

Should I have a screening test for Down syndrome during pregnancy?

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Last literature review version 18.2: May 2010 | **This topic last updated:** February 8, 2010

INTRODUCTION — This topic provides information about prenatal screening for Down syndrome to help you decide if you want to undergo this test.

More detailed information about Down syndrome screening tests is available by subscription. (See "Overview of prenatal screening and diagnosis of Down syndrome".)

WHAT IS DOWN SYNDROME? — The first decision you need to make is whether you want to know if your developing baby has Down syndrome before birth. It may help to review some facts about the condition. More detailed information is available separately. (See "Patient information: Down syndrome".)

- Down syndrome is caused by an extra number 21 chromosome
- It occurs in about 1 in 700 births
- People with Down syndrome have mild to moderate intellectual disability (mental retardation), meaning that the person can often do things independently; however, most need supervision throughout their lives.
 - People with Down syndrome have characteristic facial features (picture 1), meaning that their facial features are similar to those of other people with Down syndrome
 - People with Down syndrome may have birth defects, such as problems with how the heart or intestines develop. Other medical problems can also develop.
 - The average lifespan for an individual with Down syndrome is about 50 to 60 years

Could my baby have Down syndrome?

- A woman of any age can have a child with Down syndrome, but the risk increases as the mother's age increases
- Down syndrome usually does not run in families, except in rare cases. You should inform your doctor or nurse if you or your partner has a family member with Down syndrome.

WHAT INFORMATION DOES A DOWN SYNDROME SCREENING TEST PROVIDE? — A screening test will tell you the chances of having a certain condition, such as a fetus with Down syndrome.

Screening tests for Down syndrome cannot tell for certain whether your fetus actually has Down syndrome; rather, they tell you whether there is a low or high risk that the fetus is affected. By comparison, a diagnostic test can tell for certain if the fetus has Down syndrome.

The advantage of Down syndrome screening tests is that they only require a blood test from the mother, and possibly an ultrasound, so there is no risk to the pregnancy. The diagnostic tests for Down syndrome require putting a needle into the uterus or placenta and removing some fluid or tissue. There is a small risk of miscarriage after a diagnostic test.

The decision to have a prenatal screening test for Down syndrome is yours and depends upon your wishes, values, and beliefs. There is no right or wrong choice; you decide what is best for you and your family.

WHO IS OFFERED A SCREENING TEST FOR DOWN SYNDROME? — The American College of Obstetricians and Gynecologists recommends that all pregnant women, regardless of age, be offered the opportunity to have a screening test for Down syndrome before 20 weeks of pregnancy. Screening tests for Down syndrome are voluntary, meaning that it is your choice whether to have or not have these tests.

DECIDING TO HAVE A SCREENING TEST FOR DOWN SYNDROME

Why should I have a screening test? — These are some of the reasons that women choose to have screening for Down syndrome:

- I want as much information as possible during pregnancy about the health of my developing fetus.
- If my fetus has Down syndrome, I want to know while I am pregnant so I can learn as much as possible about the condition before the baby is born.
- I am planning to deliver my baby in a community hospital, so if my fetus has serious birth defects associated with Down syndrome (eg, heart or intestinal abnormalities), I would rather deliver at a hospital with a special care nursery.
- I have been anxious since I learned I was pregnant and if I find out that my baby's risk of having Down syndrome is low, I believe it will help ease my anxiety.
- I want to consider all of my options. If my developing fetus has Down syndrome, I would want the option to terminate the pregnancy.
- I am not sure what I would do, or how I would feel, if my developing fetus has Down syndrome. I am going to take it one step at a time. If my screening test comes back saying I am at increased risk, I will decide at that time if I want to have more testing.

Why might I choose not to have a screening test? — These are some of the reasons that a woman might choose NOT to have screening:

- I have decided that "whatever will be, will be," and I will wait until the baby's birth to find out if the baby is healthy.
- I do not want to be faced with decisions about my unborn baby. Because of religious or personal beliefs, I would never terminate an affected pregnancy.
- Since I know I would never have a diagnostic test with even a small risk of a miscarriage, I do not want to have a screening test.
- I want to know for sure if the developing fetus has Down syndrome, so I am having a diagnostic test (eg, CVS or amniocentesis) rather than a screening test. (See "Patient information: Chorionic villus sampling" and "Patient information: Amniocentesis".)

