

Maternal Serum Multiple Marker Screening for Chromosome Abnormalities and Open Neural Tube Defects: A Fact Sheet for Practitioners

The multiple marker screening test is a screening test. It is not a diagnostic test. Multiple parameters, including maternal age, gestational age, maternal weight, and levels of one or more of the following analytes: alpha fetoprotein (AFP), human chorionic gonadotropin (hCG) and unconjugated estriol (uE3), and inhibin-A, are used to calculate a risk for a woman to have a baby with Down syndrome, trisomy 18 or a neural tube defect.

Screening for Down syndrome (DS)

- The screening uses all four analytes.
- Multiple marker screening should be offered to all pregnant women who will be under 35 years of age at delivery. Using the algorithm on the back of this page, 60-70% of babies with DS will be detected in women under 35, compared to only 20% if MSAFP is used alone. If four analytes are used, the detection rate increases significantly.
- MSAFP alone should not be used as a precursor to multiple marker screening.
- Multiple marker screening is not equivalent to diagnostic testing. Women ≥ 35 should always be offered diagnostic testing for chromosome abnormalities. However, if amniocentesis and CVS are declined, multiple marker screening may be useful to women who would like refined risk information.
- Results will be reported either in terms of second trimester risks, or risks at term. For example, a 1/270 second trimester risk for DS is equivalent to a 1/400 risk at term. Second trimester risks differ from term risks because more babies with DS than with normal chromosomes are stillborn or miscarried in the third trimester.
- Typically, it is the most reassuring to a patient to present the laboratory results in the form of a percentage rather than a fraction. For example, a 1/200 risk for DS is the same as a ½% risk, or 99 ½ % chance the baby does not have DS. Even if her risk is increased, a patient can be reassured that the odds are in her favor that her baby does not have DS.
- Recalculate risks only when the reported risk is based on menstrual or exam dating, and ultrasound dating differs from original dating by two weeks or more. The risk can be recalculated by the laboratory performing the serum screening.

Screening for Trisomy 18

- The screening uses AFP, hCG, and uE3. Estriol is the most sensitive indicator.
- Multiple marker screening may detect an increased risk for Trisomy 18.
- Screen positive results cannot be explained by incorrect dating because many fetuses with Trisomy 18 are growth retarded. Therefore, recalculating the risk of trisomy 18 because of incorrect dates is not recommended.

Screening for Open Neural Tube Defects (ONTD)

- The screening uses alpha fetoprotein only.
- The detection rate for ONTD is 80-90% using MSAFP cut-off at 2.5 multiple of the median (MOM).
- An elevated MSAFP may be explained by incorrect dating, bleeding, multiple gestation, other anatomic defects, or a chromosome problem.

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Interpreting Screening Results

To accurately interpret the results, the laboratory will need to know ethnicity, if this is a twin gestation, ultrasound dating, and if the patient has diabetes.

When the maternal serum multiple marker screen is positive, genetic counseling and prenatal diagnosis should be offered. Unexplained hCG and/or MSAFP elevations may be associated with adverse perinatal outcome including: preeclampsia, premature labor, low birth weight, placental abruption or stillbirth. Follow up ultrasound an/or perinatal consult should be considered.

