



FOCUS ON RARE DISEASES



THE OFFICE OF RARE DISEASES NEWSLETTER

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HIGHLIGHTS OF THE OFFICE OF RARE DISEASES RESEARCH ACTIVITIES

Rare Diseases Clinical Research Network 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 all of 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 (RFA) for the RDCRN**

The NIH Office of Rare Diseases (ORD), in collaboration with NIH Institutes and Centers, intends to publish a Request for Application (RFA) to continue the RDCRN Program. For details see Notice of Intent to Publish a Request for Applications (<http://grants.nih.gov/grants/guide/notice-files/NOT-OD-07-062.html>). The ORD and participating ICs continue to collaborate on the development of the RFA.

- **Educational Conference “Clinical Research for Rare Diseases: Opportunities, Challenges, and Solutions”**

The educational conference supported jointly by the NIH and RDCRN, “Clinical Research for Rare Diseases: Opportunities, Challenges, and Solutions,” was held in Bethesda, MD on Wednesday, September 5, 2007. This successful one-day conference focused on the methodology used to conduct clinical research in rare diseases. The target audience for this meeting included new investigators, trainees, junior faculty, and others interested in rare diseases clinical research. Speakers included expert

clinical investigators who are actively engaged in rare diseases research as well as members of the biopharmaceutical industry, the FDA, and NIH.

- **Clinical Protocols**

In the past twenty months, more than twenty five clinical protocols have been approved by the Protocol Review Committees (PRC), Data Safety Monitoring Boards, (DSMB) and the NIH, and a few more are under review. At this time Twenty eight studies are included in ClinicalTrials.gov (<http://www.clinicaltrials.gov>) and twenty five are actively recruiting patients.

Social Security Administration

The Office of Rare Diseases has been asked by the Social Security Administration (SSA) to help them improve their Compassionate Allowance program. The idea behind this program is that some decisions regarding disability payments should be streamlined; i.e., based on certain criteria such as diagnosis and disease state. The SSA published in the Federal Register its intent to improve the Compassionate Allowance program, which included a comment period that ended October 1, 2007. In addition, the

SSA held a two-day meeting on December 4-5, 2007 to solicit additional information from the public. The effort to help the SSA extends NIH-wide, and the ORD has provided a suggestion to include the following stakeholders: the relevant NIH Institutes and Centers, the medical community, the research community, and the patient advocacy groups. ORD will continue to talk with NIH participants and with SSA representatives regarding plan for participating in this endeavor.

NIH Office of Technology Transfer

The NIH Office of Technology Transfer (OTT) evaluates, protects, markets, licenses, and manages the wide range of NIH and FDA intramural discoveries, inventions, and other intellectual property as mandated by the Federal Technology Transfer Act and related legislation.

The most important developments in medical science typically begin in laboratories, such as discoveries of new biological molecules, processes, pathways, or innovative applications of existing knowledge. In most cases, the effects of these discoveries are limited to a narrow research goal. The impact on public health generally comes after

several more steps, including further research and development, testing, approval by appropriate regulatory bodies, manufacturing, and distribution.

OTT carries out its technology transfer mandate by retaining title to inventions developed by NIH and FDA intramural laboratories, and marketing and licensing these inventions to private entities to ensure their use, commercialization, and public availability.

Over the last 15 years, NIH has executed thousands of license agreements transferring NIH and FDA inventions to the private sector for further research and development and potential commercialization that can lead to public health benefits. For more information, visit the OTT Website at <http://www.ott.nih.gov>.

The OTT and ORD launched the Rare Disease and Conditions Technologies initiative less than a year ago. The website module that was developed to provide a more collaborative, consolidated, and systematic approach to the development of products for rare diseases and conditions has seen phenomenal growth. Not-for-profit organizations, academic research centers, and foundations in the United States and abroad are encouraged to submit technologies available for licensing from their institutions. Currently, 22 participating institutions have brought 295 rare disease related technologies to this exciting new initiative. In addition, OTT has contributed to this site 237 rare disease technologies developed by the NIH and FDA intramural research programs, and is constantly adding new ones as they become available.

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;
- 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; and
- 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, more than two dozen 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 Urea Cycle Disorders (Baylor College of Medicine) and Inclusion Body Myopathy Associated with Paget Disease and/or Frontotemporal Dementia (University of California at Irvine). Duchenne Muscular Dystrophy and Becker Muscular Dystrophy testing by array technology is now available from (Emory University). Four other applications are in review.

