



## Rare Disease Day 2016: Frequently Asked Questions

### **What is a rare disease?**

In the United States, a disease is considered rare if it affects fewer than 200,000 Americans. Some countries and the European Union have similar but slightly different definitions.

### **How many people have rare diseases?**

According to the National Institutes of Health (NIH), there are approximately 7,000 rare diseases affecting between 25 and 30 million Americans. This equates to 1 in 10 Americans, or one on every elevator and four on every bus.

### **What are some examples of rare diseases?**

Rare diseases are present across the medical spectrum. Some are widely recognized by name, such as cystic fibrosis, while others are less known, such as cat eye syndrome. Most cancers (all but a few types) are rare. There are rare neurological and neuromuscular diseases, metabolic diseases, chromosomal disorders, skin diseases, bone and skeletal disorders, and rare diseases affecting the heart, blood, lungs, kidneys, and other body organs and systems. Many rare diseases are named for the physicians who first identified them. A few are named for patients or even the hospitals where they were first identified.

### **How many rare diseases are there?**

There are more than 7,000 rare diseases, according to the National Institutes of Health (NIH).

### **Where can I find out more about rare diseases?**

The National Organization for Rare Disorders (NORD) offers a database of approximately 1,300 reports on specific rare diseases written in patient-friendly language on its website, <http://www.rarediseases.org/>. The most complete listing of rare diseases in the U.S. is on the website of the NIH Office of Rare Diseases Research (ORDR), [rarediseases.info.nih.gov](http://rarediseases.info.nih.gov).

### **Who is affected by rare diseases?**

Many—but not all—rare diseases are genetic. Some are apparent at birth while others do not appear until much later in life. Approximately two-thirds of Americans with rare diseases are children. We are all connected to someone who has a rare disease. Rare diseases are an important public health concern.

### **How many rare diseases have treatments?**

Of the 7,000 known rare diseases, approximately 95 percent have no treatment. Many rare diseases and disorders are not being studied by medical researchers. Often, patients are treated “off-label” (treatments that are not approved by the FDA for the specific disease), which can lead to insurance reimbursement problems.

## **What are some of the problems people with rare diseases experience?**

- Difficulty in obtaining an accurate diagnosis (this can take years, which can be critical for stopping or halting the progression of a disease)
- Limited treatment options
- Little or no research being done on the disease
- Difficulty finding physicians or treatment centers with experience for a particular disease
- Paying for 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 health care system

## **What is Rare Disease Day?**

Rare Disease Day is an awareness event that takes place every year on the last day of February, February 28 or February 29 in Leap Years—the rarest of calendar dates to underscore the nature of these diseases—to focus public attention on rare diseases as a public health concern.

## **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, researchers, advocates and others get involved through storytelling, media interviews, posting stories, videos and blogs online, hosting or attending events, and educational initiatives in classrooms and on college campuses. A wide range of activities are planned and executed to celebrate the day.

## **Where is Rare Disease Day Celebrated?**

Rare Disease Day is celebrated all over the world. In 2015, more than 80 countries participated. Activities are organized both virtually and in-person. To learn more about these activities, you can visit [www.rarediseaseday.us](http://www.rarediseaseday.us) for US activities or [www.rarediseaseday.org](http://www.rarediseaseday.org) for global activities.

## **What can I do to participate in Rare Disease Day?**

There are many ways to get involved in Rare Disease Day. To find the activity that is right for you, visit <http://www.rarediseaseday.us> or the global website <http://www.rarediseaseday.org>.

To get involved throughout the year, visit National Organizations for Rare Disorders (NORD)® at <http://www.rarediseases.org>.

Established in 1983, NORD is the largest rare disease organization in the U.S. and the leading independent nonprofit representing all patients and families affected by rare diseases. NORD is committed to the identification, treatment and cure of all 7,000 rare diseases that affect 30 million Americans, or 1 in every 10 people. NORD provides programs of advocacy, education, research, and patient and family services to improve the lives of all people living with rare diseases. NORD represents more than 230 disease-specific member organizations and collaborates with many other organizations in specific causes of importance to the rare disease patient community. Join NORD at [www.rarediseases.org/](http://www.rarediseases.org/) and on Twitter at [@RareDiseases](https://twitter.com/RareDiseases).

For more information, email [rdd-us@rarediseases.org](mailto:rdd-us@rarediseases.org) or call 203-744-0100.