



FOCUS ON RARE DISEASES



THE OFFICE OF RARE DISEASES NEWSLETTER

May 2007

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Welcome to the first issue of the NIH Office of Rare Diseases (ORD) newsletter, *Focus on Rare Diseases*, which will be produced quarterly to keep the rare diseases community informed about rare disease activities. If you have any items of interest to add to the newsletter, please send them to ord@od.nih.gov. To contact the Genetic and Rare Diseases Information Center, please call 1-(888) 205-2311 or email: GARDinfo@nih.gov.

HIGHLIGHTS OF THE OFFICE OF RARE DISEASES RESEARCH ACTIVITIES

The Rare Diseases Clinical Research Network: NIH Office of Rare Diseases Opens Clinical Protocols, Expands Data Safety Monitoring Board and Protocol Review

Since Fiscal Year (FY) 2003 ORD, in collaboration with the NIH Institutes and Centers (ICs), has funded interdisciplinary clinical research teams through the Rare Diseases Clinical Research Network (RDCRN). The RDCRN consists of 10 clinical research consortia with more than 70 sites and a Data and Technology Coordinating Center (DTCC). The RDCRN also collaborates with 30 patient advocacy groups that represent patients and families. The vast distribution of research locations across the United States makes investigational treatments more accessible to patients with rare diseases. This network conducts research on approximately 50 rare diseases. The goals of RDCRN are to:

(1) Conduct clinical-translational research, including longitudinal studies, diagnostics, and therapeutic trials on multiple rare diseases;

- (2) Develop innovative tools to collect and manage geographically distributed clinical research data using standardized data elements;
- (3) Provide training to new clinical research investigators on rare diseases;
- (4) Improve access to information through the Web about rare diseases; and
- (5) Support demonstration projects.

In the past year, twenty-five protocols were submitted by the 10 research consortia and the DTCC and approved by the Data Safety Monitoring Board (DSMB) and the NIH. Twenty studies are now included in ClinicalTrials.gov and are actively recruiting patients.

The ORD and the following collaborating ICs will discuss and decide future plans for the RDCRN and develop a timetable for the announcement and release (re-issuance) of a

Request for Application (RFA). This will be an open competition, available to all research teams including the existing consortia. The collaborating ICs are:

- The National Center for Research Resources
- The National Institute of Child Health and Human Development
- The National Heart, Lung and Blood Institute
- The National Institute of Diabetes and Digestive and Kidney Diseases
- The National Institute of Arthritis and Musculoskeletal and Skin Diseases

- The National Institute of Neurological Disorders and Stroke.

In the Fall of FY 2007, the ORD and the collaborating ICs will host a conference focusing on the activities and research advances of the RDCRN. Expected attendees at the conference will include NIH extramural and intramural research program staff, regulatory agencies, academic research investigators, patient advocacy groups, the pharmaceutical and biotechnology industries, and foundations.

For more information please go to:
<http://rarediseasesnetwork.epi.usf.edu/>

Technologies Available for Licensing from NIH/FDA and Non-Profit Institutions on Rare Diseases and Conditions

The NIH Office of Technology Transfer (OTT) and the ORD have recently initiated a new activity that focuses on the transfer of technology for the prevention, diagnosis, or treatment of rare diseases or conditions. The OTT evaluates, protects, licenses, monitors, and manages the wide range of NIH and Food and Drug Administration (FDA) intramural discoveries, inventions, and other intellectual property.

One goal of the new activity is to execute license agreements to enable the transfer of NIH and FDA intramural inventions to the private sector for further research and development and potential commercialization that can lead to significant public health benefits. At the present time, over 500 technologies with potential application to rare diseases are available for licensing from the intramural research programs of the NIH and the FDA. For more information go to: (<http://www.ott.nih.gov/rarediseases>).

