

Genetic Disorders

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In addition, if you go to <http://www.nlm.nih.gov/medlineplus/geneticdisorders.html> you will see a comprehensive list of Internet sites on genetic disorders.

To obtain a free copy of this document, go to www.masterdocs.com.

What are genetic disorders?

A genetic disorder is a disease caused in whole or in part by a "variation" (an unusual form) or "mutation" (alteration) of a gene. Genetic disorders can be passed on to family members who inherit the genetic abnormality. A small number of rare disorders are caused by a mistake in a single gene. But most disorders involving genetic factors - such as heart disease and most cancers - arise from a complex interplay of multiple genetic changes and environmental influences.

Geneticists group genetic disorders into three categories:

- Single gene disorders caused by a mistake in a single gene. The mutation may be present on one or both chromosomes of a pair. Sickle cell disease, cystic fibrosis and Tay-Sachs disease are examples of single gene disorders.
- Chromosome disorders caused by an excess or deficiency of the genes. For example, Down syndrome is caused by an extra copy of a chromosome, but no individual gene on the chromosome is abnormal.
- Multifactorial inheritance disorders caused by a combination of small variations in genes, often in concert with environmental factors. Heart disease, most cancers and Alzheimer's disease are examples of these disorders.

How do I find more information about a specific disorder or learn whether a particular disease has a genetic component?

We now believe that all diseases have a genetic component, whether inherited or resulting from the body's response to environmental stresses like viruses or toxins. Researchers are constantly learning more about the role genes play in disease, but our knowledge is still limited.

What is genetic testing?

Genetic tests look for abnormalities in a person's genes, or the presence/absence of key proteins whose production is directed by specific genes. Abnormalities in either could

indicate an inherited disposition to a disorder. Genetic testing includes gene tests (DNA testing) and biochemical tests (protein testing).

In gene tests, DNA in cells taken from a person's blood, body fluids or tissues is examined for an abnormality that flags a disease or disorder. The abnormality can be relatively large - a piece of a chromosome, or even an entire chromosome, missing or added. Sometimes the change is very small - as little as one extra, missing or altered chemical base within the DNA strand. Genes can be amplified (too many copies), over-expressed (too active), inactivated, or lost altogether. Sometimes pieces of chromosomes become switched, transposed or discovered in an incorrect location.

Gene tests use a variety of techniques to examine a person's DNA. Some tests involve using probes - short strings of DNA - with base sequences complementary to those of the mutated gene. These probes will seek their complements within an individual's genome. If the mutated sequence is present in the patient's genome, the probe will find it and bind to it, flagging the mutation.

Another type of gene tests involves comparing the sequence of DNA bases in a patient's gene to a normal version of the gene.

Biochemical tests look for the presence or absence of key proteins which signal abnormal or malfunctioning genes.

What information can genetic testing provide?

Genetic testing can be predictive, discovering whether an individual has an inherited disposition to a certain disease, before symptoms appear. Genetic tests can also confirm a diagnosis if symptoms are present. Tests can determine whether a person is a carrier for the disease. Carriers won't get the disease, but can pass on the faulty gene to their children. Prenatal testing can help expectant parents know whether their unborn child will have a genetic disease or disorder. Newborn screening tests infants for abnormal or missing gene products.

Individuals in families at high risk for a disease live with troubling uncertainties about their own future as well as their children. A negative test - especially one that is strongly predictive - can provide an enormous sense of relief.

A positive test can also produce benefits. In the best circumstances, a positive test enables the person to take steps to reduce risk. These steps could include regular screening for the disease or lifestyle changes, such as a change in diet or regular exercise. A positive test can relieve uncertainty, and can enable people to make informed decisions about their future.

Reasons for Genetic Testing

Predictive testing identifies people who are at risk of getting a disease before any symptoms appear. Predictive tests include those that screen for some inherited predispositions to certain forms of cancer, such as colon and breast cancer. Being

predisposed does not mean that the individual will get the disease. It means the person has a certain risk of developing the disease.

Carrier testing can tell individuals if they are carriers of an inherited disorder that they may pass on to their children. A person who has only one abnormal copy of a gene for a recessive condition is known as a carrier. Carriers won't get the disease, but can pass on the defective gene to their children. Cystic fibrosis and Tay-Sachs disease are examples of disorders for which parents can be carriers.

