



## **MATERNAL SERUM SCREENING**

Maternal Serum Screening (MSS) is a standard tool in obstetrical care to identify women at higher risk for open neural tube defects (ONTD), Down syndrome (DS) and certain other defects such as trisomy 18. The screening is performed by measuring a combination of analytes in maternal serum produced by the fetus and placenta.

A multidimensional relationship between the analyte values along with maternal demographic information such as age, weight, gestational age, diabetic status, smoking status, and race are used to derive risk estimates.

The laboratory establishes specific cut-offs for each condition, which classify each screen as **screen-negative** or **screen-positive**. A positive screen is not a diagnosis but recommends that further evaluation should be considered. A negative screen identifies low risk for a specific fetal disorder, but does not guarantee the absence of a fetal defect.

### **Maternal Serum Screening Options at Marshfield Labs**

**Second Trimester Screen** (between 15 wks, 0 days and 21 wks, 6 days).

**AFP-1** is a single marker screen offered in the second trimester for open spina bifida only; measures maternal serum alpha fetoprotein (AFP) only.

**AFP-4** is a quadruple marker screen offered in the second trimester for open spina bifida, Down syndrome and trisomy 18; combines the maternal serum measurements of AFP, human chorionic gonadotropin (hCG), unconjugated estriol (uE3) and inhibin A (DIA).

**AFP-3** is a triple marker screen which is no longer an orderable test by Marshfield Labs. It has a higher false positive rate and lower detection rate compared with AFP-4.

### **Interpretation**

#### **NEURAL TUBE DEFECTS**

An AFP MoM  $< 2.5$  is reported as screen-negative.

An AFP MoM of  $\geq 2.5$  (singleton pregnancy) is reported as screen-positive.

#### **DOWN SYNDROME**

An estimated screen risk  $< 1/300$  is reported as screen-negative.

An estimated screen risk  $\geq 1/300$  is reported as screen-positive.

#### **TRISOMY 18**

An estimated screen risk  $< 1/100$  is reported as screen-negative.

An estimated screen risk  $\geq 1/100$  is reported as screen-positive.

The accuracy of estimated risk calculations and screen results are dependent on accurate information for gestational age, maternal age, race, diabetic status, and weight. Inaccurate information can lead to significant errors in the estimated risk and result in false-positive or false-negative screen results. Because of its superior accuracy, determination of gestational age by ultrasound is recommended rather than by last menstrual period.

The MSS risk assessment algorithm and patient requisitions have been updated to minimize inaccuracies due to ordering practices and optimize test performance. For details refer to Newsletter Vol 35, No 15 – September 24, 2012.

#### **Quadruple screen estimated detection and false positive rate**

Down Syndrome	Detection rate 85%; False positive rate 6.6% at 1:300
Trisomy 18	Detection rate 60%; False positive rate 0.2% at 1:100
Open NTD	Detection rate 70-75%; False positive rate 2% at 2.5 MoM*

\*MoM = Multiples of the Median

***It is recommended that women who have first trimester screening for aneuploidy should not undergo second trimester quadruple screening.***

If first and second trimester screens are performed for the same disorder, then the false positive rates may be additive and, therefore, can lead to unnecessary invasive procedures.

***However, if women who have first trimester screening performed for aneuploidy wish to assess their risk for an open neural tube defect, the AFP-1 test should be ordered.***

#### **Contact Information**

Marshfield Labs Customer Service: 800-222-5835.

#### **Questions or Comments**

Please refer any questions or comments to:  
Annu Khajuria, Ph.D., Clinical Chemist  
Joyce Flanagan, Ph.D., Clinical Chemist