The OTT carries out its technology transfer mandate by retaining title to inventions developed in NIH and the FDA intramural laboratories and licensing these inventions to private entities to ensure their use, commercialization, and public availability. Similarly, extramural recipients of NIH funds, such as universities, are allowed to seek patent protection for inventions arising from their NIH-funded research and license the rights to private entities to promote commercialization.

We invite your institution to join with us in promoting the commercialization of technologies that have possible application to a rare disease or condition. The OTT has provided a similar focus for neglected diseases where universities have elected to include their available technologies on this website. Parties interested in licensing will be directed to the institution owning the technology. Please note that your institution can submit your rare disease technologies for

inclusion on our website by visiting the following link:
(<http://www.ott.nih.gov/rarediseases/submit>).

The OTT and the ORD remain committed to ensuring that the public has ongoing access to newer and more effective health care products and procedures. Please contact

[ORD \(ord@od.nih.gov\)](mailto:ord@od.nih.gov) or [OTT \(rarediseases@mail.nih.gov\)](mailto:rarediseases@mail.nih.gov) if you have any questions about this initiative.

To access the press release on this activity, please go to:
<http://www.nih.gov/news/pr/dec2006/ord-11.htm>

Collaboration, Education, and Test Translation Program for Rare Genetic Diseases

The Collaboration, Education and Test Translation Program (CETT) is an ORD pilot initiative designed to move new genetic tests from research laboratories to clinical settings. This goal is achieved through collaborations among clinicians, laboratories, researchers and patient advocates. By establishing relationships among these groups, the process initiated through the CETT Program can:

- (1) Provide individuals with rare diseases and their families a quality diagnostic genetic test for the rare condition;
- (2) Increase access to all testing needs to individuals with rare genetic diseases and to their families;
- (3) Provide appropriate resources and educational materials to increase the understanding of the rare disease and the clinical use and interpretation of the genetic test; and
- (4) Develop a database of information on the genetic test that could help with future interpretation of the test and clinical expression of the rare disease.

The ORD supports the CETT Program by making funding available through a facilitated and rapid application process. While not all new gene tests are ready to meet the milestones necessary for clinical translation, applicants are given guidance to help them attain CETT collaborative team status.

Since the CETT Program started in March 2006, 10 new genetic tests previously not available as a clinical service have been approved for translation and many more applications are under review and receiving consideration for implementation.

The CETT Program recently broadened the test translation program to include both molecular and biochemical testing, and is developing models for clinical test report forms, educational materials and public database capabilities. More information is available on the CETT Program website at: www.cettprogram.org, or by email at info@cettprogram.org

Inventory of Bio-Specimen Repositories

The Trans-NIH Rare Diseases Research Working Group endorsed the development by the RAND Corporation of a web-based,

publicly accessible inventory of human bio-specimens for research on rare and common diseases. The database, which should be

accessible in 2007, will identify existing repositories, and the rare disease bio-specimen collection, storage, and delivery issues that can impede research on rare diseases will be assessed. The web-based

site for data entry by biospecimen repositories is currently being pre-tested. Participants will be invited to update their data on an annual basis or when changes occur in their repositories.

Expanding Research in Systemic Amyloidosis

Systemic amyloidosis is a rare disease that affects multiple organ systems and requires research attention by multiple NIH ICs. In response to Congressional interest, the ORD supported a number of amyloidosis-related conferences with NIH ICs and held workshops on amyloidosis research. In June 2006, ORD convened the *Systemic Amyloidosis Focus Group Workshop*, a meeting of international researchers and NIH institute staff. The workshop participants identified research needs and scientific opportunities in the study of amyloidosis and

steps to increase our understanding of the various forms of systemic amyloidosis in order to improve the prevention and treatment of this devastating disease. A workshop summary report entitled, “Challenges and Opportunities for Systemic Amyloidosis Research,” will be published in 2007 in the journal, *Amyloid*, and plans are underway to issue a multi-institute program announcement to inform the research community about the interest of the NIH in supporting research relevant to this disorder.

