Prenatal Tests for Down Syndrome

What is Down syndrome?

Down syndrome is a birth defect that includes mental retardation and, sometimes heart problems. Children with Down syndrome have round faces and almond-shaped eyes that slant upward. Although the learning ability of persons with Down syndrome varies greatly, all have slower mental and physical abilities. Down syndrome cannot be cured.

What causes Down syndrome?

The egg from the mother and the sperm from the father both carry chromosomes that make you who you are. Down syndrome happens when the egg and sperm join together, and the baby gets an extra chromosome.

What is the chance 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. If you are 40 years old, your chance is 1 in 100.

How can Down syndrome be detected during pregnancy?

There are 2 types of tests for Down syndrome: screening tests and diagnostic tests. Screening tests separate those who *might* from those who *probably don't* have the condition being tested for. If screening tests are abnormal, you can have a diagnostic test to tell you *for sure* if your baby has Down syndrome.

What do screening tests for Down syndrome tell me?

The blood screening test result gives you a number that tells you the *chance* your baby will have Down syndrome. If the result says "1 in 400," this means that one woman with this result *will* have a baby with Down syndrome and 399 women with this result *will not* have a baby with Down syndrome. The next page tells you about the different screening tests for Down syndrome.

Should I have any tests for Down syndrome?

There is not one right answer for all pregnant women. These tests are offered to all pregnant women, but it is your choice whether to have any of these tests done. Before you do these tests, you need to think about what you would do with the information you learn from the tests.

- Some women would chose abortion if their baby had a genetic problem. The screening tests done earlier in pregnancy will allow you to end the pregnancy earlier if there were a serious problem with the baby.
- Some women want to be prepared emotionally if their baby has a genetic problem, so they want the information even though they would not end the pregnancy.
- Other women would accept a baby with a genetic problem and feel they will only worry if a test comes back abnormal, so the information is not needed, and they would rather not have a test done.

Screening Tests for Down Syndrome

Name of the test and what it is	When in pregnancy the test is done	What the test tells you	What happens if the test is abnormal
First-trimester screen — blood test and nuchal translucency (NT) ultrasound[a]	11-14 weeks	Detects 8-9 of 10 babies with Down syndrome.	CVS or amniocentesis is offered.
Quad Screen — a single blood test	15-20 weeks	Detects 8 of 10 babies with Down syndrome.	Amniocentesis is offered.

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Name of the test and what it is	When in pregnancy the test is done	What the test tells you	What happens if the test is abnormal
Serum Integrated Screen — 2 blood tests done at different times during pregnancy	Blood test at 10-13 weeks and blood test at 15-20 weeks	Detects 8 or 9 of 10 babies with Down syndrome.	Amniocentesis is offered.
Integrated Screen — the same 2 blood tests as serum integrated screen *and* NT ultrasound	Blood test at 10-13 weeks; nucal translucency ultrasound at 11-14 weeks; and blood test at 15-20 weeks	Detects 9 of 10 babies with Down syndrome.	Amniocentesis is offered.

[a]NT is an ultrasound test that measures the area at the back of the baby's neck to see if there is more than the usual amount of fluid in this space. Extra fluid at the back of the baby's neck increases the chance that the baby has Down syndrome.

Diagnostic Tests for Down Syndrome

Name of the test and what it is	When it's done	What the test will tell you and risks of the test	What happens if the test is abnormal
CVS — needle through the lower abdomen or catheter (thin tube) through the vagina to collect cells from the placenta (afterbirth)	10-13 weeks	Diagnoses Down syndrome and some other genetic problems. One in 300 women may experience a miscarriage. Other rare risks are infection and vaginal bleeding.	You can decide to: end the pregnancy or stay pregnant and prepare for special care after the baby's birth. Depending on where you live, you may want to decide to have the baby in a hospital that has a special care unit in case the baby has problems after birth.
Amniocentesis — needle through the abdomen to collect cells from the fluid around the baby	15-20 weeks	Diagnoses Down syndrome and some other genetic problems. One in 300-500 women may experience a miscarriage. Other rare risks are infection or leaking fluid.	

For More Information

National Down Syndrome Society
Facts about Down Syndrome
http://www.ndss.org/Down-Syndrome/What-Is-Down-Syndrome/

Centers for Disease Control and Prevention
More detailed information about birth defects
http://www.cdc.gov/ncbddd/birthdefects/index.html

Mayo Clinic
More information about testing for Down Syndrome in pregnancy.
http://www.mayoclinic.com/health/down-syndrome/DS00182/DSECTION = tests-and-diagnosis