

## AFP 4

### What you should know

#### **What is Maternal Serum Screening?**

Maternal Serum screening can identify pregnant women who are at an increased risk for having a baby with certain birth defects. When a woman has reached a certain point in her pregnancy (between 15 and 21 weeks), a simple blood test can determine her risk of having a baby with an open neural tube defect, Down syndrome, or trisomy 18. AFP 4 combines four different laboratory markers to offer enhanced prenatal screening and increased detection efficiency for Down syndrome.

#### **What is AFP 4**

AFP 4 is a screening test that measures four proteins in a pregnant woman's blood: AFP (alpha-fetoprotein), hCG (human chorionic gonadotropin), uE3 (unconjugated estriol), and dimeric inhibin A (DIA). The levels of these proteins, combined with clinical information about the pregnant woman, such as her weight, race, and whether she takes insulin, can help identify a woman who has a higher chance of having a baby with certain birth defects.

#### **What birth defects does AFP 4 screen for?**

##### **Open Neural Tube Defects**

Open neural tube defects, such as spina bifida, occur when the baby's neural tube, or spine, does not close completely during development, leaving the spinal cord exposed. Approximately 1 in 1000 babies has open spina bifida. The effects of open spina bifida range from bladder control problems to paralysis and mental retardation. The AFP 4 test can identify greater than 80% of pregnancies that may have open neural tube defects.

##### **Down Syndrome**

Down Syndrome, which occurs in about 1 in 800 births, is a disorder in which the baby has an extra chromosome. Chromosomes are the units of inheritance. In a normal pregnancy, each baby inherits a total of 46 chromosomes, 23 from the mother and 23 from the father. Sometimes an error in division will cause a baby to inherit too many or too few chromosomes. In Down syndrome, instead of inheriting one chromosome 21 from the mother and one chromosome 21 from the father, the baby inherits a total of three of chromosome 21. Down syndrome is often called trisomy 21. Babies with Down syndrome have some degree of mental retardation and often have physical abnormalities, such as heart defects. AFP 4 can detect 75% to 80% of Down syndrome pregnancies.

##### **Trisomy 18**

Trisomy 18 is also called Edwards syndrome. It is rare and only occurs in 1 out of every 8000 births. Like Down syndrome, trisomy 18 is a chromosome problem, but it is caused by the new baby having three of chromosome 18. Babies with trisomy 18 have significant physical birth defects and severe mental retardation. Few live beyond the age of one year. AFP 4 can detect more than 60% of trisomy 18 pregnancies.

#### **What does it mean if my AFP 4 test is negative?**

A negative test result significantly reduces the likelihood that your baby has an open neural tube defect, Down syndrome, or trisomy 18; however, AFP4 is not diagnostic and cannot completely rule out the possibility of these problems. AFP 4 does not screen for any other defects.

**Does a positive AFP 4 test result mean that my baby has a birth defect?**

No. AFP 4 is a screening test; it cannot diagnose problems with your baby or pregnancy. The test can only identify those women who are more likely to have a baby with an open neural tube defect, Down syndrome, or trisomy 18. Normally, a positive AFP 4 result will only mean that more diagnostic testing may be offered.

**What other tests are offered if my AFP 4 test is positive?**

Follow-up options are determined privately between a woman and her physician. In general, most women with positive test results are referred for genetic counseling, ultrasound, and amniocentesis.

**What happens next?**

If your AFP 4 test is positive, your physician may recommend one or more of the following:

**Genetic Counseling**

Genetic counseling is a communication process between a patient and a trained specialist, such as a certified genetic counselor, perinatologist (high-risk pregnancy physician), or the patient's obstetrician. Genetic counseling is designed to help you understand your test results and follow-up options and may include a discussion of your family and pregnancy history.

**Ultrasound**

Ultrasound is the use of high frequency sound waves and a computer to create images of the developing baby. Ultrasound is an important tool to help determine gestational age, or how far into the pregnancy you are. The levels of the proteins measured in the AFP 4 test vary with each week of pregnancy, so knowing the exact gestation age is an essential part of the test. If the gestational age used during the AFP 4 analysis is not precise, your test results may not be accurate. If ultrasound dating changes your gestational age by 10 days or more, your physician will ask the lab to recalculate your test results. Your result may change after the gestational age adjustment.

Ultrasound may also reveal the presence of twins, which can affect the AFP 4 result. Certain birth defects, such as open spina bifida, may also be seen on ultrasound. Babies with Down syndrome and trisomy 18 may have certain features that can be seen on ultrasound, but, in general, neither can be diagnosed by ultrasound alone.

**Amniocentesis**

Amniocentesis is a test in which a trained specialist, such as a perinatologist or an obstetrician, removes a small amount of fluid from around the baby. There are some risks associated with this test that should be discussed with your physician. A thin needle is inserted into your abdomen under ultrasound guidance, and a few tablespoons of fluid are removed. This fluid contains skin cells from the developing baby. These cells are taken to the lab for chromosome analysis. This test can help to diagnose more than 99% of Down syndrome and trisomy 18, as well as other numeric and structural chromosome problems.

AFP is also measured in the amniotic fluid and, if open spina bifida is suspected, a spinal protein called AChE (acetylcholinesterase) is also measured. This combination of test can help to diagnose more than 98% of cases of open spina bifida.

**Remember:**

AFP 4 is a blood test used to help your doctor identify pregnancies that may be at increased risk for open spina bifida, Down syndrome, or trisomy 18. It does not diagnose birth defects, but can help identify those people with a higher risk who might benefit from additional testing.