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.

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. Applicants are encouraged to contact the program director (info@CETTProgram.org) early and often for information and guidance.
- CETTrack is an online application and communication tool to build collaboration among the researchers, Clinicians, laboratories, and patient advocates.

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

Inventory of Biospecimen Repositories

This Web-based database, constructed by the RAND Corporation under a contract with ORD, should be fully operational in 2007/2008. The database will list existing repositories in the United States and in other countries. Feedback from researchers over time will identify collection, storage, and delivery issues that can impede research on rare diseases. The Web-based site for data entry by biospecimen repositories has been

pre-tested. In the near future, ORD plans to expand the information gathered and add international repositories. ORD also plans updates on an annual basis or when changes occur in the repositories to ensure that information is up to date.

National Disease Research Interchange

Since 2002, ORD has worked with the National Disease Research Interchange (NDRI) to remedy the unmet rare diseases research need for human tissue. Recently, the NDRI began working with patient advocacy groups through the *Rare Disease Biospecimen Alliance* to serve the rare diseases community and encourage tissue donations for research. NDRI is currently partnering with 14 Voluntary Health Organizations (VHOs). Five additional VHO partnering arrangements are pending and discussions are ongoing with another 17 VHOs. NDRI offers members of the Rare Diseases Biospecimen Alliance the following services for an annual membership fee:

- Consent of individuals interested in donating tissue for research either from surgery or post-mortem;
- Maintenance of a donor registry;
- Tissue procurement, limited banking of tissues, and listing on the NDRI online catalog;

- Direct shipment of biospecimens to researchers;
- Researcher recruitment, application, approval, and database management; and
- Generation of DNA and cell lines (at an additional cost).

The Alliance membership has doubled in the last few months. For donations or questions, call 1-800-222- NDRI (6374) or visit the NDRI Web site at <http://www.ndriresource.org/>. For information about the Rare Diseases Program, go to http://www.ndriresource.org/Biospecimens_Programs/Rare_Disease/30/ or call Sheila M. Curristin, Ph.D., Director, Rare Disease Program at (215) 557-7361 ext. 232 or via e-mail at SCurristin@ndriresource

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 46 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/>.

A second request for scientific conferences for FY 2008 will be sent to IC Directors in early February.

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 801 scientific conferences.

NIH Partners in Research Program (R03)

This Request for Application (RFA) has been developed as an initiative of the Director, NIH. All NIH Institutes and Centers will participate and the National Institute of Child Health and Human Development and the National Institute of Nursing Research will administer the RFA on behalf of the NIH. The purpose of the Partners in Research Program is to:

- Support two-year pilot and/or feasibility research studies of innovative activities designed to improve public understanding of biomedical and behavioral science;
- Develop strategies for promoting collaboration between scientists and the community to improve public health; and
- Identify the conditions that will enhance the effectiveness of such activities.

The long-term objectives are to:

- Raise the level of public awareness and understanding of biomedical and behavioral research related to health, the role they can play in the research enterprise, and the value of public funding;

- Increase scientists' awareness of the importance of public engagement; and
- Provide a menu of techniques for researchers and communities to help implement the participation of lay communities in the health research endeavor.

The NIH Public Trust initiative (<http://publictrust.nih.gov>) is the result of two separate, but related, streams of thought. The first is to increase the public trust in and understanding of the NIH and the second is to develop a new paradigm for the future of medical research. The mission is to enable the public to understand and have full confidence in the research that NIH conducts and supports nationally and internationally. For the NIH, the "public" comprises many groups of people, including the scientists who conduct the research, individuals (and their families) with health-related conditions, health care professionals and educators who treat or teach about these conditions, research advocacy organizations, and those in racial, ethnic or cultural groups who have special needs or are underserved.

Many of these “publics” are represented on the NIH Director’s Council of Public Representatives (COPR), which has provided valuable input to the Director

about a variety of issues, such as clinical research with human subjects, input into NIH research priority setting, and how to engender public trust.

Air Transportation Options for Patients

Mercy Medical Airlift (MMA), a non-profit organization with 25 years of medical air transportation experience, manages programs and services available to patients with both common and rare diseases. Several of the resources are mentioned in this article.

