



## RARE DISEASE DAY

### RARE DISEASE DAY 2013: 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.

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

According to the National Institutes of Health (NIH), there are over 7,000 rare diseases affecting between 25 and 30 million Americans, which is nearly 1 in 10 Americans.

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

Rare diseases are present across the spectrum of medical conditions. All but a few types of cancer are rare. There are also 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. NORD has a database of more than 1,200 reports on specific rare diseases in patient-friendly language on its website ([www.rarediseases.org](http://www.rarediseases.org)). The most complete listing of rare diseases in the U.S. is on the website of the National Institutes of Health (NIH) Office of Rare Diseases Research (ORDR) at [rarediseases.info.nih.gov](http://rarediseases.info.nih.gov).

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

Nearly one in 10 Americans have 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. More than half of the people known to be affected by rare diseases at this time are children. One of NORD's continuing themes is that everyone knows someone affected by a rare disease. And, even though each disease is rare, together they significantly impact the American public as a whole. For that reason, 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.

### **When is Rare Disease Day?**

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.

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

Rare Disease Day is celebrated all over the world, in 2012, over 60 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 [www.rarediseaseday.us](http://www.rarediseaseday.us) or the global website [www.rarediseaseday.org](http://www.rarediseaseday.org).