

AFP+QUAD Test

Commonly Asked Questions

What is the AFP+ Quad Test?

It is a maternal serum test done between the 15th and 22nd week of pregnancy that allows us to measure certain substances that come from the developing fetus and placenta and are present in the mother's serum. The substances that we measure are called alpha-fetoprotein (AFP), total β hCG, estriol, and inhibin-A. By finding out what the levels of these substances are in your serum, we can learn certain things about your developing baby.

What can the AFP+ Quad Test tell me about my pregnancy?

It can tell you whether you are at high risk for having a baby with either an open neural tube defect, Down syndrome or Trisomy 18. Ultimately, about one in twelve patients will have a "screen positive" AFP+Quad Test result. If your test result is "screen positive" it does not necessarily mean that your baby has one of these disorders, but it does mean that your doctor will suggest some additional testing.

What are neural tube defects (ontd)?

Neural tube defects are a group of birth defects which include open spina bifida and anencephaly. One or two babies out of every 1000 is born with a neural tube defect.

Open spina bifida is an incomplete closure of the spine. It varies in severity depending on where it's located on the spine and how big the opening is. Surgery to close the opening is usually performed shortly after birth and additional surgery is often necessary later in infancy and childhood. Open spina bifida usually causes some amount of paralysis from the waist down and interferes with bowel and bladder control. It often leads to a condition called hydrocephalus (water on the brain) and can sometimes cause mental retardation. Almost one third of babies born with open spina bifida will not survive past age five.

Babies with anencephaly have a large part of the skull missing and the brain is not properly formed. They always die before or very soon after they are born.

The level of AFP in the mother's serum tends to be elevated when there is a pregnancy affected by an open neural tube defect. About 90% of all pregnancies with open neural tube defects will be identified through AFP+Quad Testing. Closed neural tube defects (when the spinal opening is covered with skin or thick tissue) will not be picked up by AFP testing.

What is Down syndrome?

Down syndrome is a common birth defect occurring in about one in every 700 babies. It is a disorder in which an extra chromosome (the number 21 chromosome) is present in the cells of the developing fetus from the time of conception.

Down syndrome usually occurs unexpectedly and about 98% of the time is not inherited. Although Down syndrome occurs more frequently as mothers get older, about 75% of babies with Down syndrome are born to women who are younger than 35.

Down syndrome is always associated with mental retardation, often in the mild to moderate range. Children with Down syndrome have variable but predictable physical characteristics. About 50% have medical problems such as heart defects. Often surgery can correct these defects.

The levels of AFP and estriol in the mother's serum tend to be low while the levels of total β -hCG and inhibin-A tend to be elevated in pregnancies affected by Down syndrome. Overall, approximately 80% of all pregnancies with Down syndrome will be identified through the AFP+Quad Test. However, detection rates vary with maternal age, ranging from 70% in women under 35 to more than 90% in women 35 and older.

Can other abnormalities be identified?

Yes. The risk of two other disorders can be estimated. One is Trisomy 18, a rare and usually fatal disorder caused by the presence of an extra number 18 chromosome in the cells of the developing baby. The risk of Trisomy 18 can be estimated using AFP, uE3 and total β hCG, and is reported only when the risk is high. The second is called Smith-Lemli-Opitz syndrome, a genetic disorder caused by an error in the synthesis of cholesterol. Smith-Lemli-Opitz syndrome is associated with many problems in the developing baby, most important are mental retardation and poor growth. The risk of Smith-Lemli-Opitz syndrome can also be estimated using AFP, uE3 and total β hCG, is reported only when risk is high.

Why do you take age into account?

Any woman can have a baby with Down syndrome but the chance of this happening increases as a woman gets older. We use age as one of the factors when working out your risk of pregnancy with Down syndrome. It means that an older woman is more likely to have a result in the higher risk groups (screen positive) and be offered a diagnostic test.



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What does it mean if my AFP+ Quad Test result is screen positive?

A positive AFP+Quad Test means that you are in a higher risk group for having a baby with an open neural tube defect or a chromosomal abnormality. However, it does not prove by itself that there is anything wrong with the pregnancy. In fact, only a small number of women with screen positive results will have an abnormal baby. If you have a "screen positive" result, you should consider specific counseling to discuss further testing.

