Contacting the Information Center

Experienced information specialists are available to answer questions by telephone and TTY, e-mail, fax, and letter.

Telephone and TTY calls are answered Monday through Friday from noon to 6 p.m. eastern time. Inquiries can also be submitted via e-mail, fax, or U.S. mail.

Phone: 888–205–2311
TTY: 888–205–3223
Fax: 240–632–9164
E-mail: GARDinfo@nih.gov
Mail: P.O. Box 8126
Gaithersburg, MD 20898–8126

The Information Center does not provide genetic counseling and does not offer diagnostic testing, referrals, medical treatment, or advice.

Materials provided are for informational and/or educational purposes only and are not intended as a substitute for professional medical care, advice, diagnosis, and/or treatment. Material does not represent an endorsement of any specific tests and products by NHGRI or ORD at NIH. We strongly recommend that you seek the advice of your health care provider with any questions regarding your medical care.

National Human Genome Research Institute

Office of Rare Diseases

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Genetic and Rare Diseases

Information Center

U.S. Department of Health and Human Services

National Institutes of Health

Genes and Genetic Diseases

Diseases caused by variations (unusual forms) or mutations (alterations) in genes are called genetic diseases. These variations or mutations (called anomalies) sometimes increase a person's chances of developing certain diseases.

Genes are pieces of DNA, part of the code that makes us who we are. Each human cell contains around 30,000 genes. Besides influencing features such as eye and hair color, genes can play a role in the development of diseases and in passing on diseases from parent to child.

There are three types of genetic diseases:

- Unifactorial, involving an anomaly in one specific gene out of thousands. These are also called single-gene disorders.
- Multifactorial, involving the interactions of numerous genes in the body. Environmental factors can also contribute to the development of multifactorial diseases.
- Chromosomal, resulting from an excess or deficiency in a package of genes called a chromosome, or in a segment of chromosome.

Rare Diseases

Rare diseases are defined as disorders or conditions affecting fewer than 200,000 individuals in the United States. Most rare disorders have a strong genetic component. Rare diseases are also called orphan diseases.

The Genetic and Rare Diseases Information Center

More than 6,000 rare diseases currently affect around 25 million Americans. A large percentage of these rare diseases are considered genetic. Because many of these diseases affect relatively few individuals, information about these conditions may be difficult to find.

The difficulty in finding reliable information when a patient or a family member has been diagnosed with a genetic or rare disease can often make the experience more traumatic and stressful.

The National Institutes of Health's (NIH's) National Human Genome Research Institute (NHGRI), and Office of Rare Diseases (ORD), launched the Genetic and Rare Diseases Information Center to provide free and immediate access to accurate, reliable information about genetic and rare diseases. The Information Center provides assistance to patients and families, health professionals, and other interested parties. It provides current, authoritative, and accurate information about specific conditions and illnesses from high-quality resources such as trusted and reliable Web sites, organizations, and institutions.

The National Human Genome Research Institute (NHGRI)

NHGRI leads the NIH's contribution to the International Human Genome Project, which has as its primary goal the sequencing of the human genome. As this project nears its successful completion, the NHGRI's mission has expanded to encompass studies aimed at understanding the structure and function of the human genome and its role in health and disease. NHGRI supports the development of resources and technology that will accelerate genome research and its application to human health. NHGRI also supports the training of investigators and the dissemination of genome information to the public and health professionals. For more information, visit the NHGRI Web site (**www.genome.gov**).

The Office of Rare Diseases (ORD)

ORD's mission is to provide information on rare diseases, diagnosis, and treatment; link investigators with research subjects and patients; identify rare diseases where research is lagging or lacking; identify rare diseases research opportunities; and support research in those areas. ORD responds to public inquiries about rare (or orphan) diseases; supports national and international scientific workshops and symposia; maintains the ORD Web site and interactive rare diseases list; maintains the Medical Genetics and Rare Disorders subfile of the Combined Health Information Database; and supports research on rare diseases at NIH and across the nation. For more information, visit the ORD Web site (**www.rarediseases.info.nih.gov**).