Allina Health

Triple Test Screening for Neural Tube Defects and Chromosome Abnormalities

General Information

Most babies are born healthy. But, about 1 to 2 out of 1,000 babies are born with a neural tube defect (such as anencephaly or spina bifida). Most of these babies are born to parents who have neither a family history of neural tube defects nor any obvious risk factors.

Fewer than 1 percent of all babies are born with chromosome abnormalities (too much or too little genetic information). The risks increase with maternal age. However, most babies with chromosome abnormalities are born to mothers who are younger than age 35 and have no reason to think they are at high risk. A serum test might alert unsuspecting parents to these problems.

Neural Tube Defects and Chromosome Abnormalities

- Neural tube defects develop in a baby's central nervous system before 28 days and may affect the spine or brain.
  - — Spina bifida is an opening into the baby's spine. Surgery can help close this defect but handicaps may result, such as paralysis from the waist down, walking problems, and lack of bladder and bowel control. About 80 to 90 percent of children born with spina bifida develop hydrocephalus (build up of spinal fluid in the brain). Some children can have learning disabilities or mental retardation. Normal intelligence is often possible.
  - — Anencephaly occurs when the baby's brain does not form properly due to an "opening" in the top portion of the developing neural tube. Babies are often delivered stillborn or die within hours (or days) of birth.
- Humans have 46 chromosomes in each cell. These chromosomes carry genetic information, which directs growth and development. If a baby is born with an extra chromosome, he or she will likely have a problem with mental and physical development. Two examples of such problems are:
  - — Down syndrome, which causes mild to moderate mental retardation and may cause heart defects and other physical disabilities.
  - — trisomy 18, which causes many physical problems and much more severe mental retardation than Down syndrome. Most babies born with trisomy 18 do not survive the first year of life.

The Triple Test (Serum Screening)

You may choose to take the triple test (serum screening) when you are between 15 and about 21 weeks in your pregnancy (the time varies with the lab). This screening test may identify if your unborn baby has a higher than expected chance of certain birth defects. This test is designed to help better identify pregnancies at high risk for neural tube defects and some chromosome problems so women can seek more testing (called diagnostic testing).

The screen can be normal in women who have affected children and, as is true for most screens, most women alerted to an increased risk by this screen will have healthy babies. (It may not be as helpful for women who are already identified as high risk due to family history or other concerns. A personal and family history check is also important.)

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The triple test measures three of your baby's proteins in your blood:, alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG) and unconjugated estriol (uE3).

High levels of AFP may indicate an increased risk for an open birth defect such as spina bifida. Other causes of high AFP are inaccurate gestational age, twins or triplets, or placenta problems. An ultrasound or amniocentesis can help clarify the reason for a high AFP. Most are healthy babies.

When the levels of AFP, hCG and estriol are considered with your age, the lab can estimate the chance that your baby has Down syndrome and trisomy 18. This is just a more precise guess, not a firm yes or no answer.

The test is called positive when the chance of having a baby with Down syndrome is at least the risk faced by a woman who is 35 years old (1 in 270). Amniocentesis is offered to check the baby's chromosomes. This is a diagnostic test that says "yes" or "no."

The triple test is a screen, not a diagnostic test. Not all women who carry babies with chromosome problems or neural tube defects will have a "positive" screen. Not every abnormal screen means the baby has a problem and a normal result means a low risk for Down syndrome, trisomy 18 and spina bifida. A normal result does not rule them out.

Reliability of the Triple Test*

The triple test will be "positive" in:

- 95 to 100 percent of women who carry babies with anencephaly
- 80 to 90 percent of women who carry babies with an open spina bifida
- 60 to 70 percent of the women who carry babies with Down syndrome and a smaller percentage of women who carry babies with other chromosome problems.

(*A quadruple test is more effective at alerting women who carry babies with Down syndrome and trisomy 18, and is just as good at finding babies with spina bifida.)

Many women who have healthy babies will also have a "positive test." Diagnostic tests will help detect those who truly have a birth defect from those who don't. If you are at an increased risk of having a baby with a neural tube defect or chromosome abnormalities, you will be offered diagnostic tests. They can help identify or rule out the suspected birth defects.

What to Remember About the Test

The test is your choice. Some women find having the test is reassuring. Others would rather not have the information or become anxious with an abnormal result until more testing is done. The test results may help you make decisions about other testing options. Some women who are already high risk for other reasons may have other diagnostic tests without serum screening.

Genetic counseling is available to help interpret your results and talk with you (and your partner) about what is right in your case, and to be sure you are aware of all risks and options. Please remember the following about the triple test:

- Not every positive test means a baby has a birth defect. Most women who have positive tests have healthy babies.
- Not every negative test means a baby will be born healthy. Not all cases of neural tube defects or Down syndrome can be predicted by this screening test. Diagnostic tests can help provide more information. Please remember, no test can rule out all birth defects.
- About 7 percent of women who have this test will have a positive screen. (More follow-up tests will be offered.) If you are older than age 35, the chance of a positive screen is much higher than that of a younger women. Sometimes, there is no way to explain a positive test result.

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