



FOCUS ON RARE DISEASES



THE OFFICE OF RARE DISEASES NEWSLETTER
May 2008

*Office of Rare Diseases, National Institutes of Health
6100 Executive Boulevard, Room 3B01, MSC 7518
Bethesda, Maryland 20892-7518
Phone (301) 402-4336 Fax: (301) 480-9655
Email: ord@od.nih.gov
Web Address: <http://rarediseases.info.nih.gov/>*

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

Patient Advocacy Teleconference: NIH Undiagnosed Diseases Program

The National Institutes of Health (NIH) hosted a teleconference beginning at 9:30 a.m. Eastern Time on Monday, May 19, 2008, to introduce the NIH Undiagnosed Diseases Program, a new program to study some of the most difficult-to-diagnose medical cases. The Undiagnosed Diseases Program is an unprecedented trans-NIH clinical initiative that will engage a wide range of consulting specialists to evaluate patients who suffer from conditions that have resisted diagnosis.

A research program with hopes of discovering variations of both rare and common disorders, the Undiagnosed Diseases Program will also provide hope for many Americans who now struggle with mysterious diseases. The goal of this teleconference is to introduce the program to patient advocates invested in the plight of those with undiagnosed disorders, and to provide an initial opportunity to discuss the

vision and scope of the program. Please consider participating in this informational teleconference, specially arranged for representatives of patient advocacy organizations. We will describe the program and its anticipated service to the medical community and patients from around the United States. Call-in information is below.

Teleconference. Dial-in to: 1-800-862-9098; from outside of the U.S., call: 785-424-1051. Conference ID/Password: 7NIHUDPADVO
To assist us in making sufficient call-in lines available, please RSVP by 9 a.m. Eastern Time on Friday, May 16, 2008 to mcguirema@mail.nih.gov with the name of your organization and the name of the person who will be representing your organization on the phone call. You may also RSVP by phone by dialing 301-402-0911.

25th Anniversary of Orphan Drug Act

In the decade before the Orphan Drug Act was passed by Congress and signed by President Ronald Reagan, only 10 treatments had been developed for rare diseases by the pharmaceutical industry. In the 25 years since then, more than 1,100 treatments for rare diseases have entered the research pipeline and more than 300 have been approved by the U.S. Food and Drug Administration for marketing. In addition, the Act has proven to be a potent catalyst to the growth of the pharmaceutical and biotechnology industries in the United States.

A rare or “orphan” disease is defined by the U.S. Food and Drug Administration (FDA) as a disease or condition that affects fewer than 200,000 Americans. In the past, these diseases of low prevalence were overlooked by drug and medical device developers. The Orphan Drug Act provides financial incentives that help companies recover the cost of developing a drug for small patient populations.

During 2008, the National Organization for Rare Disorders (NORD) and its members, along with the National Institutes of Health (NIH), the U.S. Food and Drug

Administration (FDA), and the pharmaceutical and biotechnology industry organizations, will be celebrating the 25th anniversary of both the Orphan Drug Act and of the founding of NORD. Special events planned for the year include the following:

- A 25th Anniversary Gala hosted by NORD at Union Station in Washington, DC, on May 20, 2008
- An international scientific conference on rare diseases hosted by the National Institutes of Health, with assistance from NORD, during the same week as the NORD Gala
- A special 25th Anniversary NORD Annual Conference in the fall of 2008

Information about these special events, and other news related to rare diseases and the development of new treatments for them, will be posted throughout the year on NORD’s web site (www.rarediseases.org).

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For information, contact:

Mary Dunkle, National Organization for Rare Disorders
(203) 744-0100, mdunkle@rarediseases.org

Newborn Screening Saves Lives Act of 2007

Newborn Screening Saves Lives Act of 2007 - Amends the Public Health Service Act to require the Secretary of Health and Human Services, acting through the Associate Administrator of the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA), to award grants to eligible entities to: (1) provide education and training in newborn screening and congenital, genetic, and metabolic disorders to health care professionals and newborn screening laboratory personnel; (2) develop educational programs about newborn screening for parents, families, and patient advocacy and support groups; and (3) establish a system to assess and coordinate treatment relating to congenital, genetic, and metabolic disorders. Additional information is available at: <http://thomas.loc.gov/cgi-bin/bdquery/z?d110:s634>:

Rare Diseases Clinical Research Network (RDCRN) Update

The RDCRN consists of 10 consortia. Each consortium focuses on a group of between three to eight rare diseases, totaling approximately 50 rare diseases for the network. In addition, the network includes a data and technology coordinating center (DTCC) that serves the RDCRN. The collaborating patient advocacy groups, through a coordinating coalition, participate as members of the network's steering committee. The network consists of more than 70 sites and includes more than 30 patient advocacy groups. The distribution of research locations across the United States makes investigational studies and treatments more accessible to patients with rare diseases. The network through the DTCC collects clinical information to develop biomarkers and new approaches to diagnosis, treatment, and prevention of rare diseases; provides training of new clinical research investigators; and supports demonstration projects.

