

What are the tests?



Ultrasound (sonogram)

Ultrasound makes a picture of the fetus using sound waves. If a visible birth defect is present, a detailed ultrasound can often detect it. Detailed ultrasound is available at State-approved Prenatal Diagnosis Centers. The ultrasound picture also shows the age of the fetus and whether there are twins.

Amniocentesis and CVS

Amniocentesis and CVS are two different ways to examine cells from the fetus. The chromosomes (genetic material) in these cells are examined to see if the fetus has certain birth defects.

Amniocentesis is usually done between **15 and 24** weeks of pregnancy. It involves removing a small amount of the fluid which surrounds the fetus. This fluid contains the cells and chromosomes of the fetus.

CVS (chorionic villus sampling) is similar to amniocentesis but is done earlier, between **10 and 14** weeks of pregnancy. A tiny piece of the placenta is removed in order to examine the chromosomes. There is usually some minor discomfort with either test.

Is prenatal diagnosis safe?

Amniocentesis and CVS are considered safe procedures when performed by medical experts at State-approved Prenatal Diagnosis Centers. The fetus is not touched. There is a small risk of miscarriage after amniocentesis or CVS. Problems such as bleeding or infection are rare. There is no known harm from ultrasound. A genetic counselor will answer any questions you may have.

If my doctor recommends prenatal diagnosis, where do I go for the tests?

It is best to go to a State-approved Prenatal Diagnosis Center. Here, experienced specialists and counselors have the most advanced medical information and equipment. Your doctor can recommend a center in your area or you can call 866-718-7915 *toll free*.

Is there a blood test that screens for birth defects?

Yes. The Prenatal Screening Program tests the pregnant woman's blood. Screening starts at 10 weeks of pregnancy. It can be one or two blood tests. These tests give your *risk* (or chance) of certain birth defects. A "screen positive" result means that there is an increased risk for your fetus having certain birth defects. Free diagnostic tests are then offered to see if the baby *actually has* a birth defect.

These blood tests screen for spina bifida (open spine), anencephaly (absent brain), abdominal wall defects, Down syndrome, Trisomy 18 and certain other chromosome disorders. Your doctor may also recommend a special screening ultrasound called Nuchal Translucency. There is more information on the website: www.cdph.ca.gov/programs/pns.

Please remember....

Most women have normal healthy babies. However, if you answered "yes" to any of the questions listed in the beginning of this brochure, be sure to talk to your doctor or a genetic counselor.

It is your choice whether or not to have any prenatal testing.

California Department of Public Health
Genetic Disease Screening Program
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866-718-7915 *toll free*
www.cdph.ca.gov/programs/pns
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Prenatal Diagnosis of Birth Defects



Should you consider it?

Prenatal Diagnosis means testing to see if your unborn baby (fetus) has certain birth defects. If you answer “yes” to any of the following questions, talk to your doctor or nurse early in your pregnancy about whether prenatal diagnosis services are recommended for you.

| | |
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| <input type="checkbox"/> | Have you or the baby's father had a child with Down syndrome or other chromosome disorder? |
| <input type="checkbox"/> | Is anyone in your family or your partner's family mentally retarded? |
| <input type="checkbox"/> | Have you or the baby's father or a close family member had . . . |
| <input checked="" type="checkbox"/> | any health problem known to “run in the family”? |
| <input checked="" type="checkbox"/> | a birth defect such as spina bifida, a heart defect, cleft lip or cleft palate? |
| <input checked="" type="checkbox"/> | a genetic disease such as sickle cell anemia, thalassemia, Tay-Sachs, cystic fibrosis, muscular dystrophy, hemophilia or PKU? |
| <input checked="" type="checkbox"/> | a baby who died during the first year of life? |
| <input type="checkbox"/> | Are you or the baby's father carriers of an inherited trait, such as sickle cell, cystic fibrosis, Tay-Sachs, or thalassemia?* |
| <input type="checkbox"/> | Have you . . . |
| <input checked="" type="checkbox"/> | had two or more miscarriages or stillbirths? |
| <input checked="" type="checkbox"/> | been diagnosed with insulin-dependent diabetes or with seizures (epilepsy)? |
| <input type="checkbox"/> | Do you drink alcohol often or use street drugs? |
| <input type="checkbox"/> | Are you related by blood to the baby's father? (For example, are you cousins?) |
| <input type="checkbox"/> | Did you have a “screen positive” Prenatal Screening blood test in this pregnancy? |
| * Some diseases are more common in certain ethnic groups. If you or the baby's father have African, Mediterranean, Asian, Jewish, Cajun, or French Canadian ancestors, talk to your doctor about carrier testing for inherited diseases. | |

Did you answer “yes” to any of these questions?

If so, you may be at higher risk than usual for having a baby with a birth defect. Most women have normal, healthy babies. But it is important for you to talk to your doctor or nurse about whether prenatal diagnosis services are recommended for you.

What are prenatal diagnosis services?

Prenatal diagnosis begins with genetic counseling. The next step is often ultrasound. After this, amniocentesis or chorionic villus sampling (CVS) may be offered. The best place for these services is a Prenatal Diagnosis Center that has earned State approval.

These tests are all voluntary. You may accept or decline any of these services.



Genetic counseling

A genetic counselor meets with you to discuss your pregnancy and family medical history. The counselor helps you understand your risks (chances) of birth defects. You will also discuss the types of tests you may choose. Counseling helps you decide whether or not to have testing.

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