Genetic Counseling & Prenatal Diagnosis

November 02, 2011
By OBGYN.net Staff [1]

Genetic counseling researches the family and medical history of potential parents using perception tests, which determine the risk of having a child with a genetic birth defect or disorder. Prenatal diagnosis tests an already conceived fetus for genetic abnormalities.

Genetic counseling researches the family and medical history of potential parents using perception tests, which determine the risk of having a child with a genetic birth defect or disorder. Prenatal diagnosis tests an already conceived fetus for genetic abnormalities.

What is it? (Overview)

Modern genetics allows us to understand how genetic diseases are inherited based on DNA, genes, and chromosomes. We can now test couples and fetuses for certain inherited disorders, as well as other chromosomal and genetic abnormalities such as neural tube defects, Down's syndrome, and cystic fibrosis. For more information on this complex subject, see heredity and disease.

Couples who are thinking about having a child may consider genetic counseling and prenatal diagnosis before conception to predict any possible abnormalities in their child. Others may use the tests after they conceive to evaluate the condition of the fetus.

People who are concerned about the following conditions and choose to undergo counseling and diagnosis before becoming pregnant, may include:

- Jews of Eastern European decent who have a high risk of having children with Tay-Sachs, a metabolic disorder that leads to death by the age of three or four.
- African-Americans who risk passing on sickle-cell anemia to their children.
- Caucasians who risk passing on cystic fibrosis to their children.
- Couples who are concerned about thalassemia, a severe form of anemia that appears in people of Asian or Mediterranean origin.
- People with family histories of inherited disorders, or those who have previously had children with genetic disorders.
  - who were exposed to toxins that could cause birth defects.
  - Women with prior medical conditions or diseases that may affect their fetus, such as .>
  - Couples who share a common ancestor.

In other cases, prenatal testing is done after the baby has already been conceived in order to discover diseases such as Down syndrome and spina bifida, as well as those conditions already discussed. Besides the couples listed above, counseling and diagnosis are also recommended for over the age of 35, and those who have received abnormal alpha-fetoprotein test results. For more information about the alpha-fetoprotein test, and when and why you might have it, see alpha-fetoprotein.

For those who may be at risk, genetic counseling and prenatal diagnosis are used to help answer some important questions, such as:

- Should we have a baby? Are the chances of having a baby with a genetic defect so high that choosing adoption or artificial insemination may be the way to start a family?
• How can we treat the fetus’ potential defects? Are there surgical techniques available or other medical interventions that may help alleviate problems?

• How do we prepare, physically and psychologically, for the possible outcome of a pregnancy? Are there special educational classes, training, or information that we need in order to raise a child with defects?

• Should we take this pregnancy to term? Are the fetus’ abnormalities so severe that choosing to have an abortion is a possibility?

It is important to remember that preconception testing can only give you the odds of having a child with a certain birth defect; a genetic counselor maps out the specific numbers. It may be determined, for example, that you have a one in four chance of having a child with a certain disorder. If you decide to conceive, the fetus can undergo prenatal testing that will reveal whether or not the baby has inherited the disorder.

How can I prepare? (Preparation)

You may wish to discuss genetic counseling with your family, as well as your health-care provider, who may refer you to a genetic counselor specially trained to understand the complex issues surrounding heredity and pregnancy. If you decide to get the blood-screening test, make sure your health-care provider talks to you beforehand and answers any of your questions.

What will happen? (Procedure)

You will be required to fill out an in-depth family history, and discuss your family's medical past with the counselor. You and your partner may also take simple blood tests, or undergo an analysis of your chromosomes known as karyotyping. All this information will be considered together to help determine what genetic patterns you may pass on to your children.

If you choose to be tested after conception, one of the following tests will be used to obtain fetal cells whose genetic and chemical makeup can be analyzed in a laboratory. You may undergo:

• Amniocentesis, in which fluid is withdrawn from the amniotic sac and analyzed. See amniocentesis for more information.

• Chorionic villus sampling (CVS), which takes a sampling of the cells of the placenta. See CVS for more information.

• Percutaneous umbilical blood sampling (PUBS), which draws fetal blood from the umbilical cord. See PUBS for more information.

What are the risks? (Complications)

The tests used for genetic counseling have virtually no risks. The tests used after conception, however, may cause infection, damage to the fetus, or miscarriage. For further information, see the articles on karyotyping, amniocentesis, CVS, and PUBS.

Frequently asked questions (FAQ)

Q: If I am in a risk group, do I have to get tested?
A: No. Whether or not to undergo genetic counseling and prenatal testing is a very personal decision. Your religious or philosophical beliefs, family circumstances, and personal desires all are a part of the decision-making process.

Q: If I discover I am a carrier of a certain disease, will I still be allowed to have a baby?
A: Absolutely. The point of genetic counseling and prenatal diagnosis is simply to provide parents with information that they need to make important decisions. A good genetic counselor will help you figure out how to use the information you discover, but will not make any decisions for you.

Glossary

Alpha-fetoprotein: A protein manufactured by the fetal yolk sac, and later by the fetus’
gastro-intestinal tract and liver, that can help detect Down's syndrome, spina bifida, and other abnormalities.
Amniocentesis: A prenatal test to detect birth defects that is performed around the 15th to 18th week of pregnancy. It involves inserting a needle through the abdomen to retrieve placental cells. Artificial insemination: Impregnating a female with male semen using methods other than intercourse.

Source URL: http://www.diagnosticimaging.com/articles/genetic-counseling-prenatal-diagnosis

Links: