

Date: May 18, 2007

To: **OB/GYN Providers**

From: Strong Health Maternal Serum Screening Program  
Department of Pathology & Laboratory Medicine  
Reproductive Genetics, Department of OB/GYN

Subject: **1st Trimester Screening Update**

Since 1st Trimester Screening became available in Rochester in Fall of 2004, its popularity has exploded as shown by the numbers below.

<b>Time frame</b>	<b># of patients screened</b>
<i>Last 3 months of 2004</i>	25
2005	400
2006	1,300
<i>First 3 months of 2007</i>	500
<i>2007 (estimate)</i>	2000

Many providers have made it a part of their standard protocol for 1st trimester care. It has replaced AFP+ (Quad Screening) as the preferable screen for chromosome disorders in women of all ages who seek prenatal care in the 1st trimester. Women over the age of 35 use the risk information from it as a way to determine if invasive testing such as amniocentesis or CVS is necessary.

**Yet it is important to be aware of its limitations, especially in "high risk" patients.**

■ **It is a screening test**

Although many patients seem to believe this test is different than other screening tests, it is not. Just as with AFP+ Screening in the 2nd trimester, most "positive" results will be "false positives" (positive screening result in a normal pregnancy), and a few "negative" results will be "false negatives" (normal result in an affected pregnancy).

■ **Patients greater than or equal to 35 years old should understand its limitations**

In the past, aneuploidy screening was used mostly by younger women to see if their risk was high enough to justify invasive diagnostic tests. Increasingly, especially with 1st Trimester Screening, many older women use it to see if their risk is low enough to "avoid" invasive procedures.

The risk information from 1st Trimester Screening can be helpful to many patients. However for patients who want definitive chromosome results, amniocentesis or CVS would still be required.

All patients who are 35 and over should still be offered diagnostic testing and should be offered genetic counseling to review the most recent information about diagnostic and screening test options in detail.

■ **It should not be used in certain situations**

- Early demise of a fetus in twin (or triplet) pregnancies

This would be expected to alter the maternal serum analytes making risk assessment for the remaining fetus unreliable. In these cases, the 2nd trimester Quad Screen should be offered.

- Women at very high risk

Some women may be at very high risk for Down syndrome in their pregnancies. For example a woman with a 14/21 chromosomal translocation may have a 10-15% chance for Down syndrome in her pregnancies.

Screening tests, due to their inaccuracies, cannot be used in these very high risk situations. If you have questions about whether to offer screening to a particular patient, please call us at (585)275-3304 for advice.

■ **It screens best for Down syndrome and Trisomy 18**

1st Trimester screening has been proven effective in screening for both Down syndrome and Trisomy 18, with detection rates in the screened population between 80-85%. It is less effective as a screen for Trisomy 13, and cannot screen at all for chromosome conditions involving other chromosomes.

Therefore, patients over 35 may be at significant risk for other chromosome abnormalities not assessed by 1st Trimester Screening. Patients with chromosomal translocations or inversions, or whose partners have chromosomal variants, should not have this screening.

Please call (585)275-3304 if you have any additional questions.

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