National Institutes of Health

Fact Sheet

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 over 6,000 rare diseases, affecting more than 25 million Americans.
- Patients with rare diseases are frequently misdiagnosed or are undiagnosed.
- Few drug companies conduct research into rare diseases since it is difficult to recoup the costs of developing treatments for such small, geographically dispersed populations.
- To advance medical research on rare diseases, a research network would facilitate 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 (RDCRN) to address the unique challenges of research on rare diseases.
- The RDCRN enables the collaboration of scientists from multiple disciplines and provides shared access to geographically distributed research resources and patient populations.
- The RDCRN consists of 10 research consortia (see listing below) and a central Data and Technology Coordinating Center (DTCC). Total NIH funding for the RDCRN, through five-year grants, is $71 million.*
- The 10 consortia are conducting more than 20 studies at greater than 50 sites in the United States and other countries. Each consortium studies a group of related rare, and sometimes fatal, diseases. These include rare bone marrow failure conditions, genetic steroid disorders, thrombotic diseases, lung diseases, neurologic and metabolic conditions, and urea cycle disorders.
- The DTCC develops and introduces novel technologies for data collection, information sharing, and performance of studies. The RDCRN is leading an effort for the incorporation of standard data elements in rare diseases research with informatics support from the DTCC. These efforts will 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. The RDCRN Coalition of Patient Advocacy Groups (CPAG), which represents more than 30 of these patient advocacy groups, is instrumental in outreach to the affected populations and provides a patient perspective to the development of studies.
- 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. More information is also available on the NCRR web site at http://www.ncrr.nih.gov/clinical/cr_rdcrn.asp.

Future

Through the coordinated international efforts of RDCRN

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investigators, new approaches to the diagnosis, treatment, and prevention of rare diseases will reduce the burden of these diseases.

- The collaboration of physicians, investigators and patient support groups will be critical in the dissemination and adoption of research results. By studying the genetic component of these rare diseases, better predictions of the course of the illness and more effective, personalized treatments may be developed.
- The next generation of rare disease investigators trained in the RDCRN will ensure that the needs of families and patients with rare diseases continue to be addressed.

The Consortia

Children's National Medical Center, Washington, DC — **Urea Cycle Disorders Consortium**

Baylor College of Medicine, Houston, TX — **Angelman, Rett, & Prader-Willi Syndromes Consortium**

Boston University School of Medicine, Boston, MA — **Vasculitis Clinical Research Consortium**

Cleveland Clinic Foundation, Cleveland, OH — **Bone Marrow Failure Disease Consortium**

Mount Sinai School of Medicine, New York, NY — **Rare Genetic Steroid Disorders Consortium**

Children's Hospital Medical Center, Cincinnati, OH — **Rare Lung Diseases Consortium**

University of Rochester, Rochester, NY — **Consortium for Clinical Investigations of Neurological Channelopathies (CINCH)**

University of North Carolina at Chapel Hill, NC — **Genetic Diseases of Mucociliary Clearance Consortium**

Childrens Hospital, Denver, CO — **Cholestatic Liver Disease Consortium (CLiC)**

Duke University School of Medicine, Durham, NC — **Rare Thrombotic Diseases Consortium**

Pediatrics Epidemiology Center, University of South Florida, Tampa, FL — **The Data and Technology Coordinating Center (DTCC)**