Making Decisions about Prenatal Tests for Birth Defects

What are the kinds of prenatal tests for birth defects?

There are 2 kinds of prenatal tests for birth defects: screening tests and diagnostic tests. All women will be offered screening tests. Some women may be offered diagnostic tests.

What are screening tests for birth defects?

Screening tests separate those pregnant women whose baby *might* have certain conditions from those who *probably don't* have the birth defect being tested for. There are no physical risks to you or your baby from having any of the screening tests.

Serum screens are blood tests. These tests tell you if there is a higher chance your baby has a defect in the spine or brain, or Down syndrome. A high or low result on this test does not mean your baby has a problem for sure. These results only identify which women should have diagnostic tests to find out if something is wrong. There are several different kinds of serum screens. Depending on the test, they are done between 11 and 20 weeks of pregnancy.

The cystic fibrosis blood test tells you if you have a gene for cystic fibrosis that can be passed on to your baby. Cystic fibrosis is a disease that can affect the lungs and stomach. 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 cystic fibrosis.

Ultrasound is a way to look at your baby inside your uterus (womb) using sound waves that make an image of the baby on a monitor. Ultrasound can pick up certain problems depending on when in pregnancy it is done. An ultrasound done at 16 to 20 weeks of pregnancy shows your baby's heart, brain, and other organs. Most women are offered an ultrasound at this time in their pregnancies. Sometimes ultrasound can miss problems.

What are the diagnostic tests for birth defects?

Pregnant women aged 35 and older and women with a family history of certain birth defects may also be offered diagnostic tests. These tests can tell you for sure if your baby has certain birth defects.

Chorionic villus sampling is a test done on a very small piece of your placenta (afterbirth). The test is done by putting a tiny tube into your uterus through your vagina or by putting a needle into your uterus through the skin on your lower abdomen. It is usually done between 10 and 12 weeks of pregnancy. This test has a small risk of causing infection or miscarriage.

Amniocentesis is a test of the amniotic fluid that is around the baby in your uterus. The fluid is taken out of the uterus with a needle that is put into the uterus through the skin in your lower abdomen. It is generally done between 15 and 18 weeks of pregnancy. The tests that are done on the fluid can find Down syndrome and a few other genetic problems that are passed from the parents to the baby. This test has a small risk of causing infection or miscarriage.

How do I decide?

Some important questions to ask when making decisions about these tests are:

- What information will the test give me?
- How accurate is this test?
- What risks are there for my baby and for me if I have this test?
- What would I do with the information from the test?
- Would I do anything different if the test results are abnormal?
- Would I agree to more tests to find out if something is really wrong with my baby?

It may help you to use the decision-making process on the next page.

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YOUR DECISION-MAKING ACTION PLAN

What are you trying to decide?

Tell your health care provider that you want to share in making decisions. Ask your health care provider to clearly state the decision that needs to be made. Ask your health care provider what the options are.

I am trying to decide about:_____

What do you need to know?

If there are things you do not know about your options or the test itself, get the facts. Use your local library, the Internet, and your health care provider. Ask about the specific birth defects being tested for and what the next steps are if a test is abnormal.

I need to know:_____

What do you think?

Some information is more important than other information for you. You will decide which risk or chance is most important based on your values. Once you think you have all the pros and cons of each choice, sort them out from most to least important. Share the list with your health care provider to make sure that you have not missed anything.

Pros:_____ Cons:_____

_____ _____

Make a Decision

After you have thought it over for a while, you might want to talk with your health care provider again to see if what you know about these tests is right. Talking with your partner or other family members may help. Then, make a decision.

I have decided to:_____

Take Action

Once you have made your decision, go forward and feel confident that you have made the best decision for you and your baby. You will have to make more decisions along the way. Learn as much as you can about your choices, and make decisions based on what you believe is best for you and your baby.

For More Information

Mayo Clinic
http://www.mayoclinic.com/health/prenatal-genetic-screening/MY01966
This Web site explains some of the prenatal genetic tests and lists questions to consider that help guide your decision making.

March of Dimes
http://www.marchofdimes.com/pnhec/159_519.asp
The March of Dimes Web site describes all the prenatal tests. The timing, meaning of results, and risks of each test are listed.

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