

Rare Diseases

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How many rare diseases are there?

What causes rare diseases?

What are some examples of rare diseases?

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What is a rare disease?

A rare disease is generally considered to be a disease that affects fewer than 200,000 people in the United States at any given time. Rare diseases are also sometimes called orphan diseases.

How many rare diseases are there?

There are more than 6,800 rare diseases. Altogether, rare diseases affect an estimated 25 million to 30 million Americans

What causes rare diseases?

For many rare diseases, the exact cause remains unknown. Still, for a significant portion, the problem can be traced to changes, or mutations, in a single gene. Such diseases are referred to as rare, genetic diseases. Many of these genetic mutations can be passed on from one generation to the next, explaining why certain rare diseases run in families.

Keep in mind that genetics are just one piece of the puzzle. Environmental factors, such as diet, smoking or exposure to chemicals, also can play a role in rare diseases. Such factors may directly cause disease, or interact with genetic factors to cause or increase the severity of disease.

What are some examples of rare diseases?

Examples of rare diseases caused by single-gene mutations include cystic fibrosis, which affects the respiratory and digestive systems; muscular dystrophies, which affect the muscles; and Huntington disease, which affects the brain and nervous system.

Single genes are also responsible for some rare, inherited types of cancer. Examples of these are the *BRCA1/2* genes, which increase the risk for hereditary breast and ovarian cancers, and the *FAP* gene, which increases the risk for hereditary colon cancer.

Rare diseases related to environmental factors include uncommon types of anemia caused by vitamin-deficient diets or certain medications. A rare cancer caused by environmental factors is mesothelioma, which affects the cells lining the chest cavity. More than 90 percent of mesothelioma cases stem from exposure to asbestos, a fibrous mineral once widely used in fireproofing and insulation materials.

What is being done to develop treatments for rare diseases?

Researchers have made considerable progress in recent years in figuring out ways to diagnose, treat and even prevent a variety of rare diseases. Still, much more remains to be done because there are no treatments for the vast majority of rare diseases.

The Orphan Drug Act of 1983 provides incentives for drug companies to develop treatments for rare diseases. In the 25 years since the Act was signed into federal law, the U.S. Food and Drug Administration (FDA) has approved more than 340 treatments for rare diseases.

Recently, the National Institutes of Health (NIH) launched a new effort, called the Therapeutics for Rare and Neglected Diseases (TRND) program, to create an integrated research pipeline to jump start the development of new treatments for rare and neglected disorders. The NIH Office of Rare Diseases Research (ORDR) handles oversight and governance of TRND. The laboratory work for TRND will be performed in a facility administered by the intramural program of the National Human Genome Research Institute (NHGRI).

To learn more about TRND, go to <http://www.rarediseases.info.nih.gov/TRND>.

Where can people get more information about rare diseases?

The NIH, which is part of the U.S. Department of Health and Human Services, established ORDR to help advance research focused on rare diseases.

To provide patients and their families with timely and reliable information, ORDR and NHGRI have created the Genetic and Rare Diseases (GARD) Information Center. GARD offers a searchable, frequently updated Web site located at: <http://rarediseases.info.nih.gov/GARD>.

If users cannot find what they need using GARD's Web search feature, they can contact GARD's information specialists by e-mail at GARDinfo@nih.gov. They can also call GARD at its toll-free number: 888-205-2311 or the international access number: 1-301-519-3194. The phone lines are answered from noon to 6 p.m. Eastern, Monday through Friday.

Requests can also be sent to GARD by fax, 240-632-9164; TTY, 888-205-3223; or by mail, P.O. Box 8126, Gaithersburg, MD 20898-8126.

In addition to GARD, there are many non-profit groups that provide information and support, promote research and advocate for policy issues related to various rare diseases. Among the groups that span a broad range of disorders are the National Organization for Rare Diseases, <http://www.rarediseases.org/>, and the Genetic Alliance, <http://www.geneticalliance.org/>.