Some common myths about screening for Down syndrome — Some of the reasons women decide whether or not to have screening are based on incorrect information, such as:

- Myth — My baby won't have Down syndrome because I am young, I exercise, and I am healthy.
- Fact — A woman of any age can have a baby with Down syndrome, regardless of her health.

- Myth — My baby won't have Down syndrome because I do not drink or smoke.
- Fact — Avoiding alcohol or tobacco during pregnancy is very important for the health of you and your baby; however, it does not affect the chance that your baby will have Down syndrome.

- Myth — My baby won't have Down syndrome because no one in my family or the father of the baby's family has Down syndrome.
- Fact — Down syndrome usually does not run in families. Your baby can be affected even if there is no one else in the family with Down syndrome. If you have a family history of Down syndrome, you should talk to your doctor, nurse, or a genetic counselor to discuss if it will increase your risk of having a baby with Down syndrome.
- Myth — I should not have screening for Down syndrome unless I know that I would terminate the pregnancy if Down syndrome were detected.
- Fact — Many people who would not terminate their pregnancy choose to have screening. These people want information about their unborn baby's health before birth to plan for delivery and newborn care.
- Myth — My friend told me that if I have a screening test, it will come back "positive" since most people who have the test end up with a "positive" result.
- Fact — Most people who have a screening test will have a "negative" result, meaning that the fetus has a low risk of having Down syndrome.

WHAT DOWN SYNDROME SCREENING TESTS ARE AVAILABLE? — There are several different screening tests available. Some important considerations include the following:

- How far along in pregnancy are you?
- What screening tests are available in your area?
- What, if any, diagnostic tests (CVS or amniocentesis) are available in your area?

There are three basic types of screening tests for Down syndrome. Some of these tests need to be done early in the pregnancy, while one is not done until 15 to 18 weeks of pregnancy (at around 4 months).

- First-trimester screening is done during the first trimester (typically at 11 to 13 weeks of pregnancy). The test involves a test of your blood and an ultrasound of the fetus.
- Second-trimester screening is done during the second trimester (typically at 15 to 18 weeks of pregnancy). The test only requires a sample of your blood.
- Integrated testing combines results from tests done during the first and second trimesters. These tests involve two samples of your blood, and some also require an ultrasound of the fetus. Results are usually available in the second trimester. In some variations of this test, called sequential screening or contingent screening, results are available earlier if your test results put you at very high or very low risk of having a fetus with Down syndrome.

Which screening test should I choose? — The "best" screening test depends upon your values and preferences. You can use the following statements to help guide your decision (table 1).

TEST RESULTS — Your risk of having a fetus with Down syndrome is based on your age and the results of the blood test and ultrasound measurement. It takes about one week to get results.

The results of your test will be given as a number. As an example, a woman with a result of 1 in 2000 would have a low risk that the fetus is affected. A woman with a result of 1 in 100 would be considered at high risk that the fetus is affected.

There is no screening result that will tell for sure if the fetus definitely does or does not have Down syndrome.

Screen positive results — If your test shows an increased risk of having a fetus with Down syndrome, you can choose to have:

- Further (diagnostic) testing, if you want to know for sure if your fetus is affected. It will take about 2 to 3 weeks to schedule, perform, and get the results back from a diagnostic test.
- No further testing during pregnancy. If needed, the infant can be tested after birth.

To help you with your decision, consider meeting with a genetic counselor. He or she can help you balance the risks and benefits of diagnostic testing. Talking with a counselor can also help you think about the issues involved in ending a pregnancy or raising a child with Down syndrome.

Two diagnostic tests are available:

- Chorionic villus sampling (CVS) — Chorionic villus sampling (CVS) is the test that would be done if you were in the first trimester of pregnancy. The test is performed between 10 and 13 weeks of pregnancy and has a small risk of miscarriage. (See "Patient information: Chorionic villus sampling".)
- Amniocentesis — Amniocentesis is the test that would be done if you were in your second trimester (after 14 weeks of pregnancy). Amniocentesis is thought to have a smaller risk of miscarriage compared with CVS. (See "Patient information: Amniocentesis".)

WHERE TO GET MORE INFORMATION — Your healthcare provider is the best source of information for questions and concerns related to your medical problem.

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