Genetic Screening Before or During Pregnancy

What is genetic screening?

If you are planning to have a baby, you may be concerned about birth defects or illnesses that your child might inherit. Genetic screening is a way to learn more about this. You can have the screening before or during pregnancy.

When is it used?

All women should be offered genetic screening during pregnancy. You should have a chance to ask questions about the different options and to get more counseling if you would like it. You can choose to decline most types of genetic screening and still receive complete pregnancy care. However, there are some specific reasons that some women decide to have genetic screening including

- The mother will be 35 years old or older at the time of delivery.
- You have already had a child with an inherited disease or birth defect.
- Someone in the family has had an intellectual disability or other chromosome problems.
- You have had stillbirths or several miscarriages. (Stillbirths are babies born dead after 20 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 a red blood cell problem such as thalassemia. Thalassemia is a disorder that causes abnormal red blood cells with anemia. 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 genetic problem that can cause early death. It is most common in certain ethnic groups, such as Ashkenazi Jews, French-Canadians, and Cajuns
- 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 were 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
- Any history of drug or alcohol abuse
- Prescription, nonprescription, and herbal medicines taken during pregnancy, including before you knew you were pregnant

How is genetic screening done?

The first step in genetic screening is simply to ask questions about you and your family's health. Your healthcare 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 you and your partner may have blood tests. Tests of the baby may also be done while you are pregnant.

Common tests used to screen for genetic conditions in pregnancy include:

- Blood tests to see if you are at risk for carrying a child with cystic fibrosis, metabolism or enzyme problems, and red blood cell problems. These tests usually look for a problem with a specific gene that might be present in either parent's blood.
- Blood tests that examine various hormones and proteins that are produced by the pregnancy. The results can identify pregnancies that have either a higher or lower chance of having a child with a genetic problem, like Down syndrome, which is a form of intellectual disability.
- Ultrasound exams in the first and second trimesters to check for birth defects, such as problems with the heart, brain, and most other organs. Ultrasound can also find genetic problems that are not found with routine blood tests

No single test or mix of tests can find all forms of birth defects or genetic problems. Most screening tests give information about the chances of a problem. They do not give a yes or no answer about whether there actually is a problem. Depending on information you give to your provider or because of screening test results, several other tests may be done. These tests are more than screening tests. They can diagnose certain conditions. These tests include:

- Chorionic villus sampling, which is a test of a sample of tissue from the placenta for chromosomal problems. This test is usually done between the 10th and 13th weeks of pregnancy.
- Amniocentesis, which is a test of the amniotic fluid around the baby. It checks for abnormal chromosomes and other substances. This test is usually done between the 15th and 18th weeks of pregnancy.
- Tests of DNA in a sample of tissue from the baby to look for problems in the genes. The sample of tissue may be obtained with amniocentesis.
- Testing of blood from the umbilical cord

Your healthcare provider or counselor will discuss the screening results with you. If there is a problem, they will help you to try to 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
  1-800-336-4363
  http://www.geneticalliance.org
- The March of Dimes
  1-888-663-4637
  http://www.marchofdimes.com/

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