



RARE DISEASE DAY

FREQUENTLY ASKED QUESTIONS

What is a rare disease?

In the United States, a disease is considered rare if it is believed to affect fewer than 200,000 Americans. Some other countries and the European Union have similar but slightly different definitions. According to the National Institutes of Health (NIH), there are nearly 7,000 rare diseases affecting between 25 and 30 million Americans.

Who is affected by rare diseases?

Many—but not all—of these diseases are genetic. Some are apparent at birth. Some do not appear until much later in life. It is believed that approximately two-thirds of those living with rare diseases are children. Everyone knows someone with a rare disease. Rare diseases are an important public health concern.

How many rare diseases have treatments?

Many rare diseases still have no approved treatment. And many are not even being studied by medical researchers at this time. Often, people with rare diseases are treated “off-label” (with treatments that are not approved by FDA for their specific disease). Increasingly, patients are experiencing reimbursement problems with insurance (including Medicare and Medicaid) for off-label treatments.

What are some of the problems experienced by people who have rare diseases?

- Difficulty in obtaining an accurate diagnosis
- Limited treatment options
- Little or no research being done on the disease
- Difficulty finding physicians or treatment centers with experience in treating a particular rare disease
- Treatments that are generally more expensive than those for common diseases
- Reimbursement issues related to private insurance, Medicare, and Medicaid
- Difficulty accessing medical, social, or financial services or assistance because those making the decisions are not familiar with the disease
- Feelings of isolation and of having been abandoned or “orphaned” by our healthcare system

What is Rare Disease Day?

Rare Disease Day is an annual global event on the last day of February to focus attention on rare diseases as a public health concern. Each year, Rare Disease Day is observed on the last day of February (Feb. 29 in leap years and Feb. 28 in other years).

When was Rare Disease Day started?

Rare Disease Day was first observed in Europe in 2008. It was established by [EURORDIS](#), the European Rare Disease Organization. In 2009, EURORDIS asked [NORD](#) to be its partner in this initiative and to sponsor Rare Disease Day in the United States.

What happens on Rare Disease Day?

Patients, their families, caregivers and researchers show their support by posting stories, videos and blogs or hosting or attending events to raise awareness about challenges associated with living with a rare disease. A wide range of activities are planned and executed to celebrate the day.