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Screening for Birth Defects

Almost all children in the United States are born healthy. Out of 100 newborns, only two or three have major birth defects. Babies with birth defects may need surgery or medical treatment. Some can be found before birth with special screening tests. Some of the most common birth defects found with testing are:

- Neural tube defect: Incomplete closure of the fetal spine that can result in spina bifida
- Abdominal wall defects: One type of defect occurs when the muscle and skin that cover the wall of the abdomen are missing and the bowel sticks out through a hole in the abdominal wall (gastroschisis). Another type is when the tissue around the umbilical cord is weak and allows organs to protrude into this area (omphalocele).
- Heart defect: The chambers or pathways through the heart are not properly developed.
- Down syndrome: Mental retardation, abnormal features of the face, and medical problems such as heart defects occur as a result of an extra chromosome 21 (trisomy 21).
- Trisomy 18: There is an extra chromosome 18, which causes severe mental retardation.

Who Should Be Tested?

Screening tests are offered to all pregnant women to assess their risk of having a baby with a birth defect or genetic disorder. If a screening test shows an increased risk of having an affected baby, further tests may be used to diagnose the problem. An abnormal screening test result, while alarming, only signals a possible problem. In most cases, the baby is healthy even if there is an abnormal screening test result. Likewise, a birth defect can occur even if the test result does not show a problem. Not all disorders can be found by testing.

The Screening Test

In the first trimester, the tests include blood tests and an ultrasound exam. This screening is done between 11 and 14 weeks of pregnancy. An ultrasound exam, called nuchal translucency screening, is used to measure the thickness at the back of the neck of the fetus. An increase in this space may be a sign of Down syndrome, trisomy 18, or other chromosomal problems. In the second trimester, between 15 and 20 weeks, a second blood test is performed and combined with the results of the first trimester test. When these tests are used, about 90–95% of the Down syndrome cases can be detected.

The Next Steps

If the results of a screening test or other factors raise concerns about your pregnancy, diagnostic tests can be done to provide more information. These tests include:

- *Detailed ultrasound exam*—A type of ultrasound exam that can help explain abnormal results and provide more detailed information about the growth and development of the fetus.
- *Amniocentesis*—A procedure in which a small amount of amniotic fluid and cells are withdrawn from the sac surrounding the fetus and tested. Your doctor can help advise you on which tests may be best for you. He or she also can explain what the results mean.

Finally

Each patient needs to decide for themselves if they want to have the screening tests performed. A common misconception is that you should only do the test if you would end the pregnancy. We do find babies every year that should not be born in Redding. This is because we do not have a pediatric neurosurgeon or heart surgeon here. When we find these babies, we set up delivery in Sacramento or San Francisco. Please be prepared with any questions you may have when you talk to the doctor so that you can make an informed decision. The doctor does not know if your insurance will pay for the testing.

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Screening for Cystic Fibrosis

Cystic fibrosis (CF) is a genetic disorder that causes problems with breathing and digestion. It is caused by an abnormal ***gene*** that is passed from parent to child. There is no cure for CF, but it can be treated. Testing can be done to see if a person carries the gene and if there is a risk of passing it on to a child.

Cause

Cystic fibrosis is a genetic disorder caused by an abnormal gene that is passed from parent to child. If both parents are carriers, each of their children has a 25% chance of having the disorder.

Risk Factors

The risk of being a CF carrier is higher in certain races and ethnic groups. It occurs more often in white people than in other racial groups. The risk also is increased in families with a history of CF.

Testing

Carrier testing can be done to find out if a person has a copy of the CF gene. The test is done on a blood sample. If testing shows that a couple is at high risk, more testing can be done during pregnancy to see whether their baby will have CF. The decision to be tested is a personal one.

Follow-up Tests

If both partners are CF carriers, further prenatal testing can be done to see if the baby has CF. Parents may want to know if the baby will have CF so that they can prepare for the care of a child with special health care needs, or they may choose to end the pregnancy.

Prenatal tests done to detect CF include amniocentesis. Amniocentesis is performed at 15–20 weeks of pregnancy. A needle is used to take a small sample of amniotic fluid from around the baby that contains some of the baby's cells for testing. In the lab, the baby's cells from the ***amniotic fluid*** are grown in a special culture. This usually takes about 10–12 days. Next, the cells are studied to detect the presence of the abnormal CF gene.

Finally

Each patient needs to decide for themselves if they want to be screened for CF. A common misconception is that you should only do the test if you would end the pregnancy. To know if a baby is going to be born with CF allows for earlier treatment to be started after the child is born
Please be prepared with any questions you may have when you talk to the doctor so that you can make an informed decision do the test or not. Also, the doctor does not know if your insurance plan will pay for the testing.