



PRENATAL TESTS FOR DOWN SYNDROME

What Is Down Syndrome?

Down syndrome is a common birth defect that includes mental retardation and—often—heart problems. Children with Down syndrome have round faces and almond-shaped eyes that slant upward. They look more like each other than like their family members. Although the learning ability of people with Down syndrome varies, all have some degree of mental retardation.

What Causes Down Syndrome?

The egg from the mother and the sperm from the father both carry chromosomes. Chromosomes are bits of genetic material. Having too few or too many chromosomes can cause birth defects. Down syndrome happens when the egg and sperm join together, and the baby that is formed gets an extra chromosome.

What Are the Chances of Having a Baby With Down Syndrome?

The chance of having a baby with Down syndrome gets higher as women get older. If you are less than 30 years old when you have your baby, your chance is less than 1 in 1000. If you are 35 years old, your chance is a bit higher than 1 in 400.

How Can Down Syndrome Be Detected During Pregnancy?

There are two types of tests for Down syndrome. Screening tests are blood tests and/or sonograms. These tests tell you what the *chance* is that your baby has Down syndrome. The other type of test is a diagnostic test. This test will tell you *for sure* if your baby has Down syndrome.

What Do Screening Tests for Down Syndrome Tell Me?

The screening test result is a number. The number tells you the *chance* your baby will have Down syndrome. If the result says 1 in 400, this means 399 women with this result will not have a baby with Down syndrome, and 1 person will have a baby with Down syndrome. There are several different screening tests available, and each gives you slightly different information. The back side of this handout tells you about the screening tests for Down syndrome that are available.

What Diagnostic Tests Can Tell Me *For Sure* if my Baby Has Down Syndrome?

The 2 tests that can tell you *for sure* are CVS and amniocentesis. Both of these tests look at the chromosomes in a few of the baby's cells. The cells are taken with a very thin needle from your bag of water or your placenta. The needle goes through your abdomen or vagina. A small number of women (about 1 in 100 women who have a CVS test and about 1 in 200 women who have an amniocentesis) who have one of these tests will have a miscarriage.

Should I Have a Screening Test or a Diagnostic Test for Down Syndrome?

There is not a single best answer for all pregnant women. Talk with your family, friends, and health care provider to make a decision that is right for you. When considering these tests for Down syndrome, it is important to remember that they do not find all the birth defects that can occur.



Table 1. Screening Tests for Down Syndrome

	When Is the Test Done?*	What the Test Tells You
Sonogram		
Nuchal Translucency (NT test)	11–14 weeks	The NT sonogram measures the amount of fluid under the skin at the back of the baby's neck and will identify about 7 of 10 babies with Down syndrome.
Sonogram and blood test		
Combined	9–14 weeks	NT test and a blood test that measures 2 different markers in your blood and will identify about 8 to 9 of 10 babies with Down syndrome.
Fully Integrated	10–13 weeks and 15–20 weeks	NT test and 2 blood tests. The first blood test is done at 10–13 weeks, and the second is the Quadruple test done at 15–20 weeks. After all 3 tests are done, the results are combined to give you 1 result, which will identify about 9 in 10 babies with Down syndrome. This test is new and not widely available yet.
Blood test		
Serum Integrated	10–13 weeks and 15–20 weeks	Two blood tests. The first blood test is done at 10–13 weeks, and the second is the Quadruple test done at 15–20 weeks. The results are combined after both are done to give you 1 result, which identifies about 8 of 10 babies with Down syndrome. This test is new and not widely available yet.
Triple Test	15–20 weeks	The triple test measures 3 different markers in your blood and will identify about 5 to 6 of 10 babies with Down syndrome.
Quadruple Test	15–20 weeks	The quadruple test measures 4 different markers in your blood and will identify about 7 of 10 babies with Down syndrome.

*Health care providers divide pregnancy into weeks starting from the first day of your last period. In order to figure out what dates to have a test done, count the number of weeks since the first day of your last period. If you don't know when you had a period, your health care provider may want to perform a sonogram first to determine how pregnant you are.

Table 2. Diagnostic Tests for Down Syndrome

	When Is the Test Done?*	What the Test Tells You
Needle through your vagina or skin to collect fetal cells		
CVS	10–13 weeks	CVS will tell you if your baby has Down syndrome or 1 of several other birth defects. About 1 woman in 100 who have this test will have a miscarriage after the test is done.
Amniocentesis	15–20 weeks	Amniocentesis will tell you if the baby has Down syndrome or 1 of several other birth defects. This test also can help tell you if the baby has certain spinal cord problems. About 1 woman in 200 who have this test will have a miscarriage after the test is done.

*Health care providers divide pregnancy into weeks starting from the first day of your last period. In order to figure out what dates to have a test done, count the number of weeks since the first day of your last period. If you don't know when you had a period, your health care provider may want to perform a sonogram first to determine how pregnant you are.

For More Information

March of Dimes

The March of Dimes Web site has information on Down syndrome, genetic testing, and other birth defects.

<http://www.modimes.org/pnhec/4439.asp>

National Institute of Child Health and Human Development

Facts About Down Syndrome

<http://www.nichd.nih.gov/publications/pubs/downsyndrome/down.htm>

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