



ALPHA-FETOPROTEIN TEST

Most health care providers now routinely offer their pregnant patients a blood test called the maternal serum alpha-fetoprotein (MSAFP) screening test. This test identifies pregnancies at higher-than-average risk of certain serious birth defects, such as spina bifida (open spine) and Down syndrome (characteristic facial features, mental retardation, and possible heart defects and other problems). The test can provide valuable information about a developing fetus. It is important for pregnant women to understand that, in most cases, an abnormal test result does not indicate a problem with the fetus and further testing can confirm this.

Q. WHAT IS ALPHA-FETOPROTEIN?

A. Alpha-fetoprotein (AFP) is a substance produced by the liver of the fetus. Some of this protein is excreted into the amniotic fluid surrounding the fetus. A small amount of AFP passes into the mother's bloodstream, where the concentration rises gradually until late in pregnancy. AFP levels can be measured during pregnancy by taking a sample of either the mother's blood or the amniotic fluid.

Q. WHEN DURING PREGNANCY IS THE MSAFP TEST DONE?

A. This blood test is most often done between 15 and 18 weeks after the last menstrual period. The results are usually available within one week. If the MSAFP levels are abnormal, the test may be repeated, as late as 20 to 21 weeks in some centers.

Q. DOES A HIGH OR LOW MSAFP READING MEAN THE BABY HAS A BIRTH DEFECT?

A. No. This test cannot diagnose a birth defect--it can only indicate an increased risk. An abnormal test result simply means that additional testing is needed. Out of every 1,000 women who take an MSAFP test, up to 100 will have an abnormal result. However, only about 10 of these 100 women will have a fetus with a birth defect.

For most of the rest, the AFP level simply indicates that the fetus is either a few weeks older or younger than originally thought. Because the range of normal results varies with the weeks of pregnancy, it is very important to know the accurate gestational age of the fetus. The gestational age of the fetus should be confirmed by ultrasound, if there is any question.

A multiple pregnancy (twins, triplets, etc.) is another common cause of an elevated reading. Pregnant women who do not understand this may experience much unnecessary anxiety.

Q. WHAT FETAL PROBLEMS CAUSE HIGH MSAFP LEVELS?

A. Neural tube defects (NTDs) are among the most common and severe problems associated with high MSAFP levels. The neural tube is the part of the developing embryo from which the brain and spinal cord form. If the neural tube does not close properly during the fourth week after conception, birth defects such as spina bifida and anencephaly may result. Approximately 2,500 babies are born in this country each year with these birth defects.

Spina bifida, often called open spine, is a birth defect of the backbone. In most cases of this defect, the spinal cord is malformed and protrudes from the back, resulting in varying degrees of leg paralysis and bladder and bowel problems. In anencephaly, the upper end of the neural tube fails to close, leading to severe malformations of the brain and skull. Babies with anencephaly do not survive.

The causes of NTDs are not well understood. Scientists believe that genetic and environmental factors act together to cause these malformations. About 90% to 95% of babies with NTDs are born to couples with no family history of these abnormalities.

Recent studies have shown that 50% to 70% of NTDs can be prevented if women of childbearing age consume folic acid (a B vitamin) before and during the early weeks of pregnancy. The U. S. Public Health Service and the March of Dimes recommend that all women of childbearing age consume 0.4 milligrams (400 micrograms) of folic acid each day prior to conception and during the first months of pregnancy. Since it is hard to get enough folic acid through diet alone, taking a daily multivitamin can make up the difference.

Certain uncommon birth defects of the abdominal wall also can raise MSAFP levels, as can certain kidney and bowel defects. In rare instances, the MSAFP level is elevated because the fetus is dying or dead.

Q. WHAT DOES A LOW MSAFP READING MEAN?

A. Low MSAFP levels are sometimes associated with chromosomal abnormalities, such as Down syndrome. However, the MSAFP screening test is not as accurate in detecting pregnancies at increased risk of Down syndrome as it is with neural tube defects.

Q. WHAT TESTS ARE RECOMMENDED FOLLOWING AN ABNORMAL MSAFP READING?

A. After an abnormal MSAFP reading, the next step is usually an ultrasound examination. This test uses sound waves to take a picture of the fetus. Ultrasound can help determine the gestational age of the fetus and show if a woman is carrying twins. If either of these factors accounts for the abnormal AFP reading, no further testing is needed. Ultrasound also can detect some serious birth defects.

If the ultrasound examination does not provide an explanation for an abnormal MSAFP reading, the health care provider may suggest amniocentesis, a test during which the doctor inserts a thin needle through the abdominal wall and into the uterus to withdraw a few teaspoons of amniotic fluid. Fetal cells contained in the amniotic fluid will be tested for Down syndrome and other chromosomal abnormalities, and the level of AFP in the amniotic fluid will be measured. When the amniotic fluid level of AFP is elevated and all other causes have been ruled out, it is very likely that the fetus has a neural tube defect.

With a high MSAFP reading, along with amniocentesis, the health care provider may recommend a high-resolution ("level three") ultrasound examination to inspect the fetal skull and spine. This procedure can quite accurately detect or rule out serious neural tube defects, but is generally available only at major medical centers. It also may help predict the severity of neural tube defects. Using MSAFP screening, ultrasound and amniocentesis, about 95% of cases of anencephaly and 80% of cases of serious spina bifida can be detected prenatally.

Q. WHAT ARE THE BENEFITS OF MSAFP SCREENING?

A. For the great majority of women, MSAFP screening provides reassurance that their fetus does not appear to have certain serious birth defects. Test results also can help a woman manage her pregnancy more effectively. For example, finding the correct gestational age helps determine whether the fetus is growing at a normal rate. And detecting a multiple pregnancy allows for special care.

When a neural tube defect or other problem is diagnosed or suspected, a couple can discuss all their options with trained personnel. They can plan for delivery in a specially equipped medical center so that soon after birth the baby can have any surgery or treatment required.

In some cases, there is no clear-cut explanation for an abnormal MSAFP test result. Abnormal readings have been linked with pregnancy problems such as detachment, or abruption, of the placenta, preterm labor and low birthweight. Unexplained high readings may be associated with an increased risk of fetal death late in pregnancy. While most women with unexplained high readings do not have fetal deaths, this study suggests that health care providers may be able to prevent some fetal deaths by monitoring these pregnancies very carefully in the third trimester. If signs of fetal difficulties are found, it is possible that some babies can be saved by early delivery.

Some health care providers now offer an enhanced MSAFP screening test to their patients. This new screening test (often called triple screening), that measures MSAFP levels along with the levels of two pregnancy hormones called estriol and human chorionic gonadotropin, appears to detect at least 60% of cases of Down syndrome, as compared to about 30% with MSAFP screening alone. (Some laboratories measure only two substances, MSAFP and human chorionic gonadotropin.) A computer calculates the individual risk of Down syndrome based on the levels of these three substances plus the woman's age and certain other factors. If the woman's risk equals that of a 35-year-old woman (about 1 in 270), she will be offered follow-up tests such as amniocentesis. Most health care

providers currently offer prenatal testing to women in this age group because the risk of Down syndrome and other chromosomal abnormalities increases with age.

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