



## **Education**

### **Genetic Screening Before or During Pregnancy**

#### **What is genetic screening?**

If you are planning to have a baby, you may be concerned about illnesses in one or both sides of your family that a child might inherit. You can ask your health care provider for genetic counseling or screening. Genetic screening is a way to learn more about inherited diseases.

It is best to have genetic screening before you are pregnant. It may also be done at your first prenatal visit and later in your pregnancy.

#### **When is it used?**

Some of the reasons for a couple to have genetic screening are:

- The mother will be 35 years old or older at the time of delivery.
- You have had a child with an inherited disease or birth defect.
- Someone in the family has had mental retardation or other chromosome problems.
- You have had stillbirths or several miscarriages. Stillbirths are babies born dead after 28 weeks of pregnancy.
- Something about the pregnancy is not normal. For example, blood test results have been abnormal. Or there is too much or not enough amniotic fluid around the baby.
- Someone in your family has had thalassemia. This is a disorder that causes abnormal red blood cells. It is most common in people from Asia and Mediterranean countries (for example, Italians and Greeks).
- Someone in your family has had Tay-Sachs disease. This is a brain problem that can cause early death. It is most common in Jews who have an Eastern European Ashkenazic ancestry.
- Members of your family have had other inherited problems, such as hemophilia, cystic fibrosis, or fragile X syndrome.
- Someone in your family has had sickle cell anemia. This problem causes abnormal red blood cells. It is most common in North American blacks.
- The mother had diabetes before she became pregnant.

#### **How do I prepare for genetic screening?**

Learn the medical history of members of your family. Try to get details of any inherited diseases in your family. Ask your parents if there have been any children in their families who have been retarded or disabled, or who had other birth defects.

Be ready to give the following information about yourself:

- past miscarriages
- exposure to chemicals, radiation (including x-rays), or other environmental hazards before or during pregnancy (for example, at work or from hobbies)
- any history of drug or alcohol abuse
- prescription, nonprescription, and herbal medicines taken during pregnancy, including before you knew you were pregnant.

Thinking carefully about your beliefs and goals will help prepare you to make choices that are best for you and your family. It's very important to discuss your choices and options before you find out any test results.

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## How is genetic screening done?

Your health care provider or genetic counselor will review your family and personal medical histories. You will be asked about diseases, disorders, and birth defects in your families. Both parents may have blood tests. If you are pregnant, tests of the baby may also be done.

The following tests may be used to look for birth defects and inherited diseases when you are pregnant:

- Tests of the mother's blood, including a check of the level of alpha fetoprotein (AFP). AFP is a protein made by the baby. The AFP test is done between the 15th and 18th weeks of pregnancy. If the amount of AFP is high or low, your health care provider may do other tests. The tests look for defects in the nervous system called neural tube defects, such as spina bifida. They also look for chromosome defects, such as Down syndrome.
- Ultrasound scans to check the baby for birth defects of the brain, heart, spine, legs, arms, or other organs.
- Chorionic villus sampling to test a sample of tissue from the placenta for chromosomal problems. This test is done between the 10th weeks and 12th weeks of a pregnancy.
- Amniocentesis to test the amniotic fluid around the baby for abnormal chromosomes and other substances such as alpha fetoprotein. This test is first done between the 15th and 18th weeks of a pregnancy.
- Tests of DNA in a sample of tissue from the baby (obtained, for example, by amniocentesis) to look for problems in the genetic code.
- Percutaneous umbilical cord blood sampling, also called PUBS, umbilical vein sampling, fetal blood sampling, or cordocentesis. This procedure, similar to amniocentesis, tests a sample of the baby's blood from the vein in the umbilical cord. The blood can be tested to look for chromosome problems, infection, blood disorders, or other problems.
- Fetoscopic tissue sampling, which involves inserting a tiny scope into the amniotic sac to test a sample of the baby's skin for certain severe skin problems.

Your health care provider or counselor will discuss the screening results with you. If there is a problem, they will help you understand the problem. They will describe your choices for prevention or treatment.

For support and information about genetic screening, you can contact:

- The Genetic Alliance Phone: 800-336-4363 Web site: <http://www.geneticalliance.org>
- The March of Dimes Phone: 888-663-4637 Web site: <http://www.modimes.org>.

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