Pregnancy: Should I Have Screening Tests for Birth Defects?

You may want to have a say in this decision, or you may simply want to follow your doctor's recommendation. Either way, this information will help you understand what your choices are so that you can talk to your doctor about them.

Non-Interactive Decision Point

PREGNANCY: SHOULD I HAVE SCREENING TESTS FOR BIRTH DEFECTS?

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Get the facts

Your options

Have a screening test to find out the chance that your baby has a birth defect.

Don't have a screening test. You may decide not to have any tests. Or, you may want to have a diagnostic test which shows for sure if there is a birth defect.

If you know that you want a diagnostic test, then you can skip screening tests and decide whether to have chorionic villus sampling (CVS) or to choose amniocentesis.

Key points to remember

Testing for birth defects means that you may find out that your baby has a serious problem. So it's important to think about what that would mean to you and your partner. Would the news that your baby has a birth defect change your parenting plans? If, for example, you know that you would continue your pregnancy even with a birth defect, you might decide not to have any tests for birth defects.

Screening tests can't be used to diagnose a birth defect. They only estimate the **chance** that your baby has a birth defect. If one of these tests shows a higher-than-normal chance of a birth defect, you would then decide whether to have a diagnostic test such as chorionic villus sampling (CVS) in the first trimester or amniocentesis in the second trimester—to find out for sure if there is a problem.

Screening tests correctly find most—but not all—cases of Down syndrome. But they sometimes show a chance of a problem when there isn't one (false-positive result).

If you plan to have a diagnostic test such as CVS or amniocentesis, you can skip screening tests.

Screening tests have less risk than diagnostic tests.

Screening tests for birth defects can't find every problem a baby could have.

A birth defects test can cost a lot. Check to see if your insurance plan will cover it.

FAQs

What are screening tests?

Screening tests for birth defects include blood tests and a certain type of ultrasound. Depending on the type of screening you have, the test can help your doctor estimate the chance that your baby may have Down syndrome, neural tube defects, or certain rare genetic problems. The blood tests are used to look for the amount of certain substances in your blood. The doctor uses an ultrasound to look for certain changes in your baby.

Other things are considered along with the test results to estimate the chance of a problem. For example, your doctor will look at your age, weight, and race, and how far along your pregnancy is.

If one of these tests shows a higher-than-normal chance of a birth defect, you would then decide whether to have a diagnostic test- chorionic villus sampling (CVS) or amniocentesis. These tests can show for sure if there is a problem. Diagnostic tests involve taking some of the baby's cells to look at the genes and chromosomes. But diagnostic tests have a small risk of causing a miscarriage.

The American College of Obstetricians and Gynecologists recommends that all women be offered a screening test for Down syndrome. The risk of having a baby with a genetic condition increases as a woman gets older.

If you choose to have a test for birth defects, you may want to talk with a genetic counselor. He or she can talk with you about your test options and about the reasons to have or not have tests.

Screening tests may be done in the first or second trimester of pregnancy.

First trimester screening tests

First trimester screening tests let you find out about Down syndrome early in your pregnancy—between 10 and 13 weeks. But they aren't used to look for neural tube defects. This screening combines the results of two tests:

Nuchal translucency test. This test uses ultrasound to measure the thickness of the area at the back of the baby's neck. An increase in the thickness can be an early sign of Down syndrome. The test is not available everywhere, because a doctor must have special training to do it.

First-trimester blood tests. These tests measure the amounts of two substances in your blood: beta human chorionic gonadotropin (beta-hCG) and pregnancy-associated plasma protein A (PAPP-A). High levels of beta-hCG and low levels of PAPP-A may be related to certain birth defects.

A newer screening test—called **cell free fetal DNA**—looks at fetal DNA in a pregnant woman's blood. It can be used to look for Down syndrome and trisomy 18. It also may find trisomy 13, which causes intellectual disability and heart defects, among other problems. This test is an option for women who are at high risk for having a baby with certain genetic conditions. But it can't help find other birth defects, such as neural tube defects. It's not used as a general screening test. And it's not available everywhere.