All are free services to patients.

www.mercymedical.org

For patients who need medical care en route, call Air Compassion America (ACAM) 1-866-270-9198. ACAM representatives locate and coordinate bed-to-bed air ambulance services with the overall goal of lowering costs. In the past year, ACAM has negotiated discounts with air ambulance companies and saved patients and their families between 30-40 percent. www.aircompassionamerica.org

One way you can help patients in medical and financial need is by donating your US Airways frequent flyer miles. Your contribution will be used for commercial airline tickets for patients who live farther than 1000 miles from their treatment or research facility. Go to <http://www.patienttravel.org/>. Click on the US Airways link. Donate to Air Charity Network (formerly Angel Flight America). Thank you for your generosity.

For patients who live within 1000 miles of their destination, Angel Flight may be an option. Angel Flight’s volunteer pilots fly children and adult patients to distant research and treatment centers throughout the United States. Pilots donate their time and the use of their planes in offering patients this free service for those who qualify. Angel Flight is a member of Air Charity Network (formerly Angel Flight America). ACN represents volunteer pilot associations across the country. www.aircharitynetwork.org

For patients traveling to any of the Rare Disease Clinical Research Network (RDCRN) sites or going to the National Institutes of Health (NIH) in Bethesda, Maryland, call Marita Eddy, 301-451-9646 or send an e-mail: meddy@mail.nih.gov. Angel Flight at NIH is supported by the Office of Rare Diseases.

www.rarediseases.info.nih.gov

For patients who are looking for travel help to other locations, call the National Patient Travel Center call 800-296-1217 for other available air transportation resources.

www.patienttravel.org

NORD’s Abbey Meyers Retires

Abbey Meyers, cofounder and President of the National Organization of Rare Diseases (NORD) for the past 29 years is retiring. On October 22 at a gala dinner in her honor numerous speakers from government,

industry, patient groups, and others celebrated her substantial contributions to the rare diseases community. Speakers included NIH director, Dr. Elias Zerhouni, FDA Commissioner, Dr. Andrew Von Eschenbach, NIH Office of Rare Diseases director, Dr.

Stephen Groft, and Dr. Tim Cote, director of the FDA's Orphan Products division. As a young mother of a child with a rare disease, Abbey Meyers learned about a major hurdle for people with rare disorders when a drug that was helping her child was terminated by the manufacturer because there were too few patients to assure financial returns for the company. In 1983, Ms. Meyers teamed with patient advocacy groups, members of Congress and their staffs, and the media to bring about passage of the Orphan Drug Act. The Act offered financial incentives to pharmaceutical companies to encourage them to develop drugs for rare conditions. The 25th anniversary of this landmark legislation will be celebrated this year on May 20th to 22nd.

As NORD president, Ms Meyers worked tirelessly for 25 years for patients with rare diseases, their families, and the patient advocacy groups representing these disorders. Besides the Orphan Drug Act, she worked to pass the Prescription Drug User Fee Act, which, among other things, established a clinical trials registry (clinicaltrials.gov) at the NIH. The registry has facilitated the awareness of, and the recruitment into, clinical trials for patients with rare diseases. Ms. Meyers and NORD also worked with Congress to pass the Rare Diseases Act of 2002, which established in law the NIH Office of Rare Diseases.

In recognition of her patient advocate contributions, Abbey Meyers has been invited to serve on many Federal government commissions and committees, including: the DHHS National Commission on Orphan Diseases (1986-1989), the DHHS National Human Research Protections Advisory Committee (2000-2002), the FDA Biological Response Modifiers Committee (1995-1999), the NIH Recombinant DNA Advisory Committee (1993-1996) and the National Human Gene Therapy Subcommittee (1989-1992), the NIH Panel on Physical Medicine and Rehabilitation Research (1989), and the NIH Special Emphasis Panel on the Coordination of Rare Diseases Research. (1987-1989).

She has been honored for her work with numerous awards, including the DHHS Public Health Service Award for Exceptional Achievement in Orphan Products Development and the FDA Commissioner's Special Citation for exceptional dedication and achievement on behalf of all people afflicted with rare disorders. She has also been Honorary President of the European Organization for Rare Disorders (EURORDIS), as well as a recipient of an honorary doctorate of humane letters in 1994 from Alfred University for her service to our nation, and has been recognized by *PharmaVoice* magazine as one of the 100 most influential people in the life sciences industry based on her work in rare diseases patient advocacy.

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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