



NORD Hosts Capitol Hill Briefing on Rare Diseases in Changing Healthcare Landscape



Presenters at NORD's recent Capitol Hill briefing on the topic "Rare Diseases in a Changing Healthcare Landscape" were (from left): Stephanie Bozarth of the National MPS Society; Andrew Emmitt, BIO; Sandy Robinson, Avalere Health; Peter L. Saltonstall, NORD; Richard Zyne, CurePSP; and Charles Mohan, United Mitochondrial Diseases Foundation. Watch for a video of this event to be posted on the NORD [website](#) soon. [More.](#)

Nearly 120 Patient Organizations Sign NORD Letter Regarding Orphan Drug Tax Credit

Nearly 120 patient organizations signed on to a letter NORD wrote to the Senate Finance Committee and House Committee on Ways and Means urging that the Orphan Drug Tax Credit (ODTC) not be repealed. This was in response to a general tax reform proposal made public recently that would include repeal of the ODTC. Read [NORD's position paper](#). [Read the letter](#).

NORD Supports FDA's Exercise of Appropriate Flexibility

A recent study published in the Journal of the American Medical Association has led to debate about whether FDA should employ a "one size fits all" approach to drug reviews. [Read letter NORD submitted to JAMA on this topic](#).

Urge Your Senators to Support NIH Funding

Senators Richard Burr (R-NC) and Robert Casey, Jr. (D-PA) are circulating a sign-on letter in support of critically needed funding for the National Institutes of Health (NIH). To help assure that NIH receives the funding it needs to support vitally important medical research, Research!America is conducting a campaign that NORD supports to encourage individuals to ask their senators to sign this bipartisan letter. [More](#).

Save The Date For The Annual NORD Gala

Please plan to join us in Washington DC on Thursday, May 8, for NORD's annual gala which will have a "Portraits of Courage" theme this year. [More](#).

FDA Issues First Pediatric Rare Disease Priority Review Voucher

FDA has issued the first Priority Review Voucher for a pediatric rare disease application, as authorized in the FDA Safety and Innovation Act of 2012. The PRV was issued to BioMarin Pharmaceutical Inc. for Vimizim, approved recently for patients with MPS type IVA. Read an [interview](#) with Dr. Anne Pariser and Dr. Andrew Mulberg, both of FDA, on this and related topics. (You'll need to sign up for a free password to Medscape to access this article.)

NIH News

Extramural Research Awards Announced

Ten projects will enable non-government researchers to conduct clinical research at the National Institutes of Health Clinical Center through three-year, renewable awards of up to \$500,000 per year. The awards will support projects on a variety of diseases and conditions, including Niemann-Pick disease type C and childhood leukemia. [More](#).

FDA News

Public Meeting on Pulmonary Arterial Hypertension Patient-Focused Drug Development Set for May 13

FDA will host a public meeting on patient-focused drug development for pulmonary arterial hypertension (PAH) on May 13. The purpose will be to obtain patient input on the impact of PAH on daily life and patients' views on currently available therapies. [More.](#)

Educational Resources on Rare Diseases for Patients, Advocacy Groups, Research Investigators and Drug Developers

FDA has launched a web page with links to recorded information about FDA and rare disease issues. [More.](#)

Advisory Committee Membership Nomination Portal Launched

An online, interactive system is now available that allows interested individuals to submit nominations for membership to any of FDA's 33 advisory committees. [More.](#)

Fiscal Year 2015 FDA Proposed Budget Announced

The President is requesting a \$4.7 billion budget for FDA, which is an 8.1% increase over the 2014 budget Congress passed earlier this year. [More.](#)

Recent Drug Approvals

Myalept (metreleptin for injection) has been approved as a replacement therapy to treat the complications of leptin deficiency in patients with congenital generalized or acquired generalized lipodystrophy. Myalept is marketed by Amylin Pharmaceuticals. [Read the press release.](#)

Kalydeco (ivacaftor) is now approved for expanded use to treat individuals age six and older who have eight additional cystic fibrosis mutations. [More.](#)

Impavido (miltefosine) has been approved to treat the tropical disease leishmaniasis caused by Leishmania, a parasite transmitted to humans through sand fly bites. Impavido previously received fast track designation, priority review, and orphan product designation. This medication is manufactured by Paladin Therapeutics. [Read the press release.](#)

Recent Orphan Drug Designations

Daval International has received orphan drug designation for Aimspro for the treatment of systemic sclerosis (scleroderma).

XOMA Corporation has received orphan drug designation for gevokizumab for the treatment of pyoderma gangrenosum. This drug previously received orphan drug designation for three other

indications: non-infectious, intermediate, posterior or pan uveitis; chronic non-infectious anterior uveitis; and Behçet's uveitis.

MEI Pharma, Inc. has received orphan drug designation for Pracinostat for the treatment of acute myeloid leukemia.

Recent Fast Track Designations

Edison Pharmaceuticals has received fast track designation for EPI-743 for the treatment of Friedreich's ataxia.

