

## **Expanded AFP Test (Quad Marker Test)**

### **What is the expanded AFP?**

The expanded AFP test is a screening test for pregnant women during the second trimester (between 15 and 20 weeks) of pregnancy who did not do a nuchal screening test. The test will help detect pregnancies at an increased risk for Down syndrome, Trisomy 18, and neural tube defects or abdominal wall defects. Occasionally, the test may also detect other chromosome abnormalities. This is not a diagnostic test; it simply indicates further testing may be advised. You will be asked to sign a consent stating your desire to take the test.

### **What does the screening test measure?**

This test measures four biochemical substances produced by fetal and placental tissues: AFP (alpha-fetoprotein), hCG (human chorionic gonadotropin), and uE3 (unconjugated estriol) and INH (dimeric inhibin-A).

### **What is Down syndrome?**

Down syndrome is a chromosome abnormality that causes mental retardation and certain types of birth defects. It is due to an extra copy of chromosome 21, so that three copies (trisomy) versus the normal two copies of this particular chromosome are present. Down syndrome affects approximately one in every 800 newborns. The chance of having a pregnancy affected with Down syndrome increases with increased maternal age. Women age 35 years and older are more likely to have a child affected with Down syndrome.

### **What is Trisomy 18?**

Trisomy 18 is a fatal chromosome abnormality that causes multiple birth defects and profound mental retardation. Few Trisomy 18 infants survive into childhood. Trisomy 18 results when the fetus has three, instead of the normal two, copies of chromosome 18. Like Down syndrome, the chance of an increased risk for fetal abnormality is determined by the test and then genetic counseling, ultrasound examination, and when needed, amniocentesis will aid in the diagnosis. Having a pregnancy affected with Trisomy 18 increases with increased maternal age.

### **What is a neural tube defect?**

A neural tube defect, such as spina bifida or anencephaly, results from a failure of complete closure of the neural tube during early fetal development. Spina bifida is an opening on the spine that exposes nerve tissue and can lead to paralysis and mental retardation. Anencephaly is an incomplete development of the brain that usually results in death.

### **What if the test result shows an increased risk?**

A positive screening test result does not mean an abnormality is present in the fetus. Often, incorrect dating of fetal age is the reason for a positive result and invasive testing may not be necessary. However, when an increased risk for fetal abnormality is determined, genetic counseling, ultrasound examination, and when needed, amniocentesis will aid in the diagnosis. If this testing is necessary, your physician will contact you and ask you to schedule an appointment with Obstetrix Medical Group (371-7111).

### **What does a negative test mean?**

A negative result indicates the risk that the fetus has Down syndrome or Trisomy 18 is not greater than that of a 35-year-old woman, and the risk for neural tube defects is not increased compared to that of the general population. However, a negative result does not completely exclude the possibility that the

fetus may have these abnormalities or other congenital abnormalities. The test detects approximately 75% of Down syndrome and trisomy 18 pregnancies, 80% of spina bifida, and 95% of anencephaly.

### **How accurate is the test?**

The test is not completely accurate. A baby may have a birth defect even though AFP levels are normal. A baby may be quite normal even though AFP levels are abnormal. For every 1,000 women tested, about 50 have an abnormal test result. Of these 50, only one or two with high AFP levels are carrying babies with a problem.

### **Who should have this test?**

All pregnant women should be offered AFP screening. If you are 35 and are having an amniocentesis, the blood test is not necessary as the amniotic fluid will be checked. If you did a nuchal screening test and are under 35, you should take the AFP only test which checks for spina bifida, not the expanded AFP test (which again looks for chemical evidence of Down syndrome but is less accurate than the nuchal translucency test

### **What are the benefits to taking the test?**

Most often, the test provides reassurance that your baby probably does not have a serious defect. Abnormal results can help you and your doctor manage your pregnancy more effectively. For example, if the test detects twins, your doctor can start providing the special prenatal care you need for a multiple pregnancy. When a brain or spinal defect is diagnosed, you and your partner can decide whether you want to continue the pregnancy. If you decide to continue the pregnancy, your doctor will be able to plan your delivery and optimize the outcome of the pregnancy for you and your baby.