

Rare Diseases Clinical Research Network



Issue

- Rare diseases, by definition, are diseases that affect fewer than 200,000 people in the United States. Yet, they present a significant health care concern since there are almost 7,000 rare diseases, affecting more than 25 million Americans and their families.
- Rare diseases may involve chronic illness, disability, and often premature death.
- Rare diseases are complex, and often with inadequate or no treatment, thereby representing a disproportionate share of health care spending.
- Patients with rare diseases are frequently misdiagnosed or are undiagnosed.
- Few drug companies conduct research into rare diseases since it is difficult to recover the costs of developing treatments for small, geographically dispersed populations.
- To advance medical research on rare diseases, a research network facilitates collaboration, enrollment in studies and trials, and sharing of data.

NIH Response

- In response to the Rare Diseases Act of 2002, the National Institutes of Health (NIH) established the Rare Diseases Clinical Research Network I (RDCRN I) to address the unique challenges of research on rare diseases.
- In 2009, the NIH funded 19 continuing and new clinical research consortia to strengthen the ongoing clinical research in rare diseases

The Rare Diseases Clinical Research Network II (RDCRN)

- Currently, the RDCRN consists of 19 Rare Diseases Clinical Research Consortia in addition to a Data Management Coordinating Center (DMCC).
- The RDCRN studies approximately 90 rare diseases at over 97 academic institutions with several hundred investigators in the United States and in other countries.
- The RDCRN enables the collaboration of scientists from multiple disciplines and provides shared access to

geographically distributed research resources and patient populations.

- Each consortium studies a group of related rare and sometimes fatal diseases.
- The DMCC develops and introduces novel technologies for data collection, information sharing, and performance of studies.
- The RDCRN leads an effort for the incorporation of standard data elements in rare diseases research with informatics support from the DMCC. These efforts allow RDCRN researchers to integrate their data with other clinical networks and scientists, enabling the development of new diagnostic tools, treatments, and preventive strategies.
- Each consortium in the RDCRN includes active participation by patient advocacy groups.
- Since its inception, investigators have made progress in every aspect of clinical research of rare diseases being studied in the network.
- Each consortium conducts longitudinal natural history studies and clinical trials of new medications, often in collaboration with industry sponsors and patient advocacy groups, and training future leaders in the field. Currently there are over 26 clinical studies actively enrolling patients.
- The RDCRN web site (<http://rarediseasesnetwork.epi.usf.edu/>) is a key source of information about rare diseases for physicians, investigators, patients, and the public. The site lists all of the studies and the participating sites.
- The RDCRN has developed a unique web-based contact registry for patients who wish to learn about their disease and participation in clinical studies (<http://rarediseasesnetwork.epi.usf.edu/registry/index.htm>). generation antiangiogenesis agents have been discovered and new and improved treatments will emerge.

The Consortia

The 19 consortia and DMCC included in the current RDCRN, as well as Principal Investigators, primary institutions and URLs for the consortia are listed below:

Urea Cycle Disorders Consortium

Batshaw, Mark L., M.D.
Children's Research Institute, Washington DC
<http://rarediseasesnetwork.epi.usf.edu/ucdc/>

Vasculitis Clinical Research Consortium

Merkel, Peter M.D., M.P.H.
Boston University School of Medicine, Boston, MA
<http://rarediseasesnetwork.epi.usf.edu/vcrc/>

Dystonia Coalition

Jinnah, H. A., M.D.
Emory University, Atlanta, GA
<http://rarediseasesnetwork.epi.usf.edu/Dystonia/>

Brain Vascular Malformation

Young, William L., M.D.
University of California San Francisco, CA
<http://rarediseasesnetwork.epi.usf.edu/BVMC/index.htm>

Genetic Disorders of Mucociliary Clearance

Knowles, Michael, M.D.
University of North Carolina at Chapel Hill, NC
<http://rarediseasesnetwork.epi.usf.edu/gdmcc/>

Chronic Graft Versus Host Disease Consortium (cGVHD)

Lee, Stephanie J., M.D., M.P.H.
Fred Hutchinson Cancer Research Center, Seattle, WA
<http://rarediseasesnetwork.epi.usf.edu/cGVHD/index.htm>

(NEPTUNE) Nephrotic Syndrome Study Network

Kretzler, Matthias, M.D.
University of Michigan, Ann Arbor, MI
<http://rarediseasesnetwork.epi.usf.edu/NEPTUNE/>

Primary Immune Deficiency Treatment

Cowan, Morton J., M.D.
University of California San Francisco, CA
<http://rarediseasesnetwork.epi.usf.edu/PIDTC/index.htm>

Lysosomal Disease Network

Whitley, Chester B., M.D.
University of Minnesota Twin Cities, MN
<http://rarediseasesnetwork.epi.usf.edu/LDN/index.htm>

Autonomic Rare Diseases Clinical Research Consortium

Robertson, David, M.D.
Vanderbilt University Medical Center, TN
<http://rarediseasesnetwork.epi.usf.edu/ARDCRC/>

Inherited Neuropathies Consortium

Shy, Michael E., M.D.
Wayne State University, MI
<http://rarediseasesnetwork.epi.usf.edu/INC/>

Rare Kidney Stone Consortium

Milliner, Dawn S., M.D.
Mayo Clinic College of Medicine, Rochester, MN
<http://rarediseasesnetwork.epi.usf.edu/RKSC/index.htm>

The Porphyria Rare Disease Clinical Research Consortium

Desnick, Robert J., Ph.D., M.D.
Mount Sinai School of Medicine, New York, NY
<http://rarediseasesnetwork.epi.usf.edu/porphyrias/index.htm>

Angelman, Rett, and Prader-Willi Syndromes Consortium

Percy, Alan, M.D.
University of Alabama at Birmingham, AL
<http://rarediseasesnetwork.epi.usf.edu/arpwsc/>

Salivary Gland Carcinomas Consortium

El-Naggar, Adel K., M.D., Ph.D.
University of Texas MD Anderson Cancer Center, Houston, TX
<http://rarediseasesnetwork.epi.usf.edu/SGCC/index.htm>

STAIR: Sterol And Isoprenoid Diseases Consortium

Steiner, Robert, M.D.
Oregon Health & Sciences University, Portland, OR

CINCH: Consortium for Clinical Investigation of Neurological Channelopathies

Griggs, Robert C., M.D.
University of Rochester, NY
<http://rarediseasesnetwork.epi.usf.edu/cinch/>

North American Mitochondrial Diseases Consortium

DiMauro, Salvatore, M.D.
Columbia University, New York, NY
<http://rarediseasesnetwork.epi.usf.edu/NAMDC/>

Clinical Research Consortium for Spinocerebellar Ataxias

Ashizawa, Tetsuo, M.D.
University of Florida, Gainesville, FL
<http://rarediseasesnetwork.epi.usf.edu/CRC-SCA/>

Data Management and Coordinating Center (DMCC) for the RDCRN

Krischer, Jeffrey, Ph.D.
University of South Florida, Tampa, FL