Advancing Research on Rare Diseases

The National Center for Advancing Translational Sciences (NCATS), a component of the National Institutes of Health (NIH), is committed to using research to address the extraordinary public health issue presented by rare diseases. Speeding development of treatments for patients will require innovation in science and technology and in how research is conducted.

In the United States, a rare disease is defined as a disease or condition that affects fewer than 200,000 people in this country. About 7,000 known rare diseases affect an estimated 30 million people in the United States, or nearly 10% of the population.

A Translational Approach to Rare Diseases Research

NCATS is focused on the science of translation, which is the process of turning observations into interventions to improve health. By studying translation as a science, the Center intends to improve this process and, ultimately, deliver more treatments to more people more quickly. This objective is particularly critical for rare diseases, most of which currently lack a treatment approved by the U.S. Food and Drug Administration (FDA). NCATS advances the science of translation by developing, demonstrating and disseminating innovations that reduce, remove or bypass bottlenecks in the translational research process.

NCATS brings together diverse groups of stakeholders, including patients and patient support organizations, to scientifically address important issues that slow down translation. Some areas of support include—

- analyzing health data to identify early indicators of disease to decrease the length of time to get an accurate diagnosis, which for patients with rare diseases can currently take years and involve many health care providers
- developing technologies that can lead to diagnostics and treatments for many diseases
- building and supporting a rare diseases research network to foster increased collaboration and improved data sharing among researchers
- improving the design and conduct of clinical trials that involve small populations with a rare disease

Cells from a type of childhood brain cancer are grown in the laboratory from patient cells. The cancer cells make different kinds of proteins, shown in blue, red and green. (Shawn Gillespie, Monje Lab, Stanford Medicine)
How NCATS Is Making a Difference

Working with Patients

NCATS is committed to engaging patients and their support organizations as essential partners. The Center works directly with them in a variety of ways to advance preclinical research, develop innovative clinical trials and methods, and raise awareness about the common issues that affect people with a rare disease. For example, NCATS includes patient support organizations as research partners in its clinical research network. This innovative approach helps achieve greater success in many areas, including the design and enrollment of clinical studies.

Learn more:

- The Genetic and Rare Diseases Information Center (GARD) features an online database and access to information specialists offering current, reliable and easy-to-understand information on rare and genetic diseases for patients, their families, health care providers, researchers and the public.
- Each year, as part of the global observance of Rare Disease Day®, NCATS and the NIH Clinical Center sponsor Rare Disease Day at NIH to raise awareness about rare diseases, the people they affect, and NIH research collaborations underway to address scientific challenges and to advance new treatments. This event invites the rare diseases community to visit NIH to learn about cutting-edge science and rare diseases research programs and highlights the patient’s voice in the research process.

Supporting Science

NCATS funds programs to accelerate medical research across rare diseases. The Center supports research on the commonalities and underlying molecular causes of diseases, including rare diseases. This has the potential to speed the development of treatments for multiple diseases simultaneously and ultimately help more patients more quickly.

Learn more:

- The Rare Diseases Clinical Research Network (RDCRN) brings scientists at hundreds of clinical sites around the world together with their patient support organization partners to study more than 200 rare diseases. The RDCRN teams focus on a group of diseases that share a clinical feature or have common causes.
- The Therapeutics for Rare and Neglected Diseases (TRND) program provides expertise and resources to research partners to move therapeutics for rare and neglected diseases through preclinical testing. The goal is to “de-risk” therapeutic candidates and make them more attractive for adoption by outside business partners. These partners then can invest the resources needed to complete development and conduct additional clinical trials.
- Conference grants support scientific meetings, conferences and workshops to advance translational science, including for rare diseases.

Sharing Resources

A core part of NCATS’ strategy for accelerating the pace of progress in rare diseases research is broadly and effectively disseminating knowledge and supporting awareness. It is not enough to generate information and publish research studies. For that reason, NCATS provides high-quality, widely used resources that educate, engage and empower the rare diseases community and reflect its input.

Learn more:

- The NCATS Toolkit for Patient-Focused Therapy Development is a repository of online resources for patient support organizations seeking reliable and relevant information about the therapy development process. The toolkit provides them with tools to advance their research.
- The Rare Diseases Registry Program (RaDaR) website provides rare diseases patient support organizations with instructions for setting up and maintaining high-quality patient registries. A registry is a collection of information about individuals, usually focused around a specific diagnosis or condition. The goal of RaDaR is to enable rare diseases patient support organizations to better 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 Softé Photography)