



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 **Down syndrome** is increased.

How Did I Get This Result?

You probably don't have a relative with Down syndrome in your family, and you may be wondering why your doctor ordered this test. Down syndrome is caused by the presence of an extra chromosome #21. Down syndrome is **not inherited** (in >97% of cases). Down syndrome usually occurs **BY CHANCE** and every pregnant woman has a chance, usually dependent upon her age at delivery, to have a baby with Down syndrome. The chance increases with increasing maternal age. In addition, it is not a risk limited only to pregnant women over 35. In fact, 75% of babies born with Down syndrome are born to mothers UNDER 35. (If you have a relative with Down syndrome, you may wish to speak directly with a genetic counselor to better determine whether it may be the inherited type).

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 Down syndrome?

In a pregnancy in which the baby has Down syndrome, the AFP and uE3 tend to be somewhat lower than average, while the hCG and DIA tend to be elevated. There are no set "cut-offs" for each analyte, however. To determine each patient's risk of carrying a baby with DS, all analytes measured from the blood test, along with the mother's age risk to have a baby with DS, are taken into account.

How many Down syndrome pregnancies does this test detect?

MSS detects approximately 75% of cases of Down syndrome in women under 35 and over 80% of cases of Down syndrome in women over 35. The detection rate is NOT 100%, and many factors unrelated to Down syndrome can cause the test to look abnormal. The MSS 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.**

What were my chances to have a baby with Down syndrome before the test, versus after the test?

The chances for a _____year old woman to have a baby with Down syndrome (based on age alone) is 1 in_____. However, taking into account the amount of AFP, hCG, uE3, and DIA found in your blood from the **MSS** test, your risk to have a baby with Down syndrome is 1 in_____. This risk is known as the "adjusted Down syndrome risk". Turning this number around, you have a _____ out of _____ chance that the baby does **NOT** have Down syndrome. Again, the **MSS** test is **not** diagnostic, but instead *screens* pregnancies (mostly for mothers under 35), to determine who, in this group, should be referred for additional testing. When the adjusted Down syndrome risk is greater than or equal to that of a 35 year old mother (1:270), ultrasound and/or an amniocentesis may be recommended.

What Other Factors Could Affect This Test Result?

- 1. The dating of your pregnancy is incorrect.** Estimating the week of pregnancy from the first day of your last menstrual period (called 'LMP') alone sometimes results in inaccurate dating of a pregnancy. Ultrasound examination (also called sonography) can correct this discrepancy, and the AFP, hCG, uE3, and DIA levels can then be recalculated.
- 2. The baby is producing the normal amounts of AFP & uE3, 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 and to make sure the placenta is doing its job. (Likewise, a high or low amount of hCG or DIA may be indicative of a placental problem).
- 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 Down syndrome or another type of abnormality in the chromosome material (numerical or structural).** Amniocentesis can detect over 99% of these problems.