

# Genetic Counseling

Genetic counseling may be recommended in many situations, for example, pregnancy in a woman over age 35, a family history of inherited disease, or diagnosis of a genetic disease in a child. Counseling is done by experts in the nature of genetic disorders, the risks of passing these disorders to future children, and caring for affected children. The information provided in genetic counseling can help you make important decisions about your child's and family's health.

## What is genetic counseling?

Many diseases are related to genetic (inherited) abnormalities. Genes, which are made up of DNA, determine all of our physical characteristics, such as height, eye color, and the shape of our noses. To some extent, genes determine our mental characteristics as well. They can also carry a wide range of inherited diseases, such as sickle cell disease, cystic fibrosis, and many others.

Sometimes a gene causing an abnormal disease is passed on from parent to child. Several members of a family may have the abnormal gene. In other cases, the abnormal gene (mutation) was not passed on from the parents but seems to be an isolated occurrence in just one family member. Other abnormalities that babies are born with result from abnormal development of the baby during pregnancy and are not genetically inherited.

Genetic counseling may be recommended in a number of situations in which genetic diseases are likely to be present. Examples include when the parents are older during pregnancy, when a routine screening test gives abnormal results, when one parent has a genetic disease that may be passed on to a child, or when a child is diagnosed with a genetic disease. Unfortunately, in some conditions, a specific diagnosis cannot be made. In this situation, different possible diagnoses and their effect on the child are discussed with the parents.

A *medical geneticist* is a doctor specializing in genetic diseases. This specialist can help to make or confirm the diagnosis in patients who appear to have a genetic disease. A *genetic counselor* is a professional who can help you to understand the risks and impact of genetic diseases on your family. The information provided by these experts can help you to understand genetic diseases, the risk of passing them on to future children, and your options for medical treatment and family planning.

## Who may need genetic counseling?

Several factors increase the risk of genetic disease. Genetic counseling may be recommended in certain situations:

- If your newborn infant has had an abnormal result on routine screening tests. These are blood tests for multiple genetic diseases that are performed in every infant.
- If your child is diagnosed with some type of congenital disease (present since birth) or birth defect.
- If you had a stillborn baby (one who died before birth) with some type of congenital abnormality.
- If the parents are older when the baby is conceived. Risk is increased when the mother is over 35 years old or the father is over 50.
- If the parents are related to each other. This increases the risk that both parents will carry an abnormal gene.
- If you or others in your family have some form of genetic disease that could be passed on to your child.
- If you were exposed during pregnancy to a drug or toxin that might lead to birth defects.
- Certain racial/ethnic backgrounds are associated with an increased risk of genetic diseases. For example, the risk of sickle cell disease is increased in some African-American families.

## What is genetic counseling like?

Genetic counseling may be performed by a genetic counselor, who is a health care professional with special training and expertise in genetic and congenital diseases. Other times, counseling is done by a medical geneticist, who is a doctor specializing in genetic diseases.

There are several steps in the genetic counseling process. The genetic counselor will:

- Ask about your family's medical history (pedigree). The questions will probably be very detailed, including health information on relatives, any stillborn children, and the ages and reasons for death of family members.
- Assemble and talk to you about all relevant medical records, for example, your child's records, if he or she has been diagnosed with a genetic disease; or the mother's prenatal, pregnancy, and delivery history.
- Discuss the results of genetic tests. Further tests may be recommended. For example, if you or your child has been diagnosed with a genetic disease; other members of your family may need testing to see if they also have the abnormal gene.
- Give information to help you make decisions regarding the health of affected children and other family members and the risk of passing the disease on to future children. The counselor doesn't tell you what you should do. Instead, his or her job is to give you accurate factual information so you can make an informed decision about what's best for your family.

- The counselor or geneticist can also provide information on available medical resources such as further tests that may be helpful, medical treatment options, or support groups for families with genetic diseases.

### What happens after genetic counseling?

Families often have to make difficult decisions after going through genetic counseling:

- Whether to terminate or continue a pregnancy. If pregnancy screening tests lead to the diagnosis of a genetic disease, some couples may consider terminating the pregnancy (having an abortion). This decision may need to be made urgently because abortions generally have to be performed early in the pregnancy. For couples who decide to continue the pregnancy, the information provided by genetic counseling can help prepare for the birth of the baby.
- Family planning issues. For many genetic diseases, the risk of inheritance is fairly predictable. The genetic counselor may be able to provide information on the likelihood that future pregnancies will be affected by a particular genetic disease.
- However, it's important to realize it's not always easy to predict the risk of passing on a genetic disease. Inheritance of genetic diseases can be complex; an exact gene abnor-

mality is known for only about 10% of genetic diseases. The genetic counselor can provide you with the most accurate information available on risks.

- Options for follow-up and support. Finding out that your family has some type of gene abnormality or genetic disease is a stressful event. The genetic counselor can help put you in touch with support groups and other valuable community resources.

### When should I call your office?

Call our office if you have any questions or concerns about genetic counseling. We can help to put you in touch with a qualified genetic counselor.

### Where can I get more information?

Here are a few good sources of general information about genetic diseases and birth defects. We may be able to recommend other resources, depending on your child's and family's situation.

- March of Dimes Birth Defects Foundation. On the Internet at [www.marchofdimes.com](http://www.marchofdimes.com) or call 1-888-MODIMES (663-4637).
- The National Center on Birth Defects and Developmental Disabilities. Information and publications are available on the Internet at [www.cdc.gov/ncbddd](http://www.cdc.gov/ncbddd).