Prenatal testing is available to people at risk for having children with a chromosomal abnormality or an inherited genetic condition. Two procedures are commonly used in prenatal testing. Amniocentesis involves analyzing a sample of amniotic fluid from the womb. CVS (chorionic villus sampling) involves taking a tiny tissue sample from outside the sac where the fetus develops. Prenatal testing is often used to look for disorders such as Down syndrome, spina bifida, cystic fibrosis, and Tay-Sachs disease.

Newborn screening, the most widespread type of genetic testing, tests infant blood samples for abnormal or missing gene products. For example, infants are commonly screened for Phenylketonuria (PKU), an enzyme deficiency that can lead to severe mental retardation if untreated.

How should I decide whether to be tested?

The decision to undergo testing is a very personal one. For many people, a pivotal consideration is whether there are preventive measures that can be taken if the test result is positive. For example, those who test positive for inherited forms of breast or colon cancer can benefit from preventive measures, screening for early detection, and early treatment.

In contrast, there are no preventive measures or cures for Huntington's disease. But a positive test for Huntington's disease might help an individual make lifestyle decisions, such as career choice, family planning or insurance coverage.

Because the decision about whether to be tested for a genetic disease is complex, most people seek guidance from a genetic counselor (see below) trained to help individuals and families weigh the scientific, emotional and ethical considerations that impact on this decision.

What are genetic counselors and what do they do?

Genetics counselors are health care professionals with specialized graduate degrees and experience in medical genetics and counseling.

Genetic counselors work as members of health care teams providing information and support to individuals or families who have genetic disorders or may be at risk for inherited conditions. Genetic counselors will help:

- Assess the risk of a genetic disorder by researching a family's history and evaluating medical records.

- Weigh the medical, social and ethical decisions surrounding genetic testing.
- Provide support and information to help a person make a decision about testing.
- Interpret the results of genetic tests and medical data.
- Provide counseling or refer individuals and families to support services.
- Serve as patient advocates.
- Explain possible treatments or preventive measures.
- Discuss reproductive options.

How do I find a genetic counselor?

Your doctor may refer you to a genetic counselor. Universities and medical centers also often have affiliated genetic counselors, or can provide referrals to a counselor or genetics clinic.

As we've learned more about genetics, counselors have grown more specialized. For example, counselors may specialize in a particular disease (such as Parkinson's disease), an age group (such as adolescents) or a type of counseling (such as prenatal).

How do I decide whether I need to see a geneticist or other specialist?

A genetics counselor may refer you to a geneticist - a medical doctor or medical researcher - who specializes in your disease or disorder. A medical geneticist has completed a fellowship or has other advanced training in medical genetics. While a genetic counselor may help you with testing decisions and support issues, a medical geneticist will make the actual diagnosis of a disease or condition. Many genetic diseases are so rare that only a geneticist can provide the most complete and current information about your condition.

Along with a medical geneticist, you may also be referred to a physician who is a specialist in the type of disorder you have. For example, if a genetic test is positive for colon cancer, you might be referred to an oncologist. For a diagnosis of Huntington's disease, you may be referred to a neurologist.

What are genetics researchers studying and how can it help people with genetic disorders?

Now that a draft of the human genome map is complete, research is focusing on the function of each gene and the role that faulty genes play in disease.

This will lead to improved diagnosis of diseases and a new approach to disease therapy. Researchers will create new classes of drugs based on gene sequencing and structure. These drugs, because they are targeted to specific sites in the body, will have fewer of the side effects common in many of today's medicines. Other medications will be customized for an individual's genetic profile.

The potential for using genes themselves to treat disease - known as gene therapy - is the most exciting application of DNA science. This rapidly developing field has great potential for treating or even curing inherited and acquired diseases. Gene therapy will use normal genes to replace or supplement a defective gene, or to bolster immunity to disease. Currently, gene therapy research is primarily concerned with establishing the safety of this approach, rather than the effectiveness of the treatment. While there are hundreds of clinical trials and studies in progress, so far no cures have resulted.

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