Second trimester screening tests

Second-trimester screening—done between 15 and 20 weeks of pregnancy—can be used to look for Down syndrome and neural tube defects.

Maternal serum triple screen. Sometimes called the triple test, it measures the amounts of three substances in a pregnant woman's blood:

Alpha-fetoprotein (AFP)

Human chorionic gonadotropin (hCG)

Estriol (uE3)

Quadruple (or quad) test. It combines the triple screen and a test for the hormone inhibin A, which is produced by the fetus and the placenta. The quad test is a little more accurate than the triple screen. But it might not be available everywhere.

Integrated screening test. It combines the results of the first-trimester tests with those of the triple or quad screening. You would get the results after the second-trimester test is done.

Ultrasound. Doctors use ultrasound between 18 and 20 weeks. They look at a fetus's organs and other features that may be signs of conditions such as Down syndrome, neural tube defects, or heart problems.

How well do these tests work to find birth defects?

First trimester screening

First-trimester screening (nuchal translucency combined with blood tests) correctly finds Down syndrome in 82 to 87 out of 100 fetuses that have it. This also means that these tests miss it in 13 to 18 out of 100 fetuses.[1]

Ultrasound

An ultrasound can find neural tube defects up to 99 out of 100 times.[2] It won't find these problems 1 time out of 100. But ultrasound isn't as good at finding Down syndrome or genetic diseases.

Triple or quad screening

The triple or quad screen finds 80 out of 100 fetuses with neural tube defects, such as spina bifida, and about 90 out of 100 with anencephaly.[2] The test misses 20 out of 100 fetuses with spina bifida and 10 out of 100 with anencephaly.

The quad test finds Down syndrome almost 81 out of 100 times. It doesn't find it 19 out of 100 times.[3] The quad test is more likely to find Down syndrome and may be less likely to be false-positive than the triple screen.

Integrated screening

The integrated screening test (first-trimester tests plus the quad screening in the second trimester) correctly finds Down syndrome in about 95 out of 100 fetuses who have it. This also means that the test misses Down syndrome in 5 out of 100 fetuses.[1]

What's next after you get the test results?

Normal results tell you that there is no need for more tests unless you have another concern, such as a known genetic disease in your family.

Positive results tell you that there is a higher-than-average chance of a birth defect. You will be offered a diagnostic test, such as chorionic villus sampling (CVS) or amniocentesis, to find out for sure if there is a problem.

Or you may decide not to have any more tests.

If a birth defect is found, you decide where to go from there. You may choose to learn all you can about raising a child with Down syndrome or a birth defect. Or you may decide to end the pregnancy.

What are the risks of having a screening test?

With the blood tests, there is little or no physical risk. A fetal ultrasound has no known risks.

Having tests may make you worry. There is a chance that the test could show that there's a problem when there isn't one. This is called a false-positive test result. Or the test could miss a problem. This is a false-negative test result.

A positive result (meaning there could be a problem) could lead you to have a diagnostic test, which has a small risk of causing a miscarriage.

But most women have normal test results. Even when the test result is positive, most pregnancies turn out to have no problems.

What are the risks of NOT having a screening test?

If you don't have a screening test to diagnose a birth defect, your baby could have a problem that you don't find out about until birth.

The birth could be higher-risk for the baby if your doctor is not expecting a newborn with health problems.

You could give birth in a hospital that does not have a neonatal intensive care unit (NICU) for sick newborns.

A fetus with a rare, severe birth defect sometimes dies before delivery.

You might not be emotionally ready for a sick baby or one with Down syndrome.

Why might your doctor recommend a screening test?

Your doctor might recommend a screening test if:

You have a family history of Down syndrome or birth defects.

You want a test for birth defects, but you aren't sure if you want to have a diagnostic test such as chorionic villus sampling (CVS) or amniocentesis.

You might change your birth or parenting plans if you knew your fetus had a serious problem.

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Credits and References

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