

MFM NEWS

Volume 7, Issue 2

Department of Obstetrics & Gynecology
Division of Maternal-Fetal Medicine

March 2007

ACOG (And EVMS MFM) Endorses First Trimester Risk Assessment

It is not every day that Obstetrics is featured on CNN, Fox News and most local and National newspapers around the country, but that was the case early in January after ACOG released its Practice Bulletin #77 which endorsed First Trimester Risk Assessment for all Obstetrical patients.

ACOG first addressed Nuchal Translucency (NT) measurements in their Committee Opinion # 223 in October 1999. Although increasingly used in Europe for Down Syndrome screening, ACOG recommended that "First trimester screening for chromosomal, cardiac, and other abnormalities using the NT marker alone or in combination with serum markers appears promising but remains investigational". ACOG called for "the need for standardization and for further studies". Until then, NT measurements were not recommended for routine clinical use.

Since then studies in Europe (SURUSS) and in the U.S. (BUN and FASTER trials) have been performed which have given us a wealth of information on the effectiveness of First Trimester screening. In July 2004, ACOG Committee Opinion #296 replaced the 1999 opinion. At that time, ACOG recommended that "Although First trimester screening for Down Syndrome and Trisomy 18 is an option, it should be offered only if the following criteria can be met: 1. Appropriate ultrasound training and ongoing quality monitoring programs are in place. 2. Sufficient information and resources are available to provide comprehensive counseling to women regarding the different screening options and limitations of these tests and 3. Access to an appropriate diagnostic test is available when screening tests results are positive".

The Nuchal Translucency Quality Review (NTQR) program, which members of our division have had significant input, has taken that challenge to establish education, training, and quality monitoring, providing both web based www.ntqr.org and land courses for physicians and sonographers, providing outreach, genetic counseling and diagnostic services.

In response to these efforts and those of other organizations,

ACOG in January 2007 has issued Practice Bulletin #77 replacing the earlier Committee Bulletins. Using the data published from multiple studies comparing first and second trimester screening strategies for Down Syndrome, they have concluded that:

1. All obstetrical patients should be offered aneuploidy screening before 20 weeks of gestation regardless of maternal age.
2. First trimester screening with NT and serum analytes (PAPP-A and Free or total BHCG) is an effective screening test for the general population and is comparable to 2nd trimester screening with serum markers (MSAFP, BHCG, uE3, and Inhibin) with the additional advantage of earlier and thus safer and more available pregnancy terminations if desired.
3. Combinations of first and second trimester screening will lead to the higher sensitivity with lower False Positive rates.
4. Women with increased risk of aneuploidy should be offered diagnostic testing with either CVS or second trimester amniocentesis.
5. The caveat of necessary training, standardization, appropriate ultrasound equipment and ongoing quality assessment are necessary to achieve optimal NT measurements.

As can be seen, NT measurements have gone from a promising investigational tool (1999), to an option in appropriate circumstances (2004), to standard practice for all patients (2007). It is now our challenge as an obstetrical community to implement these recommendations in our region. From a practical perspective this will change established practice patterns. Previously Down syndrome screening has been a second trimester issue. It will now need to be addressed in the first trimester and frequently at the first prenatal visit. We anticipate that patients at risk for Down syndrome (AMA, previous affected pregnancy) will be referred for genetic counseling in the first trimester so that all their options can be reviewed in a timely fashion. Low risk patients will need to be screened in your offices as is currently done with MSAFP screening in the second trimester. They can then be referred if found to be at risk. Dr. Steve Warsof, Director of Genetics and Prenatal Diagnosis, EVMS, is available to assist you in developing a system for managing this through your office. We hope the table on the reverse side will be helpful to you in different Down Syndrome screening strategies.

Prenatal Screening Options at a Glance

	10.5 –14 Weeks				16 - 20 Weeks	
	First Trimester Screening	Sequential Screening (Stepwise)	Biochemical Integrated Screening	Integrated Screening	Quad Screen	URAD (ultrasound)
Timing	11w-13w6d (Blood can be drawn 9-14 weeks)	Step 1: 11w - 13w6d Step 2: 15-20 weeks	1 st part: 11w-13w6d 2 nd part: 15-20 weeks	1 st part: 11w-13w6d 2 nd part: 15-20 weeks	15-20 weeks	18-22 weeks
Procedure	Blood sample and Ultrasound	Blood sample and ultrasound in 1 st trimester; blood sample in 2 nd trimester	Blood sample in 1 st trimester (NT cannot be obtained); blood sample in 2 nd trimester	Blood sample and ultrasound in 1 st trimester; blood sample in 2 nd trimester	Blood sample	Ultrasound
Detection Rate	83% of Down syndrome 80 % of Trisomy 18	91% of Down syndrome 90% of Trisomy 18 80% of open neural tube defects	87% of Down syndrome 90% of Trisomy 18 80% of open neural tube defects	92% of Down syndrome 90% of Trisomy 18 80% of open neural tube defects	75-80% of Down syndrome 60% of Trisomy 18 80% of open neural tube defect	50% of Down syndrome
Measures	PAPP-A, β-HCG, Nuchal translucency	PAPP-A, β-HCG, AFP, estriol, Inhibin A and nuchal translucency	PAPP-A, hCG, AFP, estriol, and Inhibin A	PAPP-A, hCG, AFP, estriol, Inhibin A, and nuchal translucency	AFP, HCG, estriol, Inhibin A	Detailed Anatomy Scan
Twin Pregnancy	Trisomy 18 risk is not available; Down sx risk is "pseudorisk"	Trisomy 18 risk is not available; Down sx risk is "pseudorisk"	Trisomy 18 risk is not available; Down sx risk is "pseudorisk"	Trisomy 18 risk is not available; Down sx risk is "pseudorisk"	Trisomy 18 risk is not available; Down sx risk is "pseudorisk"	Each baby can be evaluated separately
Results	5-7 days	Preliminary result –5 days Final result - 5 days after second blood draw	~5 days after second blood draw	~5 days after second blood draw	5-7days	Immediate
Follow-up	- If Low risk: MSAFP at 16-18wks Morphology scan (18-22wks) - If high risk: possible CVS/ Amnio	- If Low risk: second blood draw at 16-18wks Morphology scan (18-22wks) - If high risk (>1/50): CVS is offered	If low risk, morphology scan. If high risk, genetic counseling and possible amniocentesis.	If low risk, morphology scan. If high risk, genetic counseling and possible amniocentesis.	Morphology scan and possible amniocentesis	Genetic counseling and possible amniocentesis if abnormalities are detected

If you have any questions or suggestions regarding the MFM Newsletter, please contact the editor Sue K. Sayegh, M.D. at sayeghsk@evms.edu



**EASTERN VIRGINIA
MEDICAL SCHOOL**
ESTABLISHED 1973

#616
EASTERN VIRGINIA MEDICAL
SCHOOL
P.O. BOX 1980
Norfolk, VA 23501-1980