

NATIONAL CENTER FOR ADVANCING TRANSLATIONAL SCIENCES (NCATS), OFFICE OF RARE DISEASES RESEARCH (ORDR)

ORDR stimulates and coordinates research on rare diseases as part of the NIH National Center for Advancing Translational Sciences (NCATS). The office supports a variety of programs to serve the needs of patients who have any one of the approximately 7,000 rare diseases known today. ORDR supports scientific conferences, a database of human biospecimen repositories, and research consortia of the Rare Diseases Clinical Research Network. ORDR works with many stakeholders within the rare disease community to accelerate the development of new therapies and diagnostic tools through programs like the Global Rare Diseases Registry.

For more information, visit the ORDR Web site at <http://rarediseases.info.nih.gov/>.

NATIONAL HUMAN GENOME RESEARCH INSTITUTE (NHGRI)

NHGRI led NIH's participation in the International Human Genome Project. The main goal of that project was the sequencing of the human genome, which was completed in April 2003. Now, NHGRI's mission has expanded to include studies that investigate the structure and function of the human genome and the role of the human genome in health and disease. NHGRI develops resources and technology that will speed up genome research and its application to human health. NHGRI also trains investigators and distributes genome information to the public and health professionals.

For more information, visit the NHGRI Web site at <http://www.genome.gov/>.



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GENETIC AND RARE DISEASES INFORMATION CENTER

NATIONAL CENTER FOR
ADVANCING TRANSLATIONAL SCIENCES,
OFFICE OF RARE DISEASES RESEARCH

AND

NATIONAL HUMAN GENOME
RESEARCH INSTITUTE

OF THE

NATIONAL INSTITUTES OF HEALTH

DEPARTMENT OF HEALTH AND HUMAN SERVICES

What is the Genetic and Rare Diseases Information Center (GARD)?

GARD provides the public with access to current and reliable information about diseases that are rare or genetic or both. GARD is staffed by experienced Information Specialists who provide accurate information about rare or genetic diseases in English and Spanish.

What is a rare disease?

A rare disease (also called an orphan disease) is a disease or condition affecting fewer than 200,000 persons in the United States. Up to 25 million people in the United States have rare diseases.

What is a genetic disease?

A genetic disease is caused by a change in a person's DNA. Some genetic diseases are inherited from parents, and others are caused by new changes that occur during a person's lifetime. Most genetic diseases are considered rare because they affect fewer than 200,000 people in the United States.

Who can GARD help with information?

- ❖ People who have rare or genetic diseases.
- ❖ Parents, family members, and friends of someone with a rare or genetic disease.
- ❖ Doctors, nurses, genetic counselors, other health care providers, social workers, and teachers who work with people with rare or genetic diseases.
- ❖ Scientists who are studying rare or genetic diseases and need information for their research or for people taking part in studies.
- ❖ Community leaders who are helping people find resources about rare or genetic diseases.

- ❖ Advocacy groups who want up-to-date disease information for their Web sites.
- ❖ Members of the general public who want to learn more about a rare or genetic disease.

Where should I start my search for information on a rare or genetic disease?

Visit the GARD Web pages on the NCATS/ORDR Web site at <http://rarediseases.info.nih.gov/GARD/> to search for a disease. Each disease has its own Web page, where Information Specialists post the answers to questions GARD has received.

Check the GARD Web pages for the following and more:

- ❖ Questions answered by GARD Information Specialists.
- ❖ Links to resources where you can find more information.
- ❖ Information about genetic testing and genetic services.
- ❖ Scientific conferences that have been sponsored by NIH.
- ❖ Organizations that provide information and support.
- ❖ Information about research studies and clinical trials.
- ❖ FDA-approved medications for rare diseases.

Where does GARD get information?

Information Specialists search the following and more:

- ❖ NIH resources.
- ❖ Medical textbooks.
- ❖ Journal articles.

- ❖ Web sites.
- ❖ Advocacy groups' literature and services.
- ❖ Medical databases.

What if I can't find the information I'm looking for?

Reliable information about rare or genetic diseases is often hard to find. If you can't find the information you need on our Web site, you can call or write to GARD, and our Information Specialists will answer your questions.

How do I contact GARD?

Information Specialists are available by phone Monday through Friday, 12:00 p.m. to 6:00 p.m. Eastern time (excluding federal holidays).

- ❖ PO Box 8126
Gaithersburg, MD 20898-8126
- ❖ Toll-free: 1-888-205-2311
- ❖ International number: 1-301-251-4925
- ❖ TTY: 1-888-205-3223
- ❖ Fax: 301-251-4911
- ❖ E-mail:
GARDinfo@nih.gov
- ❖ Web site:
<http://rarediseases.info.nih.gov/GARD/>