required to draw samples.

In summary, not only are detection rates increased with this method but the number of screen positive anxious women is decreased from the triple screen because freeBeta is the most specific biochemical marker and nuchal translucency the most specific ultrasound marker for Down syndrome.

**GeneCare Medical Genetics Center, in Chapel Hill, offers genetic screening, including 1<sup>st</sup> trimester Ultrascreen and 2<sup>nd</sup> trimester double and AFP only tests, as well as diagnostic laboratory services. For more information please call (919) 942-0021 or 1-800-277-4363 or visit our web site: [www.genecare.com](http://www.genecare.com).**