

Answering Your Questions About: Quad Screening

What is Quad screening?

Quad screening (sometimes called AFP4 screening) is a blood test that screens for open neural tube defects (NTDs) such as spina bifida and anencephaly, for abdominal wall defects and for chromosome changes such as Down syndrome (trisomy 21) and trisomy 18 (Edwards syndrome). A sample of blood from the mother is obtained between 15 and 20 weeks of pregnancy, and the amount of alpha-fetoprotein (AFP), human chorionic gonadotrophin (hCG), unconjugated estriol (uE3) and dimeric inhibin (DIA) are measured. A refined estimate of a woman's risk for carrying a child with one of the above conditions is calculated using this screening. An elevated level of AFP in the mother's blood may indicate an increased chance of having a baby with an open NTD or abdominal wall defect. The risk for Down syndrome is increased when the levels of AFP and uE3 are lower and the levels of hCG and DIA are higher than expected. The risk for trisomy 18 is increased when the levels of AFP, hCG, and uE3 are lower than expected.

What if the Quad screen results are abnormal?

An abnormal screening result does not mean that your baby has a neural tube defect, Down syndrome or trisomy 18. Many normal pregnancies have higher or lower levels of AFP, uE3, hCG or DIA than expected. An abnormal test simply means further evaluation is necessary to determine the significance of the screening results. Additional tests may include a second blood test, an ultrasound examination or an amniocentesis. Most women who have abnormal quad screening results go on to deliver healthy babies.

How accurate is a normal Quad screening result?

A normal result from this screening does not guarantee that the baby does not have one of these conditions. Almost all cases of anencephaly will be detected with quad screening, but only four out of five cases of spina bifida (80 percent), approximately three out of four cases of Down syndrome (70-75 percent) and about three out of five cases of trisomy 18 (60 percent) will be detected. There are also other birth defects that do not cause quad screening levels to be abnormal.

Who should have Quad screening?

Anyone who desires more information about her baby's development should consider the screening. More than 95 percent of babies with neural tube defects or Down syndrome are born to families with no history of these conditions. The quad screening can also alert the doctor to complications of pregnancy that might otherwise go undetected. The diagnosis of a problem before birth may sometimes lead to special management of the pregnancy and delivery to improve the outlook for the baby.

What is Down syndrome?

Down syndrome is a chromosomal abnormality. Chromosomes carry our genetic information. Typically, a person has 46 chromosomes in each cell of his or her body. Anytime a child receives extra or missing chromosomal material, developmental problems may occur. A person with Down syndrome has 47 chromosomes because he or she has an extra copy of chromosome 21. Because of this extra chromosome, people with Down syndrome have some degree of mental retardation (typically mild to moderate). They also have a predisposition to certain medical conditions, such as heart defects.

The risk of having a baby with Down syndrome increases with a woman's age. Women who are 35 or older at the time of delivery have an increased risk of having a baby with Down syndrome or other chromosome abnormality, and should be offered amniocentesis in their pregnancies. Even though a woman is not considered to have a high risk until she reaches 35 years of age, babies with Down syndrome can be born to women of any age.

Maternal Age	Second trimester risk of Down syndrome
25	1/890
30	1/690
35	1/270
40	1/84
45	1/25

What is Trisomy 18?

Trisomy 18 is a more severe chromosomal condition that causes profound mental retardation and multiple birth defects. Few babies with trisomy 18 live more than one year, though some do survive into childhood. Like Down syndrome, the chance to have a baby with trisomy 18 increases with maternal age.

What are spina bifida and anencephaly?

Neural tube defects (NTDs) are serious conditions that involve incomplete development of the brain, spinal cord and/or protective covering of these organs. The two types of NTDs that may be detected with the AFP screening are anencephaly and spina bifida. Anencephaly is a lethal condition that occurs when there is incomplete development of the skull and brain. Spina bifida occurs when there is an opening along the spine. The severity of this condition varies a great deal. Some individuals have learning disabilities and require a wheelchair for mobility, while others may have normal intellect and mobility.

Possible explanations for an abnormal Quad screening result:

- Normal variation
- The pregnancy is earlier or further along than expected
- The mother is carrying twins
- Bleeding or changes in the placenta
- The baby has spina bifida, Down syndrome or other birth defect

*When individual levels are extremely high or low, this may indicate a need to offer additional testing or monitoring of the pregnancy.

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