Things to Know About Prenatal Testing for Birth Defects

Most babies are born healthy. But any woman can have a baby with a birth defect. There are different ways to find birth defects during pregnancy. Some tests will tell for sure if a baby has certain conditions. Others will not. No test can find all birth defects.

It is your choice whether or not to have any testing. When you are deciding whether or not have a test, this is a helpful question to ask: do I want to know if my baby has a birth defect before the baby is born?

Here are some facts to help you sort out the all the information about different kinds of prenatal tests

Regular Screening Tests

- Safe, but not 100% accurate
- Will not tell for sure if a baby has a birth defect, but can tell you the chances
- Most women with "low risk" test results have healthy babies
- Many women with "high risk" test results also have healthy babies
- A "high risk" test result means that more tests should be done
- Anyone who is pregnant can have one
- Some examples of these tests are contingency screens, sequential screens, quad screens, and nuchal translucency ultrasounds.

What does it test for?	How is it done?	When is it done?	Pros	Cons
• Down syndrome • Trisomy 18 • Some spine and brain defects	• Blood tests • Ultrasound	• 11 to 24 weeks • Sooner is better	• No risk to baby • Less cost than diagnostic tests	• Some false alarms • Follow-up tests offered if abnormal • Can miss some babies with these birth defects

New Screening Tests

- Safe
- Not perfect, but better than regular screening tests
- Done for women who have known "risk factors" for Down Syndrome or certain other conditions called "chromosome abnormalities" These factors are:
    - Mother over 35 years old
    - Have had a baby with chromosome abnormality
    - A regular screening test that shows "high risk"
    - An ultrasound that shows signs of chromosome abnormality

Since these tests are so new, they are not yet recommended for everyone. If you think you might want a new screening test, you should meet with a genetic counselor to learn more.

What does it test for?	How is it done?	When is it done?	Pros	Cons
• Down syndrome • Trisomy 18 • A few other chromosome abnormalities	• Blood tests	• Any time after 10 weeks	• No risk to baby • Tests for more things than regular screening • More likely to find these things if the baby has them • Fewer false alarms than regular screening • Can sometimes be done for little or no money	• Rare false alarms • Follow-up tests offered if abnormal • Not as accurate as diagnostic tests • Not all birth defects can be found • Can cost a lot

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Diagnostic Tests

- Will tell for sure if a baby has certain birth defects
- Not as safe as screening tests

If you think you might want a diagnostic test, you should meet with a genetic counselor to learn more.

What does it test for?	How is it done?	When is it done?	Pros	Cons
• Down syndrome and most other chromosome abnormalities • Certain other diseases that can be passed down from parents	• Doctor takes a small piece of the placenta (CVS) • Doctor takes some fluid from around the baby (amnio-centesis)	• 11 to 13 weeks • 15-16 weeks and later	• Definite information about Down syndrome and some other genetic conditions	• Small chance of miscarriage • Costs more than screening tests • Not all types of birth defects can be found

Ultrasound

We use ultrasound to see a baby's body, the fluid that surrounds a baby, and the placenta that connects Mom to baby. Ultrasound—

- Can give clues about abnormal chromosomes, as part of a screening test
- Can also be used as a diagnostic test
- Is the best way to find certain physical birth defects such as cleft lip or open spine (spina bifida)
- Cannot detect all birth defects, not even all serious birth defects

The best time to do ultrasound to check for birth defects and signs of abnormal chromosomes is around 20-22 weeks.

Ultrasound is very safe. But there should always be a medical reason to have it done. Fun information (like finding out the sex of a baby) can come from ultrasound, but it is a medical exam. The reason to do an ultrasound is to get information about the health of a baby and the pregnancy.

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Carrier Testing for Genetic Diseases

Some genetic diseases happen when both parents carry a gene for the same disease.

- People who are gene carriers for these types of diseases are not sick. They do not show signs of disease.
- Most people who have a baby with a condition like this did not know they were gene carriers.
- Most people who have a baby with one of these genetic diseases **have no family history** of the same thing.

Tests can be done to see if you are a carrier of certain genetic conditions. It is best if **both** parents are tested. **Both** parents must be carriers for a baby to be at risk. If you think you might want genetic disease carrier testing, you should meet with a genetic counselor to learn more.

What does it test for?	How is it done?	When is it done?	Pros	Cons
• Up to 100 genetic diseases	• Blood test • Spit (saliva test)	• Any time. In fact, it's better to have the test before you are pregnant	• Gives information for family planning • Could prevent the birth of a child with certain serious illnesses	• Not all carriers are found • Can cost a lot

Questions? Call Us!

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