Fiscal Year 2006 Biennial Report on Rare Diseases Research Activities

The Rare Diseases Act of 2002, P.L. 107-280, instructed the Director of the Office of Rare Diseases to prepare a biennial (every two years) report that describes rare diseases research and related activities at the NIH and identifies future plans.

The FY 2004 biennial report is available at: http://rarediseases.info.nih.gov/html/reports/fy2004/FY2004_index.html

The FY 2006 biennial report will provide an overview of ongoing research in extramural and intramural programs, recent scientific advances, new or planned research initiatives, and related activities such as scientific workshops and symposia, public and

professional education and training, and information development and dissemination. Many advances presented are the direct result of years of rare diseases research sponsored by NIH. Patients with rare diseases continue to benefit from the treatment applications realized by the emphasis NIH places on both basic and clinical intramural and extramural research programs.

The ORD has also been responsible for an annual report on rare diseases research and related activities.

The FY 2005 annual report is available at: http://rarediseases.info.nih.gov/html/reports/fy2005/Annual_Report_FY_05_Final.pdf

Genetic and Rare Diseases Information Center

The Genetic and Rare Diseases Information Center (GARD) provides individually tailored information to patients and their families, health professionals, researchers, and the public. Since 2002, GARD has

responded to more than 17,000 inquiries for more than 4,732 rare and/or genetic diseases. For more information go to: http://rarediseases.info.nih.gov/html/resources/info_cntr.html

Scientific Conferences

Every year, the ORD collaborates with NIH ICs and other Federal agencies to stimulate rare diseases research by supporting scientific conferences when research is lagging or to identify scientific opportunities. The outcomes of these scientific conferences have included:

- (1) The establishment of research priorities;
- (2) The development of collaborative research protocols;
- (3) Criteria for diagnosing and monitoring rare diseases;
- (4) New research endeavors; and
- (5) Publications in the scientific literature.

These scientific conferences have also contributed to the exchange of ideas and information among basic and clinical investigators, patient advocacy groups, NIH staff, and the pharmaceutical, biotechnology, and medical devices industry.

The ORD has attached a list of the most recent conferences supported by the ORD with this issue of the newsletter and will do so with each future issue. For additional information on the scientific conferences program, visit the ORD website at: <http://rarediseases.info.nih.gov/html/workshops/sicon.html>

Angel Flight

ORD and National Human Genome Research Institute (NHGRI) jointly invited Angel Flight to participate in the NIH Clinical Center efforts. This not-for-profit organization provides transportation free of charge to and from the Clinical Center for patients and family members. Since it began operations through the Office of the Clinical Director of the NHGRI in January 2004, Angel Flight has flown more than 400 patients and family members to and from the NIH Clinical Center. The patients, with a variety of diagnoses, have come for

education, treatment, or research purposes. Studies include alkaptonuria, autosomal recessive polycystic kidney disease (ARPKD), cystinosis, leukemia, Hermansky-Pudlak Syndrome (HPS), Pallister-Hall syndrome, periodic fever syndrome, and several types of cancers. Angel Flight also facilitates air transportation for patients who are traveling to participate in research studies for the RDCRN. For more information please visit: www.angelflightatNIH.org

About ORD

The Office of Rare Diseases (ORD) was established in 1993 within the Office of the Director of the NIH, the Nation's medical research agency. Public Law 107-280, the [Rare Diseases Act of 2002](#), established the office in statute. The goals of ORD are to stimulate and coordinate research on rare diseases and to respond to the needs of patients who have any one of the almost 7,000 rare diseases known today.

Definition of rare diseases: (Orphan Drug Act as amended in 1984 by P.L. 98-551 to add a numeric prevalence threshold to the definition of rare diseases.)

“...the term, rare disease or condition means any disease or condition which (a) affects less than 200,000 persons in the U. S. or (b) affects more than 200,000 persons in the U.S. but for which there is no reasonable expectation that the cost of developing and making available in the U. S. a drug for such disease or condition will be recovered from sales in the U. S. of such drug.”

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