THE ALPHA-FETOPROTEIN (AFP) BLOOD TEST

A blood test is available called the alpha-fetoprotein (al-fa-fee-toe-pro-teen) (AFP) test. This test helps identify a small number of women whose unborn babies may have certain birth defects.

If you are thinking of having the AFP blood test, you should be aware that the test itself is not a guarantee. It is possible that the neural tube defects may be missed, and there is a very slight chance, even with extensive follow-up testing, that a healthy unborn baby may be incorrectly identified as having a neural tube defect. After reading this brochure, discuss the possibility of incorrect results with your doctor before you ask for an AFP test.

WHAT ARE SOME TYPES OF NEURAL TUBE DEFECTS?

Two common and serious types of neural tube defects are anencephaly and spina bifida.

Anecephaly- In this disorder, much of the brain, head, and possibly the spinal cord do not develop normally. Newborn children with this severe disorder usually die shortly after birth.

Spina Bifida- This disorder is also called "open spine". It is a defect of the spinal column. Spina bifida has several forms.

Normal developement of the spina bifida child with little physical handicap is possible. Often, however, paralysis of the lower limbs, repeated urinary tract infections, hydrocephalus ("water on the brain"), mental retardation, and incontinence (inability to control bladder or bowel movements) occur.

The cause of neural tube defect is not known. The defects may be inherited; they may also be caused by environmental factors.

WHAT ARE THE CHANCES OF A BABY BEING BORN WITH A NEURAL TUBE DEFECT?

In the United States, about one to two live births per thousand involve a neural tube defect. About 3,000 to 6,000 of the 3,000,000 babies born each year will have a neural tube defect. These birth defects are about equally split between anencephaly and spina befida.

Of the 3,000 to 6,000 births that result in a neural tube defect, approximately 90 to 95 percent of the babies with such a defect will be born to women who have no "special risk" characteristics. The remaining 300 to 600 babies with neural tube defects will be born to parents who have "special risk" characteristics such as:

Women who have already given birth to a child with neural tube defect.

One or both parents have neural tube defects.

Women whose mothers or fathers have a history of neural tube defects.

WHAT IS THE AFP TEST?

The AFP blood test is a simple laboratory procedure. A sample of blood is taken from your arm and sent to a laboratory. The laboratory tests it to see how much AFP is present and then sends the results to your doctor.

If the results of the first blood test is elevated, then other tests are required before your doctor can determine whether your unborn baby has a neural tube defect.

WHEN SHOULD A PREGNANT WOMAN HAVE THE AFP BLOOD TEST?

The AFP blood test should be conducted at 15-20 weeks after the last menstrual period. The best time is 16-18 weeks. Unreliable results may be obtained if the blood sample is taken too early during pregnancy.

WHAT DO THE TEST RESULTS MEAN?

If your blood has normal levels of AFP the test is considered negative and there will be no need for further tests. Parents should understand that an AFP blood test result in the normal range cannot guarantee a normal baby at birth. The first AFP blood test may miss as many as 20 percent of those babies affected with neural tube defects. It can offer much greater assurance, however, that the baby is not likely to have an open neural tube defect.

The spinal malformation and nerve damage caused by this disorder hamper movement in varying degrees. Some individuals with spina bifida can walk by themselves. Others use braces and crutches or wheelchairs. Modern surgical and corrective techniques can help many children born with spina bifida lead healthy and productive lives.

An elevated result indicates that there are high levels of AFP in your blood. This does not always mean a neural tube defect is present. It means that you should have additional tests to see whether or not your unborn baby has a neural tube defect.

Research has also shown that in cases where there is an unexplained elevated blood level, these pregnancies may be at higher risk for other problems. Compared to normal pregnancy, these pregnancies with an unexplained elevated level may have a problem with premature births, stillborn infants, placental abruptions or problems with fetal growth.

After the first elevated blood sample, a second sample of blood will be taken from your arm. If the result of the second test is also elevated there is only about a 4 to 10 percent chance that your unborn baby has a neural tube defect. More likely, you may be carrying twins or your pregnancy may be further advanced than you think. For these reasons the doctor will want to conduct additional tests.

After the test was in use a few years, it was discovered that a low AFP level may also detect an infant with a chromosomal defect such as Downs Syndrome. The AFP test will detect 2 out of 3 of these cases like Downs Syndrome.

WHAT ARE OTHER TESTS THAT MAY BE NEEDED IF THE SECOND AFP BLOOD TEST IS ELEVATED?

Ultrasonography. This is a procedure in which sound waves are used to obtain a television-like picture of the unborn baby. The picture will enable your doctor to tell whether the blood test was elevated because of twins or because the week of pregnancy was estimated incorrectly. Ultrasonography can also show if an unborn baby has died, and often whether a neural tube defect is present. If ultrasonsography does give a reason for the elevated AFP values, amniocentesis may be recommended by your doctor.

Amniocentesis. In this test the doctor will take a sample of the amniotic fluid (the fluid filling the sac around the unborn baby) and send it to the laboratory to measure the AFP level in the fluid. If the AFP level is high and other possible causes (twins or an incorrect estimate of the week of pregnancy) have been ruled out, there is a high chance that the unborn baby has a neural tube defect. This test cannot tell how severe the defect will be or the possible degree of the handicap.

Although relatively safe, amniocentesis presents a small risk. You may want to discuss the nature of this risk with your physician.

SOME OTHER TESTS. If the level of AFP in the amniotic fluid is elevated, the physician may wish to attempt to locate the possible defect by means of high resolution ultrasonography (a more advanced and accurate type of ultrasonography). To further reduce the chance of a wrong diagnosis, a new test may be used in which amniotic fluid is analyzed for the presence of the enzyme acetylcholinesterase.

COUNSELING

You should ask your doctor about these tests, the risks that they present, how much they cost, where these tests are given, genetic counseling that is available, and other questions that may concern you.