



Maternal Serum Screening

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You were informed that your:

_____ Maternal serum screening (MSS) test, also known as the quad screen

_____ Other: _____

indicates that your risk for having a baby with **trisomy 18 (an extra chromosome #18)** is increased.

How Did I Get This Result?

You probably don't have a relative with trisomy 18 in your family, and you may be wondering why your doctor ordered this test. Trisomy 18 is caused by the presence of an extra chromosome #18. It is the second most common chromosome abnormality next to Down syndrome (which is trisomy 21). Trisomy 18 is **not inherited**. Trisomy 18 usually occurs **BY CHANCE** and every pregnant woman has a chance, usually dependent upon her age at delivery, to have a baby with trisomy 18. The chance increases with increasing maternal age. Most pregnancies with trisomy 18 are miscarried. Those babies who go full term are born with many health problems, which are usually incompatible with long term survival.

What do these tests measure? The MSS test measures AFP, hCG, uE3, and DIA.

AFP, or alpha-fetoprotein, is made by the baby's liver during the second trimester of pregnancy and is normally filtered into the fluid surrounding the baby (amniotic fluid). AFP finally crosses the placenta into the mother's blood. Adults usually do not produce AFP in measurable quantities (unless they have a specific medical problem), so that AFP comes strictly from the baby (or babies).

hCG, or human chorionic gonadotropin, is produced only by the placenta, or afterbirth. Like AFP, it is also found in the mother's blood during pregnancy. hCG is, in fact, the chemical which is usually measured in either blood or urine to determine that a woman is pregnant.

uE3, or unconjugated estriol, comes from both the baby's liver and the placenta, and is therefore only produced during a pregnancy.

DIA, or dimeric inhibin A, comes from the placenta. Levels in maternal serum remain relatively constant through the 15th-18th week of gestation in normal pregnancies.

The normal values of each of these substances change with each week of pregnancy. That is why the results are expressed in multiples of the median, or MOMs. The MOM tells how close your value is to the median (average) value for that week of pregnancy. For example, "0.5 MOMs AFP" means there is **half** as much AFP as is usually found during that week of the pregnancy, while "2.5 MOMs AFP" means there is **two and one half times** the usual amount of AFP for that particular week of pregnancy.

How do these measurements indicate an increased chance for trisomy 18?

In a pregnancy in which the baby has trisomy 18, all four markers tend to be somewhat lower than average.

How many trisomy 18 pregnancies does this test detect?

MSS detects 70% of cases of trisomy 18. The detection rate is NOT 100%, and many factors unrelated to trisomy 18 can cause the test to look abnormal. The **MSS** test is a **SCREENING** test, and is **not** diagnostic. **A normal baby is still the most likely outcome, even after an abnormal maternal serum screening test.**

You had the MSS, which measures AFP, hCG, uE3, and DIA. All four values were low for this gestational age, and therefore the risk for your baby to have trisomy 18 is increased. Ultrasound and/or amniocentesis may be recommended.

What Other Factors Could Affect This Test Result?

- 1. The baby is producing the normal amounts of AFP, hCG, uE3, and DIA, but only small amounts are filtering through the placenta, into your bloodstream.** This may mean that the baby is healthy, but the placenta may have a problem. Ultrasound examinations are sometimes recommended in this situation to monitor the growth of the baby, to make sure the placenta is doing its job.
- 2. The dates of your pregnancy.** The levels of AFP, hCG, uE3, and DIA that are considered "normal" are different for each week of pregnancy. Estimating the week of pregnancy from the first day of your last menstrual period (called LMP) sometimes results in inaccurate dating of a pregnancy. Ultrasound examination can usually correct this discrepancy, and the AFP, hCG, uE3, and DIA levels can then be recalculated if the discrepancy is significant.
- 3. The levels of AFP, hCG, uE3, and DIA found in your blood simply reflect a normal variation in these levels, even though the amounts found are outside of the range considered 'normal'.**
- 4. The baby has trisomy 18 or another genetic condition or chromosome abnormality (numerical or structural).** Amniocentesis can detect over 99% of these problems.