



KIRKLAND 425 899 2200; BELLEVUE 425 688 8111

Second Trimester Multiple Marker Screening (The Quad Screen)

What is this test? The quad screen is a blood test that measures the levels of certain hormones and proteins in a pregnant woman's blood.

Why is this test offered to me? The purpose of the quad screen is to measure the chance that your baby has one of three birth defects: neural tube defects, Down syndrome, and trisomy 18.

Will this test find all birth defects? NO!! Most birth defects are not detected by this or any other test.

What is a neural tube defect? Neural tube defects are birth defects that affect a baby's spine or brain. Spina bifida is an opening along the spine where the bones and skin do not cover the spinal cord. The effects range from poor control of the legs, leg paralysis, loss of bowel and bladder control, to severe physical disability and quadriplegia. About 1 in 1000 babies are born with spina bifida. Anencephaly is the most severe form of neural tube defect in which the baby's brain does not form completely. Babies with anencephaly do not survive. About 1 in 2000 babies are born with anencephaly. For more information: http://www.sbaa.org/html/sbaa_facts.html

What is Down syndrome? Down syndrome is the most common genetic form of mental retardation. It is not inherited, except in rare cases. It is caused when a baby receives an extra chromosome (package of genetic instructions) from the egg or sperm at the time of conception. In the case of Down syndrome, the extra chromosome is always a number 21. Adults with Down syndrome can be mildly or severely mentally retarded, but most are in the moderate range. Adults with Down syndrome may have jobs but will rarely be able to live independently. About 1 in 800 babies are born with Down syndrome. For more information: <http://www.ndss.org/aboutds/aboutds.html>

What is trisomy 18? Trisomy 18 is also a chromosome disorder, but the extra chromosome is a number 18. Most babies conceived with trisomy 18 are miscarried or stillborn, but rarely they can survive years after birth. Children with trisomy 18 often have birth defects such as clubfoot or heart defects, and are always severely mentally retarded. About 1 in 5000 babies are born with trisomy 18. For more information: <http://homepages.tig.com.au/~karens/t18.html>

How does the test work? The lab measures AFP, HCG, estriol, and inhibin A in your blood. Only babies make these proteins and hormones, which pass through the placenta and sac into their mother's blood. Your amounts are compared to normal levels to see whether your levels are normal, high, or low. Using these levels, and other factors such as your age, your weight, and your race, the lab calculates the chance your baby has a neural tube defect, Down syndrome, or trisomy 18.

Why would I take this test? You may want to know during the pregnancy whether your baby has one of these three birth defects. The quad screen can give you some information about the health of your baby before birth, without any physical risk to the baby. If the results are abnormal **and** follow-up tests diagnose a birth defect, you can discuss all your options with trained healthcare providers



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such as genetic counselors and perinatologists. Some parents decide to end the pregnancy. For those who continue their pregnancy, knowing the diagnosis can help them choose the best delivery and treatment plans. This may improve the short-term and/or the long-term outlook for the child.

Why do some people NOT take the quad screen? Taking the test itself has no risk to the baby, and the only risk to you is pain from having your blood drawn. BUT before having this test done, you should think about whether you want this type of information about your baby. Getting an abnormal result and deciding whether or not to have more tests causes most people a lot of stress. If you feel you would never end a pregnancy under any circumstances, and you do not want to know before birth whether your baby has a neural tube defect, Down syndrome, or trisomy 18, the quad screen may not be the right choice for you.

When is the test done? A common time for the quad screen is between 16 and 18 weeks of pregnancy. It can be done as early as 15 weeks and as late as 22 weeks.

How long do results take? Results are usually available within 2-3 days.

Does an abnormal (or “positive”) test result mean the baby has a birth defect? NO!! The quad screen cannot diagnose a birth defect. About 1-2% (1 to 2 in 100) of all women will have a quad screen “positive” for a neural tube defect; about 5% (5 in 100) of all women will have a quad screen “positive” for Down syndrome; and about 0.5% (1 in 200) of all women will have a quad screen “positive” for trisomy 18. *Most of these women will have healthy babies.* Abnormal test results ONLY mean that the chance of one of these three birth defects is higher and that you should be offered other tests to try to find the cause of the abnormal results. A result of 1 in 100 chance (1%) of Down syndrome from the quad screen means 99 out of 100 times (99%) the baby does NOT have Down syndrome.

What happens next after an abnormal test result? Most women who have an abnormal quad screen result are referred to specialists, such as those at Eastside Maternal-Fetal Medicine. The first part of the visit is spent with a genetic counselor, who spends as much time as needed answering questions parents have. The genetic counselor reviews the quad screen results and discusses the follow-up tests available to the parents. A family and pregnancy history is also taken, to identify other, unrelated, risks to the baby. Parents then meet with a radiologist or a perinatologist to do a high-resolution ultrasound. Some parents also decide to do an amniocentesis.

Does a normal (or “negative”) test result mean the baby does not have a birth defect? NO!! Normal test results only mean that the chance of these three birth defects is not higher than certain levels set by the laboratory. The quad screen will be “negative” in (it will not detect) about 5-10% of babies with anencephaly, 20-25% of babies with spina bifida, 20-25% of babies with Down syndrome, and 20-40% of babies with trisomy 18.

Where can I get more information? Your healthcare provider may be able to give you more information. Also: http://www.vtmednet.org/~m145037/vhgi_mem/triple/toc.htm