



# Delivering Hope for Rare Diseases

**Common. Costly. Actionable.** These three words may not come to mind when thinking about rare diseases. But all three describe the challenges and opportunities faced every day by those living with rare diseases.

Nearly 10% of people in this country have a rare disease — that's about 30 million people. Their estimated yearly medical care costs total about \$400 billion, which is on par with common diseases like cancer, heart failure and Alzheimer's disease.

Safe, effective treatments exist for only a few hundred of about 10,000 known rare diseases. 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 rare diseases.

The National Center for Advancing Translational Sciences (NCATS) is the heart of rare diseases research at the National Institutes of Health (NIH). We develop ways to get more treatments for all people more quickly. We work with patients, advocates, scientists and health care providers to improve lives today, while also paving a path to the treatments of tomorrow.

Our research priorities include the following:

- Shortening the time to diagnosis by finding early indicators of rare diseases.
- Tackling many diseases at a time by focusing on what they have in common. If several diseases have the same cause, they could be treated the same way.
- Making it easier and more efficient for scientists to discover and develop drugs for rare diseases.

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

## Rare Diseases by the Numbers

- ✓ **1 in 10 people** are affected by a rare disease.
- ✓ **1 in 2 people** diagnosed with a rare disease are children.
- ✓ **3–15 years** is a common timeline for diagnosis.
- ✓ **95% of rare diseases** lack an FDA-approved treatment.
- ✓ People with rare diseases face **3–5 times higher medical costs** than people with non-rare diseases.

## Transforming Rare Diseases Research

We partner with scientists and people across the rare diseases community to share ideas, experience, know-how and resources. This collaboration 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](#) 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 program](#) moves basic research discoveries in the laboratory closer to becoming new drugs. To date, the program has advanced 15 new drug candidates to human testing.
- The [Platform Vector Gene Therapy program](#) and the [Bespoke Gene Therapy Consortium](#) aim to improve the process for developing effective gene therapies.
- [Clinical Trial Readiness for Rare Diseases, Disorders and Syndromes](#) grants help researchers collect information they need to better prepare clinical trials to test treatments. Information can include how rare diseases progress and ways to measure how well treatments are working.
- [The IDeaS initiative](#) uses medical and insurance data to show the impact rare diseases have on society. It also looks for ways to help people with rare diseases get an accurate diagnosis sooner.
- NCATS' other programs, including the [Clinical and Translational Science Awards \(CTSA\) Program](#), also support rare diseases research at institutions across the country.

Our strategy for speeding progress in rare diseases research goes beyond generating new scientific knowledge. We provide up-to-date, trusted and easy-to-understand 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 aims to support people living with a rare disease and their families with public information and resources for thousands of rare diseases and offers access to information specialists.
- The [NCATS Toolkit for Patient-Focused Therapy Development](#) is an online 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.

*Banner Image: Nine-year-old Amber (center), who is part of a gene therapy trial at the NIH Clinical Center and a resident of The Children's Inn at NIH, poses with her parents, Miguel and Leticia (right); Shazia Ahmad (left); and Jennie Lucca, CEO of The Children's Inn at NIH, at Rare Disease Day at NIH on Feb. 28, 2019. (Daniel Soñé Photography)*

## Partners in Rare Diseases Research

Our scientists, grantees and collaborators are changing lives through new research. Projects include the following:

- Carrying out important laboratory studies to help [develop a genetic treatment for children with a rare condition](#) that affects the brain and central nervous system.
- Developing a [potential new therapy](#) for a rare, possibly deadly lung disease.
- Aiding the development of the [first treatment](#) to replace a faulty gene in people with a form of a rare inherited immune disease through the [Primary Immune Deficiency Treatment Consortium](#).
- [Finding a drug pair](#) that kills cancer cells and counters the effects of a mistake in a gene that causes a group of deadly childhood brain cancers.
- [Identifying three forms](#) of a rare and potentially dangerous food allergy disease. This information could help physicians better diagnose and treat the disease.