Things to Know About Prenatal Testing for Birth Defects

Most babies are born healthy, but sometimes babies have birth defects. A birth defect is a problem that happens while a baby is developing in the mother's body. Birth defects can be minor or severe. They may affect appearance, organ function, and physical and mental development.

We don't always know why your baby's has a birth defect. Sometimes we know the reason. Your baby may have a problem with their genes, may have been exposed to chemicals or viruses while in the womb, or a combination.

There are different tests we can do during your pregnancy to learn more about the health of your baby. **It is your choice to have the testing done.**

Read more about prenatal testing, what to think about before the tests, types of testing, who should get the tests, when to get the test, and what they tell you.

What tests check for birth defects?

Your healthcare provider can use screening and diagnostic tests to look for certain birth defects. Every pregnant woman will be offered screening tests. Some women may be offered diagnostic tests because of their specific risk factors.

Should I get the testing done?

We recommend for you to think about what you might do with the test results before you have the test done. **It is your choice to get any of the tests done.**

Questions to Ask Before the Tests

- What information will the test give me?
- How correct is this test?
- What are the risks for me and my baby if I have the test?
- What will I do with the information from the test?
- Will I do anything different if the test results are unusual (abnormal)?
- Will I get more tests to find out if something is really wrong?

What Might I Do with the Results?

Some women may choose to end their pregnancy if their baby has a birth defect or a serious problem. Most screening tests are done early in your pregnancy. This gives you time to make the decision that is right for you.

Some women want to be ready emotionally if their baby will have a birth defect. They want to know the test results even though they would not end their pregnancy.

Some women may not want to have the tests done. They will only worry about their baby if a test comes back unusual. They are ready to care for their baby even with a birth defect.

Screening Tests

Screening tests can help your healthcare provider know if your baby is healthy or might have certain birth defects. There are **no** physical risks to you or your baby from a screening test.

Ultrasound

This medical test looks at your baby inside your uterus (womb). Your healthcare provider will use sound waves to make an image of the baby on a monitor. An ultrasound gives you information about the health of a baby and your pregnancy. This test can show how your baby's heart, brain, and other organs are forming and any problems. You can learn fun information like the sex of your baby.

- An ultrasound can be done around 20 weeks of pregnancy.
- Ultrasounds are very safe but sometimes this test can miss problems with your baby. There should be a medical reason to have one done.

Serum Screens

This blood test will tell your healthcare provider if your baby has a higher chance to be born with a problem in their backbone, brain, or have Down syndrome. There are several different kinds of serum screens and the results can vary. The test results only help us know what women should have diagnostic tests to find out if something is wrong. **It is important to discuss the results with your healthcare provider.**

- These blood tests can be done between 11 and 22 weeks of pregnancy.

Carrier Testing for Genetic Diseases

These are blood tests to look for genetic diseases if both parents carry a gene for the same disease. These tests check the mother to see if you are a carrier of certain genetic conditions like cystic fibrosis.

- This blood test can be done anytime during pregnancy.
- **If you have the gene:** Your partner can be tested to see if your baby has a chance of having a problem from a gene. Both parents must have the gene for your baby to be at risk.

Talk to your healthcare provider about meeting with a genetic counselor. You can learn more if you think you might want this testing.

What is Cystic fibrosis?

Cystic fibrosis is a genetic disease that can cause problems in your baby's lungs and stomach.

Cell-free DNA Screening

This is a new blood test and not recommended for everyone. This test is for women who have risk factors for Down syndrome or other genetic conditions.

- This test can be done at 10 weeks of pregnancy or later.

The risk factors include:

- A mother's age is over 35 years
- A regular blood (serum) screening test that shows you are high risk
- An ultrasound that shows signs of chromosome abnormality
- Having a baby with a genetic birth defect

Talk to your healthcare provider about meeting with a genetic counselor. You can learn more if you think you might want this testing.

What is Down Syndrome?

Down syndrome is a birth defect where your child has mental and developmental delays. Sometimes your child will have heart problems.

There is no cure for Down syndrome.

Children with Down syndrome have:

- Round faces
- Almond-shaped eyes

Children with Down syndrome have different levels of learning abilities – all have slower mental and physical abilities.

Diagnostic Tests

These tests can correctly tell you if your baby has certain birth defects. We may offer these tests to pregnant women who:

- Are 35 years and older
- Have a family history of certain birth defects
- Have abnormal screening tests or ultrasounds

Chorionic Villus

This test uses a very small piece of your placenta (afterbirth). Your healthcare provider will put a tiny tube into your uterus through your vagina. They may use a needle to collect the small piece from your uterus through the skin on lower belly.

- This test can be done between 10 and 12 weeks of pregnancy.
- This test has a small risk of miscarriage or an infection.

Amniocentesis

This test uses some of the amniotic fluid that is around the baby in your uterus. Your healthcare provider will take the fluid from the uterus with a needle. This needle is put through the skin in your lower belly into your uterus. This test will let you know if your baby has Down syndrome or other genetic problems. Inherited problems are passed from the parents to the baby.

- This test can be done between 15 and 24 weeks of pregnancy.
- This test has a small risk of miscarriage or an infection.

Find Out More

Mayo Clinic: Explains about the types of prenatal genetic tests and lists questions to think about to help guide your decisions for the tests. http://www.mayoclinic.com/health/prenatal-genetic-screening/MY01966

March of Dimes: Talk about the tests, risks, when to get them, and what the results mean. http://www.marchofdimes.com/pnhec/159_519.asp