

IF THE AFP+ TEST INDICATES AN INCREASED RISK FOR A BIRTH DEFECT, WHAT WILL HAPPEN?

Your doctor will discuss your AFP+ result with you and discuss your options. These may include having a repeat blood test or an ultrasound to confirm how far along you are. In some cases, your doctor may offer amniocentesis. Amniocentesis is performed by removing a small amount of fluid from the amniotic sac (the bag of water that surrounds the fetus). The fetal chromosomes can then be examined in this fluid to check for Down syndrome and Trisomy 18, and testing can be done to detect neural tube defects and abdominal wall defects. You may be referred to the Reproductive Genetics Program for further information about your test results and about amniocentesis. Remember, most women who have further testing get normal results and have healthy babies.

MORE QUESTIONS?

If you have further questions about AFP+ screening, discuss them with your doctor or call **(585) 487-3480**.



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AFP+ SCREENING

Would you want to take a simple test to let you know if you have a higher than average chance of having a baby with certain birth defects?

If you were told you had an increased risk, would you want further testing to tell for sure?

If you think the answer to these questions is “yes,” then you should consider Maternal Serum AFP+ (Quad) Screening.

WHAT IS AFP+ SCREENING?

AFP+ Screening measures the levels of certain substances (alpha-fetoprotein (AFP), unconjugated estriol, human chorionic gonadotropin, and Inhibin-A) in a pregnant woman’s blood. These substances come from the fetus and placenta and are found in the blood of all pregnant women. It is normal for the levels of these substances to vary among different women, but some women with particularly high or low values may be at an increased risk for having a baby with certain birth defects.

The AFP+ test is called a “screening” test because it cannot tell for certain whether the fetus does or does not have a birth defect. It can identify women who have a greater than average risk for having a baby with certain birth defects. These women may then choose to have other tests (such as ultrasound or amniocentesis) that can help tell with greater certainty if a birth defect is present. About 7 out of every 100 women who have AFP+ testing will need some further testing, but most of the time those women will deliver a normal, healthy baby.



WHAT BIRTH DEFECTS CAN BE DETECTED BY AFP+ SCREENING?

NEURAL TUBE DEFECTS (SPINA BIFIDA AND ANENCEPHALY): Neural tube defects occur in only about one pregnancy out of every 1,000. They are birth defects in which the brain (anencephaly) or part of the spinal cord (spina bifida) does not form normally and may not be covered with skin or bone. Some babies with neural tube defects are stillborn or die shortly after birth. Children who survive usually have medical problems, some more severe than others. Complications may include infections, hydrocephalus (“water on the brain”), and sometimes learning problems or mental retardation. Spina bifida often results in paralysis of the legs and impaired bladder and bowel control. Elevated levels of AFP may indicate an increased risk for a fetus with a neural tube defect.

ABDOMINAL WALL DEFECTS: These birth defects are somewhat less common than neural tube defects. They are caused by an opening in the layer of muscle and skin near the “belly button.” Although these defects can be serious, they can usually be corrected with surgery. Elevated levels of AFP may also indicate an increased risk for an abdominal wall defect.

DOWN SYNDROME: Down syndrome is a condition where the fetus has an extra 21st chromosome. Therefore, there are 47 chromosomes instead of the normal 46. The chromosomes contain the genetic material that determines our inherited characteristics. Down syndrome results in mental retardation and sometimes in physical problems such as heart defects. Although the risk of having a child with Down syndrome is higher in older women, it also can occur in young women. The AFP+ test is used to indicate whether a woman is at an increased risk for having a baby with Down syndrome. If the AFP+ test indicates that the chance for Down syndrome is high enough, further testing may be recommended.

TRISOMY 18: Trisomy 18 is caused by an extra 18th chromosome. It is a very severe birth defect causing many health problems and severe mental retardation. The AFP+ test indicates an increased risk for Trisomy 18 in only about 1 out of every 200 women, and these women can have further testing to know for sure if the baby is healthy.

WHAT FACTORS MIGHT INFLUENCE THE AFP+ TEST RESULT?

Many factors must be taken into consideration to know if your AFP+ test results are normal for you. These include how far along you are in your pregnancy, your weight, your race, and whether or not you are carrying twins. Things such as your diet, the number of times you have been pregnant, or the results of other blood tests do not affect the AFP+ test.

WHEN SHOULD AFP+ TESTING BE DONE?

The AFP+ test is best performed between 15-18 weeks from the beginning of the last menstrual period.

IF THE AFP+ TEST IS NORMAL, CAN THE MOTHER BE CERTAIN EVERYTHING IS ALL RIGHT?

The answer is “No.” Although results of the AFP+ test are useful, they do not guarantee a healthy baby. AFP+ testing is a screening test and will detect approximately 90 percent of fetuses with open neural tube defects and approximately 75-80 percent of fetuses with Down syndrome. There are other kinds of problems the AFP+ test cannot detect. But remember, most babies are born healthy.