



“The New Era in Health Care” to be Theme of 3rd Annual U.S. Conference on Rare Diseases and Orphan Products



How will the Affordable Care Act affect people with rare diseases? What is the investment outlook for orphan products? And what will our healthcare system look like in the future? These are all questions that will be addressed at the U.S. Conference on Rare Diseases and Orphan Products to be hosted by NORD and DIA Oct. 7 – 9 in Bethesda. [More](#).

FDA Posts Draft Guidance on Expedited Programs for Serious Conditions

FDA has published a draft guidance to outline policies and procedures for all four programs for expedited development and review of drugs and biologics to address unmet medical need in the treatment of serious or life-threatening diseases. August 26 is the deadline to submit electronic or written comments. [More](#).

Journal of Rare Disorders Now Online

The first issue of the new [Journal of Rare Disorders](#) is now available online. A call for manuscript submissions has been issued for future editions. Guidelines for submitting manuscripts are posted [here](#).

Policy News

Patient Organizations Join NORD in Supporting Compassionate Allowances Program

Approximately 60 patient advocacy groups signed a letter drafted by NORD seeking continued support for the Social Security Compassionate Allowances Program instituted by former Commissioner Michael Astrue. The program expedites processing of SSDI and SSI disability applications for patients with medical conditions that clearly meet SSA's requirements. NORD submitted the letter to Acting Commissioner Carolyn Colvin and members of the committees in Congress that oversee SSA. [Read the letter.](#)

NIH News

Public-Private Partnership Grants Announced

The National Institutes of Health has awarded \$12.7 million to nine academic research groups working with pharmaceutical industry compounds to explore new treatments in eight disease areas, including Duchenne muscular dystrophy and lymphangi leiomyomatosis (LAM). [More.](#)

FDA News

Request for Nominations

The FDA is requesting nominations for voting and/or nonvoting consumer representatives on public advisory committees. [More.](#)

Recent Drug Approvals

The FDA has approved Rixubis [Coagulation Factor IX (Recombinant)] to prevent or reduce the frequency of bleeding episodes prior to surgery in people with hemophilia B who are 16 years of age and older. [Read the press release.](#)

Recent Orphan Drug Designations

The National Tay-Sachs and Allied Diseases Association has received orphan drug designation for development of a gene therapy for Tay-Sachs disease and Sandhoff disease.

Elorac, Inc. has received orphan drug designation for naloxone lotion for topical treatment of pruritus accompanying cutaneous t-cell lymphoma (CTCL, also known as mycosis fungoides).

Alexion Pharmaceuticals, Inc. has received orphan drug designation for Soliris (eculizumab) for the treatment of neuromyelitis optica (NMO also known as Devic's disease).

Activartis has received orphan drug designation for its cancer immunotherapy AV0113 for the treatment of malignant glioma.

River Vision, Inc has received orphan drug designation for teprotumumab to treat active (dynamic) phase Grave's orbitopathy.

Recent Breakthrough Therapy Designations

GlaxoSmithKline and Prosensa Therapeutics have received breakthrough therapy designation for drisapersen, an exon-51 skipping compound, for the potential treatment of Duchenne muscular

dystrophy.

News from NORD Member Organizations

NORD Welcomes New Member Organization

NORD is happy to welcome the following new Member Organization:

CADASIL Association

Alpha-1 Foundation

The Foundation has announced that research grant opportunities are available for both in-cycle and out-of-cycle timelines. In-cycle letters of intent are due Sept. 20. [More](#).

American Partnership for Eosinophilic Disorders (APFED)

APFED is accepting applications from researchers for the next cycle of their HOPE Pilot Grant awards. Investigators initiating new projects relevant to eosinophil-associated diseases that are likely to lead to future external funding are invited to submit an application by October 1. [More](#).

Association for Glycogen Storage Disease (AGSD)

The 2013 AGSD Conference will be held Sept. 20-21 in Raleigh-Durham, NC. [More](#).

Canadian Organization for Rare Disorders (CORD)

CORD has announced that its annual conference will be held Sept. 29-Oct. 1 in Ottawa. [More](#).

