National Center for Advancing Translational Sciences

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U.S. Department of Health and Human Services
National Institutes of Health
*NIH…Turning Discovery Into Health*

# Rare Diseases Clinical Research Network

## Program Overview

An estimated 25 million Americans are affected by one of the more than 7,000 known rare diseases. Only a few hundred of these disorders have any treatments available. Some obstacles to developing rare disease treatments include difficulties in timely patient diagnosis, small numbers and geographically dispersed patients and scientific experts, a lack of data from natural history studies, and missing biomarkers to support the clinical development of new therapeutics. NCATS seeks to tackle these challenges by working to identify common problems among rare diseases and addressing them collectively, rather than individually.

One way to explore this common ground is through the collaborative Rare Diseases Clinical Research Network (RDCRN) — an NCATS-led program that partners with nine NIH Institutes and Centers. The RDCRN aims to address many of the unique challenges researchers face in developing rare disease therapies. The network consists of 20 individual consortia and a Data Management and Coordinating Center (DMCC) that work together with representatives of nearly 130 patient advocacy groups to advance rare diseases clinical research and investigate new treatments for patients.

Established in 2003, the RDCRN has supported 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. Since the program's launch, nearly 40,000 participants have been enrolled in network clinical studies. The consortia, made up of researchers from multiple disciplines at hundreds of clinical sites around the world, are intended to advance the diagnosis, management and treatment of rare diseases.

## How the RDCRN Works

The RDCRN is designed to promote highly collaborative, multisite, patient-centric translational and clinical research. The Rare Diseases Clinical Research Consortia (RDCRC) focus on unmet clinical trial readiness needs that will move the field of research forward from its current state.

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; and
- access to information about rare diseases for basic and clinical researchers, academic and practicing physicians, patients, and the public.

Each consortium focuses on at least three related rare diseases, participates in multisite studies and actively incorporates patient advocacy groups as

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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 can apply that knowledge to improving diagnosis and treatment.

## Research Collaboration

Funding and scientific oversight for the RDCRN are provided by NCATS and nine other NIH entities: the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development; 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 Dietary Supplements. In addition, patient advocacy organizations may contribute funding.

## An RDCRN Success Story

The RDCRN consortia have a rich history of accomplishment. For example, Lysosomal Disease Network scientists led crucial natural history studies and gene editing research that provided a foundation for first-in-human genome editing clinical studies for a rare metabolic disease. Primary Immune Deficiency Treatment Consortium members showed the advantage of early stem cell transplants for patients with a rare immune system disorder and severe combined immunodeficiency, and the group's work contributed to advances in gene therapy–based treatments for the disease.

## 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 systemwide 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 crosscutting 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

### Contact

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### About the RDCRN

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### RDCRN Consortia

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