Updates:

- **Request for Applications (RFAs) for the RDCRN**

The NIH Office of Rare Diseases (ORD) in collaboration with ten NIH Institutes has published the following Request for Applications (RFAs) to continue the RDCRN Program.

1) Rare Diseases Clinical Research Consortia (RDCRC) for the Rare Diseases Clinical Research Network (U54)

<http://grants.nih.gov/grants/guide/rfa-files/RFA-OD-08-001.html> (RFA-

OD-08-001)

2) Data Management and Coordinating Center (DMCC) for the Rare Diseases Clinical Research Network (U54)

<http://grants.nih.gov/grants/guide/rfa-files/RFA-OD-08-002.html> (RFA-OD-08-002)

- **Pre-application meeting for RDCRN RFAs**

ORD held a public pre-application meeting on March 17, 2008. The purpose of this meeting was to provide prospective applicants with an overview presentation on the above mentioned RDCRN RFAs followed by an exchange of questions and answers pertinent to preparing applications in response to these RFAs with representatives of the Office of Rare Diseases and participating NIH Institutes and Centers (IC). This opportunity is not intended to replace more detailed discussions that applicants may wish to have with the IC program staff. The public pre-application meeting for RDCRN RFAs can be accessed at [Pre-Application Meeting for RFA-OD-08-001 and RFA-OD-08-002, Rare Diseases Clinical Research Network](http://rare diseases.info.nih.gov/Wrapper.aspx?src=asp/resources/extr_res.asp).

VideoCast proceedings of this meeting are available.

http://rare diseases.info.nih.gov/Wrapper.aspx?src=asp/resources/extr_res.asp

- **Clinical Protocols**
In past 24 months more than thirty clinical protocols have been approved by the Protocol Review Committees (PRC), Data Safety Monitoring Board

(DSMB) and the NIH. At this time 31 studies are included in ClinicalTrials.gov (<http://www.clinicaltrials.gov>) and 29 are actively recruiting patients.

International Conference on Rare Diseases and Orphan Drugs in Washington DC (ICORD 2008)

ICORD will be held May 20-22, 2008 in Washington, DC, in honor of the 25th Anniversary of the Orphan Drug Act. ICORD 2008 is planned in association with the National Organization for Rare Disorders' (NORD) annual Tribute Banquet and the NORD Corporate Council on May 20. To obtain the most recent information on the conference, please visit the ICORD website: http://www.icord.cc/washington_2008.php

Mission

The ICORD mission is to improve the welfare of patients with rare diseases and their families world-wide through better knowledge, research, care, information, education and awareness.

Aims of ICORD

- To organize International Conferences on Rare Diseases and Orphan Drugs (ICORD)
- To promote research, ethics, policies and actions on rare diseases and orphan products in all regions of the world

- To facilitate and provide a global forum for all stakeholders for effective communication, formation of opinion and public debate, concerning rare diseases and orphan products
- To enhance international discussion, cooperation and coordination of research, policies and actions of all bodies active in the field of rare diseases and orphan products
- To exchange best practices between existing bodies and develop international approaches and tools to address common issues in rare diseases and orphan products

Past Conferences have been held in the following locations:

- ICORD 2007 in Brussels
- ICORD 2006 in Madrid
- ICORD 2005 in Stockholm

Collaboration, Education, and Test Translation Program for Rare Genetic Diseases (CETT)

In the more than two years since ORD initiated the Collaboration, Education and Test Translation for Rare Genetic Diseases (CETT) Program, at least 35 new tests for

rare genetic diseases have either become available or are in development. The CETT Program is a pilot project of ORD, designed to move tests for rare genetic diseases from the research laboratory to

clinical settings by establishing collaborations among clinicians, researchers, laboratories and patient advocates.