News from NORD Member Organizations

NORD Welcomes New Members

NORD is happy to welcome the following new members:

[The XLH Network](#)

[RASopathies Network USA](#)

Alagille Syndrome Alliance (ALGSA)

The ALGSA is seeking candidates for associate executive director. The application deadline is April 11. [More.](#)

Association for Glycogen Storage Disease (AGSD)

The 36th Annual AGSD Patient/Family/Professional Conference will be held Sept 19-20 in Detroit-Dearborn. [More.](#)

Friedreich's Ataxia Research Alliance (FARA)

FARA is seeking applications for the Phillip Bennett and Kyle Bryant Translational Research Award that focuses on pre-clinical and clinical investigations that will advance treatments for FA. The deadline for letters of intent is May 15. [More.](#)

The LAM Foundation

Sue Byrnes, Co-founder of the LAM Foundation, will retire later this year. The Sue Byrnes Legacy Fund has been established to ensure the future growth of the Foundation. [More.](#)

The Marfan Foundation

Carolyn Levering, President & CEO of the Marfan Foundation, has announced a two-year

transition plan leading to her retirement. [More.](#)

Myotonic Dystrophy Foundation (MDF)

The MDF Annual Conference will be held Sept 12-13 in Washington, DC. [More.](#)

National Hemophilia Foundation (NHF)

The NHF has launched a new [advocacy website](#). Also, nominations are being sought for the 2014 Annual Meeting Awards of Distinction in Programs and Awards of Excellence. The deadline for submissions is May 19. [More.](#)

NephCure Foundation

The Foundation has launched the NephCure Kidney Network, a registry of people with focal segmental glomerulosclerosis, minimal change disease, membranous nephropathy, and other rare diseases that cause primary nephrotic syndrome. [More.](#)

Patient Recruitment

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

Rare Disease Diagnosis

Childhood Stroke Gene Identified

Researchers have found that mutations in the CECR1 gene can prevent production of the adenosine deaminase 2 (ADA2) enzyme that is important to the integrity of blood vessel walls, leading to inflammation and an increased risk for stroke in children. Further study of the ADA2 pathway may yield clues to the underlying cause of stroke and other vascular disorders. [More](#)

Gene for Severe Cushing Syndrome Identified

A sub-group of patients with Cushing syndrome have benign or malignant tumors of the adrenal glands that secrete excessive cortisol. A recent study found that mutations in the PRKACA gene appear to increase production of the PKA enzyme, resulting in increased growth of adrenal tissue and an overproduction of cortisol. [More.](#)

International News

International Pompe Day

The first International Pompe Day will be held April 15. [More.](#)

European Conference on Rare Diseases and Orphan Products

Patients, policy makers, healthcare professionals, industry, payers, regulators, researchers and academics will gather for this conference in Berlin May 8-10. [More.](#)

EnRich Innovative Trial Design Course

This course is designed for pediatric healthcare and research professionals with experience in clinical trial development and an interest in better designed and more impactful trials. This three-day course will be held May 21-23 in Toronto. [More.](#)

Upcoming Meetings and Webcasts

Disability Policy Seminar

The 2014 Disability Policy Seminar will be held April 7-9 in Washington, DC. [More.](#)

NCCN Policy Summit: Designing Clinical Trials in the Era of Multiple Biomarkers and Targeted Therapies

The National Comprehensive Cancer Network (NCCN) will host this summit April 25 in Bethesda, MD. The program is designed for multiple stakeholders including physicians, industry, government, patients, and other interested parties. [More.](#)

Miscellaneous

Patient Advocacy Leadership (PAL) Awards

Genzyme has launched its fourth annual PAL Awards program for non-profit organizations that serve the lysosomal storage disorder patient community. Grants will be awarded for initiatives that support disease awareness and education, community mobilization, non-profit development and good governance activities, patient care and support programs. Proposals must be received by June 6. Awards will be announced by September 30. [More.](#)

Websites and Blogs of Interest

Putting Patients First

The National Health Council has launched two tools on [PuttingPatientsFirst.net](#) that provide resources to help people make informed decisions about purchasing health insurance on the marketplace. The [Estimate My Costs](#) tool helps people project how their annual anticipated medical services and drug usage will impact their health care costs across the "metal" plans offered in their state. The [Explore My Options](#) tool – allows people to review a set of six different health scenarios to see how different health conditions, medical and drug use, and “metal” plans

impact out-of-pocket costs.

PhenomeCentral

PhenomeCentral is a secure data repository for clinicians and scientists working in the rare disease community to share information. The platform enables researchers to learn about patients with similar undiagnosed conditions, which may eventually lead to understanding the underlying cause. Several major rare disease research programs, including the NIH Undiagnosed Diseases Program, are contributing cases to PhenomeCentral. [More](#).

MassGenomics

Posts on this blog cover in-depth reviews of recent research publications or editorials from the perspective of a researcher working at a large-scale sequencing center. The most recent post is regarding analysis of exome data in rare Mendelian disorders. [Visit the blog](#).

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