If My Test Result Is Positive, What Happens Next?

Follow-up options are discussed between you and your doctor. If your screening test is positive, your doctor may recommend one or more of the following:

- **Genetic counseling**. Genetic counseling is designed to help you understand your test results and follow-up options and may include a discussion about your family and pregnancy history. Genetic counseling may be provided by a certified genetic counselor, a perinatologist (a specialist in high-risk pregnancies), or your own obstetrician.
- **Ultrasound**. This procedure uses high-frequency sound waves and a computer to create images of the developing baby. In the second trimester, a detailed ultrasound examination of the baby may be able to identify some birth defects, such as open spina bifida. Babies with Down syndrome and trisomy 18 may have certain features that can be seen on ultrasound, but in general, neither can be diagnosed by ultrasound alone.
- **Amniocentesis**. This procedure is usually performed after the 15th week of pregnancy. Ultrasound is used to guide a thin needle through the abdomen into the uterus, and a small amount of fluid (amniotic fluid) from around the baby is removed. The cells in the fluid are examined in the laboratory to determine whether a chromosomal abnormality like Down syndrome or trisomy 18 is present. Amniocentesis can diagnose most chromosomal abnormalities but cannot diagnose or identify all birth defects. Alpha-fetoprotein (AFP) is also measured in the amniotic fluid, and if open spina bifida is suspected, a spinal protein called acetylcholinesterase (AChE) is measured as well. This combination of tests can diagnose most, but not all, babies with open spina bifida.[6]

References

1. Wald NJ, Watt HC, Hacksaw AK. Integrated screening for Down's syndrome based on tests performed during the first and second trimesters. *New England Journal of Medicine*. 1999 Aug 12; 341(7):461-467.
2. Wald NJ, Rodeck C, Hackshaw AK, Walters J, Chitty L, Mackinson AM. First and second trimester antenatal screening for Down's syndrome: the results of the Serum, Urine and Ultrasound Screening Study (SURUSS). *Health Technology Assessment*. 2003;7(11):1-77.
3. American College of Obstetricians and Gynecologists. Maternal Serum Screening. *Educational Bulletin*. 1996; September (228):3,5-8.
4. Nussbaum Robert L., editor. *Thompson and Thompson Genetics in Medicine*. 6th ed. Philadelphia: W.B. Saunders Company; 2001.
5. Palomaki GE, Neveux LM, Kinght GJ, Haddow JE. Maternal serum-integrated screening for trisomy 18 using both first trimester- and second-trimester markers. *Prenatal Diagnosis*. 2003 Mar; 23(3):243-247.
6. Bradley LA, Palomaki GE, McDowell GA. ONTD Working Group, ACMG Laboratory Quality Assurance Committee. Technical standards and guidelines: Prenatal screening for open neural tube defects. *Genetics in Medicine*. 2005 May-Jun;7(5):355-369.

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Serum Integrated Screening

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What Is Serum Integrated Screening?

Serum integrated screening is a two-stage test offered during pregnancy to identify women who are at increased risk of having a baby with Down syndrome.[1,2] It also permits screening for open neural tube defects, such as open spina bifida, and the identification of pregnancies at high risk for trisomy 18.

The **first stage** of serum integrated screening is offered between the 10th and 13th weeks of pregnancy and requires a blood sample to measure a protein called pregnancy-associated plasma protein A (PAPP-A).

The **second stage** of serum integrated screening is offered between the 15th and 21st weeks of pregnancy and requires a blood sample to measure four additional substances in a pregnant woman's blood: alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), unconjugated estriol (uE3), and dimeric inhibin A (DIA).

The measurements from the 2 blood samples are combined with information about you, such as your age and weight, to determine your baby's risk for having Down syndrome. The AFP measurement is used to screen for open neural tube defects, and the combination of markers may identify babies at increased risk for trisomy 18.

What Are Down Syndrome, Trisomy 18, and Open Neural Tube Defects?

Down syndrome and trisomy 18 are disorders caused by chromosomal abnormalities. Chromosomes are present in every cell of the body and contain genetic information that helps determine how we look, how our bodies grow and develop, and our health. A baby normally receives 23 chromosomes from each parent, so there are a total of 23 pairs of chromosomes in each cell. Sometimes a baby can be born with too many or too few chromosomes. Errors in the number of chromosomes may cause a variety of birth defects, ranging from mild to severe.

In **Down syndrome**, also called trisomy 21, the baby has an extra copy of the #21 chromosome. All babies with Down syndrome have some degree of mental retardation and often have physical abnormalities, such as heart defects. About 1 in 800 babies is born with Down syndrome.[3] Serum integrated screening is expected to detect 88% of pregnancies with Down syndrome in the early second trimester, with 6.0% of unaffected pregnancies having positive results (ie, a false-positive rate of 6.0%).[2]

Trisomy 18 is also known as Edwards syndrome. Babies with this condition have an extra copy of the #18 chromosome. Trisomy 18 causes severe mental retardation and physical abnormalities. Most babies with trisomy 18 die within the first year of life. Trisomy 18 is rare, occurring in 1 in every 7500 births.[4] Serum integrated screening is expected to detect 90% of pregnancies with trisomy 18 with little increase in the 6% false positive rate.[5]

Open neural tube defects (ONTD), such as open spina bifida, occur when the baby's neural tube, or spine, does not close completely during development. About 1 in 1000 babies is born with open spina bifida.[3] The effects of open spina bifida range from bladder control problems to paralysis and hydrocephalus. Screening is performed by measuring AFP levels in the second trimester and will identify 80% of pregnancies with open spina bifida with a false-positive rate of 1% to 3%.[3,6]

What Does it Mean if My Serum Integrated Screening Test Is Negative?

A negative test result indicates that, compared to unscreened women, the risk that your baby has Down syndrome is significantly reduced, but it cannot completely rule out the possibility of this condition in your baby. The measurements used in the serum integrated screening test can also be used to identify pregnancies with a relatively low risk of trisomy 18 and open neural tube defects, but again, they cannot rule out having a baby with either of these conditions. Screening using the serum integrated test does not detect other chromosomal abnormalities or birth defects.

Does a Positive Serum Integrated Screening Test Mean My Baby Has a Birth Defect?

No. Screening tests cannot indicate for certain whether your baby has a birth defect. A positive test result can only tell you that your baby is at increased risk of having Down syndrome, and the measurements are also used to identify pregnancies at high risk for trisomy 18 or an open neural tube defect if the AFP level is high. Typically, a woman who has a positive screening result is offered diagnostic tests to determine if the baby has one of these birth defects.