



Note: Below are some facts about Enhanced AFP screening, a test offered to pregnant women. If you have any questions after reading this, please ask your nurse or doctor. Remember, it is your decision whether to have this test or not.

What is the Enhanced AFP Screening Test and why is it done?

This test helps identify pregnant women who are at increased risk for having a baby with either neural tube defect or Down syndrome. It is a blood test that is usually done between 16 and 18 weeks of pregnancy. Three different substances produced by the fetus (growing baby), called alpha-fetoprotein (or “AFP”), estriol and hCG, are measured in the mother’s blood. A positive (abnormal) test result does not necessarily mean that your baby has a problem. However, additional testing will be suggested.

What are neural tube defects?

Neural tube defects are defects of the spinal cord and/or the brain. They occur in about 1-2 of every 1,000 babies born. The two common types of neural tube defect are called “anencephaly”, and “spina bifida”. In anencephaly, the baby’s brain and skull do not develop normally. These babies are usually stillborn or die shortly after birth. In spina bifida, the spine does not completely close. How severely the baby is affected depends on the size and location of the opening in the spine. Most children with spina bifida have medical problems, and some are mentally retarded. Many individuals with spina bifida cannot control their bowels or bladder, or may require operations, leg braces and physical therapy. With medical treatment, many babies with spina bifida grow up to lead productive lives.

What is Down syndrome?

Down syndrome is a birth defect that occurs in about one of every 800 babies born. An extra #21 chromosome causes it. Children with Down syndrome are mentally retarded. All have similar physical features, and some have serious medical problems such as heart defects, which can usually be corrected with surgery. Although Down syndrome occurs more frequently as mothers get older, more than 80 percent of children with Down syndrome are born to women who are less than 35 years old. The Enhanced AFP Screening Test can help detect these pregnancies in women under age 35. If you are 35 years old or older at the time your baby is due, you should talk to your doctor about the best testing for you.

How is the Enhanced AFP Screening Test done?

The test requires taking a sample of blood from the mother, usually at 16-18 weeks of pregnancy. The laboratory tests the blood sample to see how much AFP, estriol, and hCG are present. A high AFP level means the fetus has an increased risk for having a neural tube defect. The levels of AFP,

estriol and hCG, plus the mother’s age, are all used to determine the risk of the fetus having Down syndrome.

What if my test is positive?

Your doctor or nurse will recommend further tests. In most cases, an ultrasound study (sonogram) will be recommended if you have not already had one. Sonograms can sometimes explain a positive test by finding that your pregnancy is either farther along or earlier than expected. If you are carrying twins, this may also explain a high AFP level. If the sonogram does not explain your test results, an amniocentesis and/or a more detailed ultrasound may be suggested. The equipment for the detailed ultrasound is available only in large medical centers. It can often detect spina bifida and some other birth defects, but cannot diagnose Down syndrome. Amniocentesis is a test where a small sample of the fluid that surrounds the fetus is obtained. This fluid can be tested to diagnose both spina bifida and Down syndrome.

If these diagnostic tests are recommended, you will be able to discuss them with a physician or genetic counselor who will explain them completely. You will be able to choose whether or not you want further testing. If a birth defect is found, your doctor, nurse or a genetics staff will be available to help you plan the remainder of our pregnancy.

Remember, fewer than 5 out of every 100 women with a positive test result are carrying a fetus with neural tube defect or Down syndrome.

If the Enhanced AFP Test is negative, does that mean everything will be all right with the baby?

A negative (or normal) result can be reassuring and gives your doctor useful information, but it does not guarantee a normal baby. The Enhanced AFP Test will detect most (about 80-85 percent), but not all fetuses with neural tube defects. About 6 out of 10 cases of Down syndrome will not be detected by the Enhanced AFP test. Also, many other birth defects cannot be diagnosed with this or any other test.

Enhanced AFP Screening is another way we can help provide optimal care for you.

- The test is simple and safe – just a blood test.
- It gives your doctor valuable information about your pregnancy and your developing baby, and may alert him/her to the presence of twins, a change in your delivery date, or other conditions which may alter your prenatal care.