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Prenatal Screening for Down syndrome

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

What is Down syndrome?

The first decision you need to make is if you want to know, before birth, whether your developing baby has Down syndrome. It may help to review some facts about the condition.

- Down syndrome is a congenital disorder arising from a chromosome defect,causingintellectualimpairmentandphysicalabnormalities including short stature and a broad facial profile. It arises from a defect involving chromosome 21, usually an extra copy (trisomy-21).
- It occurs in about 1 in 700 births and is more common earlier in pregnancy
- 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, meaning that their facial features are like 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 baby with Down syndrome, but the chance gets higher as a woman gets older.
- Down syndrome usually does not run in families, except in rare cases. You should inform your provider if you or your partner has a family member with Down syndrome.

What information does a prenatal screening test for Down syndrome provide?

A screening test will tell you the chances of having a certain medical condition.

Screening tests for Down syndrome cannot tell for certain whether your babyhas Down syndrome; rather, they tell you whether there is a low or high risk that the

baby is affected. By comparison, a diagnostic test can tell for certain if the baby 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 (about 1/200 for chorionic villus sampling [CVS] and 1/300 to 1/600 for amniocentesis) 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. Some couples who have a positive test decide against having a diagnostic test and some decide to continue the pregnancy even when Down syndrome is diagnosed.

Who is offered a screening test for Down syndrome?
The American College of Obstetricians and Gynecologist 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 baby.
- If my baby 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 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 my options. If my developing baby 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 baby 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 women 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 consider terminating an affected pregnancy.
- Since I know I would never have a diagnostic test, even with only a small risk of a miscarriage, I do not want to have a screening test.
- I want to know for sure if the developing baby has Down syndrome, so I am having a diagnostic test (e.g., chorionic villus [CVS] or amniocentesis) rather than a screening test.

Some common myths about screening for Down syndrome:
Some of the reasons women decide whether 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 may 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 provider 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 baby 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 (chorionic villus sampling [CVS] or amniocentesis) are available in your area?

There are four 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 15-18 weeks of pregnancy (at around 4 months).

- First-trimester screening is typically done at 11-13 weeks of pregnancy. It involves a test of your blood and an ultrasound of the developing baby.
- Second-trimester screening is typically done at 15-18 weeks of pregnancy. The test only requires a sample of your blood.
- Integrated screening combines results from tests done during the first and second trimesters. These tests involve two samples of your blood, and often include an ultrasound of the developing baby. 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 you are at very high or very low risk of having a baby with Down syndrome.
- A newest screening method is the measurement of circulating cell free DNA in maternal plasma, which can be done beginning at 10 weeks of gestation. This test only requires a sample of your blood. It is currently recommended for women at high risk of having a Down syndrome pregnancy and can markedly reduce the need for invasive diagnostic testing (amniocentesis, CVS). It may not be paid for by your health insurance.

Test Results
Your risk of having a baby 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.

For most tests, the results will be given as a number. For example, a woman with a result of 1 in 2000 would have a “low” risk that the baby is affected. A woman with a result of 1 in 50 would be considered “high” risk.

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

Screen positive results
If your test shows a “high” risk of having a baby with Down syndrome, you can choose to have:

- Further (diagnostic) testing, if you want to know for sure if your baby is affected. It will take about 2 to 3 weeks to schedule, perform, and get the results.
- 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) – 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 (about 1 miscarriage for every 100 procedures).
- 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 (less than 1 miscarriage for every 200 procedures) compared with CVS.