

Virtual Rare Disease Day at NIH



Event brings people together to raise awareness, highlight research, and promote collaboration

In the United States, diseases that each affect less than 200,000 people are called “rare diseases.” There are about 7,000 – 10,000 rare diseases, and more than half of them are diagnosed in children. Many rare diseases are caused by genetic mutations. Most of them are complicated and hard to diagnose, and only a few have any effective treatment options. Rare diseases can cause serious, chronic, and long-term health problems, and many of them are fatal.

Individually rare, collectively common

The diseases themselves are each rare, but as a group they have a large impact. Each rare disease might only affect a few hundred to several thousand people, but all together there are about 30 million people in the United States living with a rare disease.

Annual events raise awareness

Every year, the National Center for Advancing Translational Sciences (NCATS) and the NIH Clinical Center host Rare Disease Day at NIH to raise awareness about rare diseases and the people living with them.

Rare Disease Day 2022 was held on February 28th with events to support and raise awareness for rare disease patients, their families, and caregivers.

The event’s goals were to:

- **Bring together** groups of people affected by rare diseases – including patients, caregivers, patient advocates, health care providers, and researchers.
- **Increase awareness** about rare diseases and the people living with them.
- **Share information** about how the NIH advances rare disease research and highlights patients’ voices and experiences.

The virtual event was free and open to the public. It featured rare disease patient stories, scientific posters, panel discussions, and an art exhibit.

Topics included:

- Diversity in rare disease research
- Individualized therapies
- Telehealth during the COVID-19 pandemic
- The diagnostic odyssey (the long, complicated journey that many rare disease patients go through to get diagnosed)