Tests recently put into development and expected to be available soon include: DNMT2 Centronuclear Myopathy, ROR2 Robinson Syndrome, ASPM, CDK5RAP2, CENPOJ and MCPH1 for Autosomal Recessive Primary Microencephaly (University of Chicago); and ATP1A3 Rapid-Onset Dystonia Parkinsonism (Neurogenetics DNA Diagnostic Lab in Boston). Other tests put into development earlier include Urea Cycle Disorders (Baylor College of Medicine); Inclusion Body Myopathy Associated with Paget Disease and/or Frontotemporal Dementia (University of California at Irvine); and Duchenne Muscular Dystrophy and Becker Muscular Dystrophy (Emory University).

Kate Reed, MS, MPH, recently joined the CETT Program to coordinate educational material development. She is a project director at the National Coalition for Health Professional Education in Genetics (NCHPEG). Her work focuses on creating tools to help health professionals and the

public assess the accuracy and completeness of educational materials. She also works as a pediatric genetic counselor at Johns Hopkins University. Previous to her training in genetic counseling, Kate earned a M.P.H. in Public Health Genetics, which considers the impact of genetic information on the population.

The CETT Program Web site has two new features:

- New application is more focused, and has an accompanying guide to make it easier for applicants to complete. Nevertheless, applicants are encouraged to contact the program director (info@CETTProgram.org) early and often for information and guidance.
- CETTrack is an online application, communication tool to build collaboration among the researchers, clinicians, laboratories and patient advocates, and provide a review process.

For more information, see the CETT Program web site at www.CETTProgram.org or by email at info@CETTProgram.org

Scientific Conferences

The request for nominations for scientific conferences was sent to the IC Directors in early August 2007. The ORD review group made its decisions in October and memoranda of intent were sent on October 12. Of the 47 nominations received, 38 were selected to be co-supported by ORD. A list of the FY 2008 scientific conferences is posted on the ORD web site <http://rarediseases.info.nih.gov/Wrapper.aspx?src=asp/workshops/scicon.asp&PageID=5>.

A second request for scientific conferences for FY 2008 was sent to the IC Directors in early February. Forty-three selected to receive support from ORD.

The Office of Rare Diseases is pleased to announce that from the time this program began in FY 1995 until the present time we have co-supported 845 scientific conferences.

Biennial Report

The 2006 Biennial Report on the Rare Diseases Research Activities at the National Institutes of Health is now available on the Office of Rare Diseases website.

<http://rarediseases.info.nih.gov/asp/files/BienniaReportRareDiseasesFY2006Final.pdf>

This will be the final ORD initiated report. Future Rare Diseases Research Activities at the NIH will be reported in the NIH Annual Report.

ORD Website

The Office of Rare Diseases website (<http://rarediseases.info.nih.gov>) has been completely redesigned and recently was launched as the new public site. In addition to a more user-friendly layout, the new site features a more robust search engine, allowing visitors to find all relevant information in a format that is very similar to Google searches. Visitors to the new site have the option of going directly to a subject page using the tabs at the top or using the subject sections below to find out what links are being featured. Like the tabs at the top, the headings of the subject sections will take the visitor to the subject page where an expanded selection of links is available. We are continuing to make website improvements and will highlight some of these changes in future editions of the newsletter. The ORD welcomes all feedback regarding our website, as we continually strive to make it as useful as possible.

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.”

For additional information about ORD, contact:

Office of Rare Diseases
National Institutes of Health
6100 Executive Boulevard, 3B-01
Bethesda, Maryland 20892-7518
Telephone: (301) 402-4336
Fax: (301) 480-9655
ord@od.nih.gov

ORD Staff

Stephen C. Groft, Pharm.D.
Director
301-435-6041
ord@od.nih.gov

Mary Demory
Public Outreach and Education Program
301-402-4338
ord@od.nih.gov

Marita Eddy
Angel Flight NIH Coordinator
301- 451-9646
ord@od.nih.gov

David J. Eckstein, Ph.D.
Senior Health Science Administrator
301-496-0141
eckstein@od.nih.gov

John H. Ferguson, M.D.
Medical Advisor
301- 496-0109
jferguson@healthinfoconsultants.com

William Gahl, M.D., Ph.D.
Clinical Director, Intramural Research Program
301-402-8255
bgahl@helix.nih.gov

Rashmi Gopal-Srivastava, Ph.D.
Extramural Research Program
301-402-4336
ord@od.nih.gov

Chris Griffin
Program Analyst
301-496-0266
ord@od.nih.gov

Henrietta D. Hyatt-Knorr, M.A.
Policy and Program Planning and Analysis
301-435-6045
ord@od.nih.gov

Geraldine B. Pollen, M.A.
Senior Policy Advisor
301-402-4336
ord@od.nih.gov

Giovanna M. Spinella, M.D.
Science and Program Consultant
301-496-0139
ord@od.nih.gov