



Join NORD in Promoting Rare Disease Day 2014!



Are you wondering how to show your support for Rare Disease Day 2014? NORD has suggested activities for individuals, schools and universities, companies, patient organizations and others. Visit the [official national website](#) for Rare Disease Day to learn about opportunities to get involved in the U.S. Visit the [official global website](#) to learn what others are doing around the world.



Legislative Awareness Events Being Organized in Several States

Patient advocates, medical professionals, orphan product developers and others are collaborating on Rare Disease Day “State House Events” to increase understanding and awareness among legislators in several states. Read about how to connect with others in your state to organize such an event. [More](#).

NORD Commends Expansion of SSA’s “Compassionate Allowances” List

The Social Security Administration (SSA) has announced that 25 medical diagnoses have been added to the Compassionate Allowances list. This initiative provides expedited review of applications for disability assistance from individuals diagnosed with conditions that invariably meet SSA criteria. [Read NORD’s press release](#).

Policy News

PCIP Will Extend Coverage Through March

The federally-run Pre-existing Condition Insurance Plan (PCIP) will offer the option of two

additional months of coverage to people currently enrolled in PCIP who have not yet found new health insurance coverage. This transitional coverage through March 31 will allow PCIP enrollees more time to review Marketplace plan options. [More.](#)

NIH News

NIH Campaign To Improve Access to Pediatric Palliative Care

The National Institute of Nursing Research has launched a new campaign to increase the use of palliative care — comprehensive treatment of the discomfort, symptoms, and stress of serious illness — for children with serious illness. [More.](#)

FDA News

Recent Drug Approvals

Orenitram (treprostinil) has been approved for the treatment of pulmonary arterial hypertension (PAH). Orenitram is manufactured by United Therapeutics Corporation. [More.](#)

Tretten, a recombinant factor XIII replacement product, has been approved for the prevention of bleeding in adults and children who have the rare clotting disorder congenital factor XIII A-subunit deficiency. Tretten is distributed by Novo Nordisk, Inc., USA. [Read the press release.](#)

Mekinist (trametinib) in combination with Tafinlar (dabrafenib) has been approved to treat patients with advanced melanoma that cannot be removed by surgery or is late-stage. These are the first drugs approved for combination treatment of melanoma. Mekinist and Tafinlar are marketed by GlaxoSmithKline. [Read the press release.](#)

Recent Orphan Drug Designations

Omeros Corporation has received orphan drug designation for OMS721, a human monoclonal antibody targeting mannan-binding lectin-associated serine protease-2 (MASP-2), the key regulator of the lectin pathway of the immune system, for prevention of complement-mediated thrombotic microangiopathies.

Veloxis Pharmaceuticals has received orphan drug designation for Envarsus, a once-daily form of tacrolimus, for the prevention of kidney-transplant rejection.

RegeneRx Biopharmaceuticals, Inc. has received orphan drug designation for its drug candidate, Thymosin beta 4 (Tβ4), for the treatment of neurotrophic keratopathy, a serious degenerative disease of the outside layer of the eye.

DiaVacs, Inc. has received orphan drug designation for DV-0100, the company's type 1 diabetes mellitus therapy.

Alexion Pharmaceuticals has received orphan drug designation for Soliris for the prevention of

delayed graft function, a complication for kidney-transplant patients.

News from NORD Member Organizations

NORD Welcomes New Member Organization

NORD is happy to welcome the following new Member Organization:

Rothmund-Thomson Syndrome Foundation

Alpha-1 Foundation

The TV series "Healthy Body, Healthy Mind" has done a 2-part segment on Alpha-1, featuring Alpha-1 specialists and offering the patient perspective. [More.](#)

Applications are now being accepted for the Gordon L. Snider Scholar Award, a research grant named in memory of a great researcher and longtime Board member who helped to create the Foundation's research program. Applications are due March 28. [More.](#)

Alport Syndrome Foundation (ASF)

The ASF, Macquarie, Pedersen Family and the Kidney Foundation of Canada (KFOC) have announced the availability of funding for basic science and clinical research on the natural history, biochemical basis, and treatment of Alport syndrome. Two research projects will be selected for funding, totaling \$100,000 USD each. Applications are due March 17. [More.](#)

March will be the first-ever Alport Awareness Month, a time for Alport syndrome families to raise awareness in their local communities during National Kidney Month, which occurs annually in March throughout the US. [More.](#)

