



Rare Diseases Clinical Research Network

Program Overview

There are several thousand rare diseases, of which only a few hundred have any treatments available. Combined, rare diseases affect an estimated 25 million Americans. Some obstacles to developing rare disease treatments include difficulties in patient diagnosis, geographically dispersed patients and scientific experts, a perceived high risk of development failure, and a lack of data from natural history studies, which follow a group of people with a specific medical condition over time. NCATS seeks to tackle these challenges in a systematic way by working to identify what is common among rare diseases.

One way to explore this common ground is through the collaborative [Rare Diseases Clinical Research Network](#) (RDCRN) — an NCATS-led initiative that aims to address many of the unique challenges researchers face in developing rare disease therapies. Through the network, physician scientists and their multidisciplinary teams at RDCRN consortia work together with representatives of nearly 100 patient advocacy groups to advance rare diseases clinical research and investigate new treatments for patients.

Established in 2003, the RDCRN supports [consortia of medical research centers](#) that work together to investigate groups of related rare diseases, including performing long-term natural history studies and clinical trials of new medications. The RDCRN develops robust data on more than 200 rare conditions, enabling scientists to better understand and learn from the common features among diseases.

How the RDCRN Works

Composed of about 2,600 researchers from multiple disciplines at hundreds of clinical sites around the world, the RDCRN is designed to advance medical research on rare diseases by facilitating collaboration, study enrollment and data sharing. Since the program's launch, nearly 29,000 participants have been enrolled in network clinical studies.

The RDCRN facilitates clinical research in rare diseases through support for:

- Collaborative activities, including multisite longitudinal studies of individuals with rare diseases and/or clinical trials.
- Training of clinical investigators in rare diseases research.
- Pilot and demonstration projects.
- Uniform data collection protocols.
- Access to information about rare diseases for basic and clinical researchers, academic and practicing physicians, patients, and the public.

The RDCRN currently consists of [22 clinical research consortia and a Data Management and Coordinating Center](#) (DMCC). Each consortium focuses on at least three related rare diseases, participates in multisite studies and

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actively incorporates patient advocacy groups as research partners. The DMCC enables uniform high-quality data collection and analysis and facilitates information sharing across the network. These data help scientists better understand the commonalities of rare diseases so they may apply that knowledge to improving diagnosis and treatment.

Research Collaboration

Funding and scientific oversight for the RDCRN are provided by NCATS and 10 other NIH entities: the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, the National Cancer Institute, the National Heart, Lung and Blood Institute, the National Institute of Allergy and Infectious Diseases, the National Institute of Arthritis and Musculoskeletal and Skin Diseases, the National Institute of Dental and Craniofacial Research, the National Institute of Diabetes and Digestive and Kidney Diseases, the National Institute of Mental Health, the National Institute of Neurological Disorders and Stroke, and the Office of the Director. In addition, patient advocacy organizations may contribute funding.

An RDCRN Success Story

A success from the RDCRN-supported [Urea Cycle Disorders Consortium](#) illustrates the kind of outcomes RDCRN consortia can produce. Urea cycle disorders (UCDs) are caused by genetic defects that render the body unable to remove ammonia from the blood. Ammonia is toxic and can damage organs, including the brain. Scientists have discovered a number of UCDs, all caused by defects in different genes but related by the biochemical pathway they affect and the symptoms they cause. By tackling UCDs as a group rather than individually, the UCD Consortium team catalyzed the development and approval of three drugs to treat UCDs. This advance provided patients with treatments for these life-threatening conditions.

About NCATS and Translational Science

NCATS is one of 27 Institutes and Centers at the National Institutes of Health. The Center was established to transform the translational process so that new treatments and cures for disease can be delivered to patients faster.

Translation is the process of turning observations in the laboratory, clinic and community into interventions that improve the health of individuals and the public — from diagnostics and therapeutics to medical procedures and behavioral changes.

Translational science is the field of investigation focused on understanding the scientific and operational principles underlying each step of the translational process. NCATS studies translation on a system-wide level as a scientific and operational problem.

NCATS focuses not on specific diseases, but on what is common among them. The Center serves as an adaptor to enable other parts of the research system to work more effectively.

Through its cross-cutting programs in rare diseases, translational technologies, strategic alliances and other areas, NCATS is:

- Developing new approaches, technologies, resources and models;
- Demonstrating their usefulness; and
- Disseminating the data, analysis and methodologies to the community.

For More Information

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About RDCRN

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2014 RDCRN

ncats.nih.gov/rdcn-awards2014.html