# ASHG Rare Diseases

American Society of Human Genetics

## What is a rare disease?

Any condition affecting fewer than 200,000 people in the U.S. is considered a rare disease. The majority of rare diseases have a genetic basis, directly caused by mutations (changes) in genes or chromosomes. In some cases, these genetic mutations are passed from one generation to the next. Other times, they occur spontaneously in a person who is the first in a family to be diagnosed.

Rare diseases often have severe health impacts, such as physical and intellectual disabilities and premature death. Some examples of rare diseases are cystic fibrosis, muscular dystrophy, Huntington disease, and spina bifida.

## How many people are affected by rare diseases?

It is estimated that there are around 7,000 rare diseases, although the number could be as high as 10,000. Almost one in ten Americans has a rare disease, underscoring the fact that these diseases are individually rare, but collectively common.

## What is being done to develop treatments for rare diseases?

In recent years, researchers have learned a considerable amount about how to diagnose, treat, and even prevent a variety of rare diseases. However, much more remains to be done, as there are no treatments for the vast majority of rare diseases.

One challenge is that we still do not know the specific genetic mutations involved in many rare diseases. A more complete understanding of the genetic causes of rare diseases will be necessary to develop effective treatments. Gene therapy and gene editing technologies, such as CRISPR, are increasingly moving from research labs into clinical trials.

Technologies like these have been used to fix or replace mutated genes in a small number of diseases. They have the potential to treat a wide spectrum of rare diseases, but this is still an emerging area.

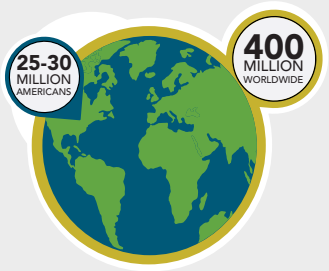
## Rare Disease Fast Facts


In the U.S., a rare disease is defined as a condition affecting fewer than 200,000 people.


There are at least 7,000 individual rare diseases.

More than 90% of rare diseases do not have FDA-approved treatments.

The majority of rare diseases begin in childhood. Approximately 30% of children with rare diseases will not live to see their 5th birthday.




Rare diseases affect 25-30 million Americans and approximately 400 million people worldwide.

For more information on rare diseases from discovery to diagnosis to treatment, please see ASHG's Success Stories in Human Genetics and Genomics Research on this topic.

6120 Executive Blvd, Suite 500, Rockville, Maryland 20852 · (301) 634-7300 · www.ashg.org

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