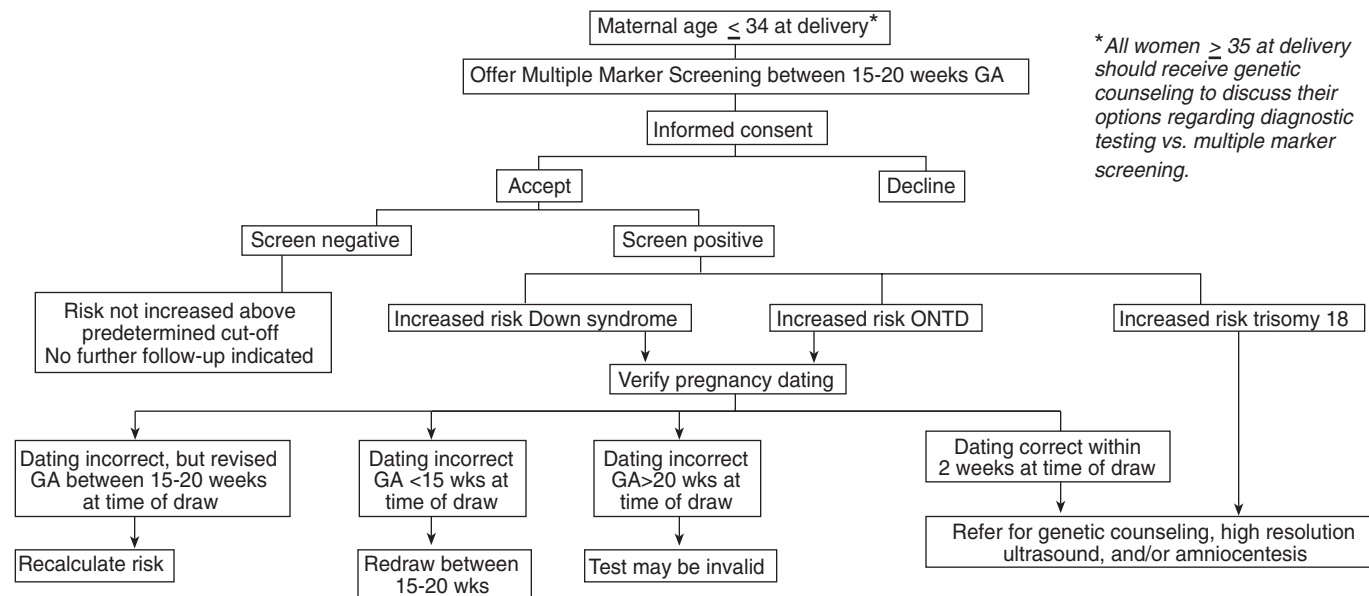
Each laboratory may have its own interpretation for what is considered screen positive. Please consult with the testing laboratory regarding their screening parameters.

Remember, maternal serum multiple marker screening is not a diagnostic test.

Please see page 3 for an algorithm and a list of prenatal genetics clinics in the Pacific Northwest.

This fact sheet is available from:
Pacific Northwest Regional Genetics Group
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<http://mchneighborhood.ichp.edu/pacnorgg>

Maternal Serum Multiple Marker Screening Algorithm for Practitioners



*All women ≥ 35 at delivery should receive genetic counseling to discuss their options regarding diagnostic testing vs. multiple marker screening.

Please consult with the testing laboratory regarding their screening parameters, specimen handling, and test availability.
Source: consensus of the PacNoRGG Prenatal Genetics committee

Where can I get further information?

Prenatal Genetics Clinics in the Pacific Northwest

ALASKA

Anchorage
Providence Hospital, 907/261-3097

IDAHO

Boise
St. Luke's Hospital, 208/381-3088

OREGON

Eugene
Center for Genetics and Maternal Fetal
Medicine, 541/349-7600

Portland

Kaiser Permanente NW, 503/331-6593
Legacy Emanuel Hospital & Health
Center, 503/413-4726
Northwest Perinatal Center,
503/297-3660
Oregon Health and Science University,
503/494-7577

WASHINGTON

Bellevue
Eastside Maternal-Fetal Medicine,
425/688-8111

Kirkland

Evergreen Hospital Maternal-Fetal Medicine,
425/899-2200

Renton

Perinatal Obstetrix Medical Group, Inc.,
425/656-5520

Seattle

Swedish Medical Center, 206/386-2101
University of Washington Medical Center,
206/598-8130

Spokane

Inland Northwest Genetics Clinic,
509/473-7115

Tacoma

Madigan Army Medical Center,
253/968-1252 Prenatal Appts.,
Consult for Reproductive Genetics
253/968-1160
Obstetrix Medical Group of
Washington, Inc., 253/552-1037

Walla Walla

Blue Mountain Genetics Clinic,
509/525-1302

Wenatchee

Central Washington Hospital Genetics
Group, 509/667-3350

Yakima

Yakima Valley Memorial Hospital,
509/575-8160



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