What are the tests that will be offered if my AFP+ Quad Test is screen positive?

It depends on your particular result. In most cases, after counseling, an ultrasound study (sonogram) will be recommended. Depending on your AFP+Quad Test and the results of the sonogram, an amniocentesis or further diagnostic ultrasound may be recommended.

What is ultrasound and what will it show?

An ultrasound machine uses sound waves to look at the developing baby. One of the things it can do is check fetal age. Many women will have a "screen positive" AFP+Quad Test result because the dates of their pregnancies have been misjudged. When this date is adjusted, the test result may become "screen negative". Occasionally twins will be discovered and will explain the "screen positive" result.

If the gestational age is correct and you are not carrying twins, either amniocentesis or level II sonography may be suggested. Level II sonography is a detailed examination of the fetus. It cannot be used to diagnose Down syndrome or Trisomy 18, but often can identify spina bifida and other fetal abnormalities.

What is amniocentesis and what will it show?

Amniocentesis is a procedure in which the doctor obtains a small sample of fluid that surrounds the developing fetus. The sample is then sent to the laboratory for testing. This fluid sample can be used to diagnose both chromosomal problems such as Down syndrome and Trisomy 18, as well as open neural tube defects such as spina bifida. Amniocentesis is an invasive procedure, which means that there is a small risk of miscarriage (less than 1 in 200) associated with it. Results of the test for Down syndrome and Trisomy 18 will take about 7-14 days. Results of the test for spina bifida will take about 2-5 days.

What happens if a birth defect is discovered through the AFP+ Quad Test ?

Your healthcare provider and/or genetic counselor will be available to discuss your baby's diagnosis in detail and options available to you. One option would be to continue the pregnancy and make arrangements for appropriate medical services at and after delivery. Other options such as adoption and termination of pregnancy will be discussed with you by your healthcare provider.

If my AFP+ Quad Test is screen negative, does that mean that my baby will be normal?

A "screen negative" result means that your risk for a child with a neural tube defect is 1 in 1000 or less. It also means that your risk for a child with Down syndrome is less than that of a 35-year-old woman.

It is never possible to be sure that your baby is going to be normal. The AFP+Quad Test will allow us to identify at least 5 out of 6 cases of open spina bifida and almost all cases of anencephaly. It can also lead to the diagnosis of about 8 out of every 10 cases of Down syndrome and Trisomy 18. Remember, a screening test can never completely rule out the possibility of an open neural tube defect, Down syndrome or Trisomy 18. There are also many other birth defects that cannot be identified with this test.

What are the advantages of having the AFP+ Quad Test?

The test may give you and your healthcare provider important information about your pregnancy and your developing baby. Twins may be discovered or your expected date of delivery may be corrected so your prenatal care and visits can be adjusted accordingly.

If your baby is found to have a serious birth defect, you can receive professional counseling about how your child's physical and mental development may be affected. The individual capabilities and potential of children with birth defects are considerations which you may wish to discuss with your genetic counselor or with other healthcare providers. Other options such as adoption and termination of pregnancy may also be discussed.

Further information and support are available through groups such as your local Down Syndrome Society and Spina Bifida Association as listed below:

March of Dimes www.marchofdimes.com
National Down Syndrome Society www.ndss.org
National Association for Down Syndrome www.nads.org
Smith-Lemli-Opitz Syndrome www.smithlemliopitz.org
Spina Bifida Association www.sbaa.org
Trisomy 18 www.trisomy.org



AFP+QUAD INFORMED CONSENT

I have read and understand the information in this pamphlet regarding screening for the AFP+QUAD Test.

- Yes, I want to have the AFP+QUAD Test.
 No, I do not want to have the AFP+QUAD Test.

Patient Name: _____

Patient Signature: _____

Date: _____ **IMPORTANT:** Retain Copy in Patient File



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The AFP+QUAD Test

Information for Patients

Second Trimester Risk Assessment for Down Syndrome and Open Neural Tube Defects



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