National Center for Advancing Translational Sciences

Delivering Hope for Rare Diseases

Individually Rare, Collectively Common

More than 10,000 known rare diseases together affect millions of people. Only about 5 percent of these rare diseases have safe, effective treatments and it takes more than 6 years, on average for patients and their families to receive an accurate diagnosis. In the United States, a rare disease is defined as one that affects fewer than 200,000 people. Because the number of people with any given rare disease is small, many drug companies don't invest the millions of dollars needed to develop treatments for these chronic diseases, leading to long and expensive health care journeys.

The National Center for Advancing Translational Sciences (NCATS) is the heart of rare diseases research at the National Institutes of Health. We strive to address the significant and often unmet needs of people with rare diseases. We work with patients, advocates, clinicians and researchers to meet the public health challenge of rare diseases to improve lives today, while paving a path to the treatments of tomorrow.

A key research priority is shortening the patient's time to diagnosis. We also work on quickly testing multiple treatments at a time, which makes it easier for scientists to discover and advance rare disease therapies efficiently.

Everything we do centers on patients. They drive our mission to speed the delivery of life-changing treatments through collaboration, innovation and other translational science approaches.

NCATS Impact on Rare Diseases

TRND
49 Investigational New Drugs

RDCRN
200 diseases being studied
295 clinical sites
12 approved treatments

GARD
120,000 calls answered

8 NCATS Rare Pediatric Diseases and Orphan Drug Designations

Transforming Rare Diseases Research

NCATS partners with scientists and people across the rare diseases community to share ideas, experiences, abilities and resources. This model helps ensure that benefits and breakthroughs are shared by all.

We have several key programs that address rare diseases.

The Rare Diseases Clinical Research Network (RDCRN) brings scientists around the world together with rare diseases organizations led by patients and families. They study and develop treatments for more than 200 rare diseases.

The Therapeutics for Rare and Neglected Diseases (TRND) program moves basic research discoveries in the laboratory closer to becoming new drugs. To date, the program has advanced 15 new drug candidates to clinical testing.

The Platform Vector Gene Therapy pilot project and the Bespoke Gene Therapy Consortium aim to improve the process for developing effective gene therapies.

Providing Critical Resources

Our plan for speeding progress in rare diseases research goes beyond generating new scientific knowledge. We provide up-to-date, simple and trusted resources that educate, engage and empower rare disease patients, their families and their health care providers.

The Genetic and Rare Diseases (GARD) Information Center is a public health resource that supports people living with rare diseases and their families by providing free access to reliable, easy-to-understand information. GARD offers information and resources for thousands of genetic and rare diseases, as well as access to information specialists who can help.

The NCATS Toolkit for Patient-Focused Therapy Development is a resource for patient organizations seeking reliable and useful information about how treatments are developed.

The Rare Diseases Registry Program helps rare diseases patient organizations promote and advance patient-focused research.

Read our rare diseases research news.

Visit our Research Activities webpage.

Learn more about our Impact on Rare Diseases.