Aplastic Anemia & MDS International Foundation

Aplastic Anemia and MDS Awareness Week is set for March 1-7 in conjunction with [Rare Disease Day](#), which is observed on February 28. [More.](#)

Genetic Alliance

Genetic Alliance, as part of the National Genetics Education and Consumer Network (NGECN), is inviting proposals for the Impact Awards. Up to \$25,000 will be offered per award, and a total of \$175,000 will be distributed as part of the award process. Letters of intent are due Feb 15. [More.](#)

The LAM Foundation

The International Lymphangiomyomatosis Research Conference and Patient & Family Educational LAMposium will be held March 28-30 in Chicago. [More.](#)

National PKU Alliance (NPKUA)

The NPKUA has announced that Henry Joel Mroczkowski PhD, MD, is the 2013 recipient of the Dr. Koch Memorial Scholarship. [More.](#)

National Tay-Sachs & Allied Diseases Association, Inc. (NTSAD)

The NTSAD is soliciting proposals for innovative research projects that involve basic research, translational studies or clinical studies in the areas of neurodegenerative disorders affecting the central nervous system, especially lysosomal storage disorders and pediatric leukodystrophies. Proposals are due Feb 7. [More.](#)

PRISMS (Parents and Researchers Interested in Smith-Magenis Syndrome)

PRISMS 8th International Conference, "Building Bridges of Hope" will be held July 31-Aug 3 in St. Louis, MO. [More.](#)

United Leukodystrophy Foundation (UDF)

The UDF is accepting applications to support research on cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) and plans to award one grant of \$25,000. Applications are due Feb 1. [More.](#)

Provider Resources

New NCHPEG Programs for Pediatricians and Neurologists

The National Coalition for Health Professional Education in Genetics (NCHPEG), The Jackson Laboratory, and partners have announced the launch of two new educational programs for pediatric and neurology providers. [More here](#) and [here](#).

Treatment

ACMG Guidelines for Management of PKU

The American College of Medical Genetics (ACMG) has published a new guideline that supports the need for life-long management of phenylalanine levels in individuals with PKU. [More.](#)

Diagnosis and Treatment Recommendations for Neuromyelitis Optica (NMO)

The German Neuromyelitis Optica Study Group has published a report stating that testing for the serum marker AQP4-Ab and imaging techniques are necessary for the diagnosis of NMO. Azathioprine and rituximab are suggested for first-line treatment, and other immunosuppressive drugs are recommended for second-line treatment. [More.](#)

International News

Genomic Disorders 2014: The Genomics of Rare Diseases

This meeting will be held March 5-7 in Cambridge, UK, focusing on how human genome analysis can best assist future clinical practice and patient care. [More.](#)

International Euromit Patient Meeting

This meeting will be held June 18-19 in Tampere, Finland in conjunction with the International Meeting on Mitochondrial Pathology. Lectures and panel discussions on mitochondrial disease, treatment, and latest research findings will be included. The working language of the meeting will be English, and simultaneous translation will be provided into and from Finnish and Swedish, plus into other languages subject to demand. [More.](#)

Upcoming Meetings and Webcasts

Assessing Genomic Sequencing Information for Health Care Decision Making: A Workshop

The Institute of Medicine's Roundtable on Translating Genomic-Based Research for Health will host a public workshop to evaluate how evidence for genomic applications is gathered and assessed for clinical decision-making, reimbursement decisions, and guideline development in the absence of an ideal information base. This workshop will be held Feb 3 in Washington, DC. [More.](#)

Miscellaneous

Million Dollar Bike Ride for Rare Disease Research

The first annual [Million Dollar Bike Ride](#) to raise money for rare disease research, organized by the Penn Center for Orphan Disease Research and Therapy and Rare Disease Cycling, will be held May 3. Several NORD Member Organizations plan to participate.

Websites and Blogs of Interest

Rare Coagulation Disorders Resource Room

The Resource Room provides current and searchable information on the basic science, clinical management, available laboratory and genetic testing, clinical trials, and global research initiatives for very rare and heterogeneous coagulation disorders. This website was developed through a collaboration of the [International RBDD Registry](#), the [Indiana Hemophilia & Thrombosis Center](#), and the Rare Coagulation Disorders Subcommittee of the [National Hemophilia Foundation](#). [Visit the website.](#)

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