Cystinosis Research Network (CRN)

The CRN Family Conference will be held July 18-20 in Washington, DC. NORD representatives will participate, and speakers will include William A. Gahl, M.D., Ph.D., Director of the NIH Undiagnosed Diseases Program, and Jess Thoene, MD, Professor Emeritus of Pediatrics, University of Michigan [More](#).

Fibromuscular Dysplasia Society of America (FMDSA)

The FMDSA has formed the Mount Sinai Heart Center for Fibromuscular Disease Care and Research in order to study the cause and full clinical spectrum of FMD. [More](#).

Global Foundation for Peroxisomal Disorders (GFPD)

The GFPD will hold its 2013 Family and Scientific Conference July 26-30 in Lincoln, Nebraska. [More](#).

Lymphangiomatosis & Gorham's Disease Alliance (LGDA)

The LGDA has announced that funding is available for research grants. [More](#).

TNA-The Facial Pain Association

The TNA has announced that its 2013 National Conference will be held Oct. 4-6 in San Diego, CA.

[More](#).

Patient and Family Resources

Apps for Children with Special Needs and Learning Differences

Common Sense Media provides recommendations for products that help children who struggle with traditional methods of learning. [More](#).

Patient Recruitment

*Anyone considering participating in a clinical trial should discuss the matter with his or her physician. **NORD does not endorse or recommend any particular studies.***

Chronic Thromboembolic Pulmonary Hypertension (CTEPH)

Bayer HealthCare Pharmaceuticals is sponsoring a clinical trial to provide access to riociguat for patients with inoperable CTEPH or recurrent or persisting pulmonary hypertension after surgical treatment who are not satisfactorily treated or cannot participate in any other CTEPH trial. The study will also evaluate the safety, tolerability and clinical effects of riociguat. [More](#).

Congenital Adrenal Hyperplasia (CAH)

The National Institute of Child Health and Human Development is sponsoring a study to compare the effectiveness of a cortisol pump with standard cortisol pill therapy for adults with classic CAH. [More](#).

Rare Disease Testing

ACMG Standards and Guidelines for Fragile X Testing Revised

Mutations in the FMR1 gene are associated with fragile X syndrome, fragile X tremor ataxia syndrome, and premature ovarian insufficiency. This document provides updated information regarding FMR1 gene mutations, including prevalence, genotype–phenotype correlation, and mutation nomenclature. [Read the guidelines](#).

Research

Genes Identified for Primary Focal Dystonia

Analysis of data from the Dystonia Coalition DNA Biorepository has resulted in identification of four candidate genes for adult-onset focal dystonias. [More](#).

Promising Gene Therapy Trials for Metachromatic Leukodystrophy and Wiskott-Aldrich Syndrome

Early results from two separate phase I/II trials involving six patients with these diseases indicate that a lentiviral vector to deliver the genes appears to be safe and effective in halting disease

progression. In addition, no evidence was found that this method inserted the genes near cancer causing genes, which was a problem in earlier studies. [More](#).

Treatment

Diagnosis and Management Recommendations for Q Fever

The Centers for Disease Control and the Q Fever Working Group have developed the first national recommendations for clinical and laboratory diagnosis, treatment, management and reporting for Q fever. [More](#).

American Heart Association (AHA) Consensus Statement on “Cardiovascular Function and Treatment in Beta-Thalassemia Major”

The AHA statement indicates that serial cardiac MRI is the best predictor of cardiac iron overload, which can lead to heart failure. The statement also points out that acute heart failure in thalassemia major requires continuous uninterrupted infusion of high-dose deferoxamine, augmented by oral deferiprone. [More](#).

International News

New Zealand Organisation for Rare Disorders (NZORD)

NZORD is sponsoring a one-day seminar to discuss access to orphan drugs for rare diseases in New Zealand on August 1 in Wellington. [More](#).

International Conference on Rare Diseases and Orphan Drugs (ICORD)

The 8th ICORD meeting will be held Nov. 1-2 in Saint Petersburg, Russia. [More](#).

Upcoming Meetings and Webcasts

7th International Patient Symposium on Myeloproliferative Diseases

The Cancer Research & Treatment Fund is sponsoring this symposium on Nov. 6 in New York City. [More](#).

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