NORD and RDLA to Co-Host Briefing with Rare Disease Congressional Caucus Co-Chairs

NORD and Rare Disease Legislative Advocates (RDLA) will host a briefing on May 7 in coordination with Rare Disease Congressional Caucus Co-Chairs Rep. Leonard Lance (R-NJ) and Rep. Joe Crowley (D-NY) on “Access to Care and Therapies in the New Healthcare System: A Rare Disease Perspective”. This will take place from noon to 1 p.m. in the Rayburn House Office Building, Room B-318. Seating is limited. **RSVP to attend.**

“Portraits of Courage” to Honor Spirit of Patients and Caregivers at NORD Gala

With a Portraits of Courage theme, NORD will feature patients, parents and caregivers – many of whom will be present to participate in the program – at its annual gala on Thursday, May 8, at the National Building Museum in Washington DC. Honorees will include two members of Congress – Representative Fred Upton and Senator Sherrod Brown – for advancing research on diseases affecting children. **More**.

NORD Senior VP Named to PCORI Advisory Panel on Rare Disease

J. Russell Teagarden, senior VP of medical & scientific affairs at NORD, is one of 13 individuals named to an inaugural Advisory Panel for Rare Disease for the Patient-Centered Outcomes Research Institute (PCORI). Panel members will advise PCORI on its research priorities in the area of rare disease, as well as on engaging with the rare disease research community. **More**.

FDA News

New Expedited Access Program for Medical Devices Proposed

The Expedited Access Premarket Approval Application for Unmet Medical Needs for Life Threatening or Irreversibly Debilitating Diseases or Conditions ("Expedited Access PMA" or

"EAP") program has been proposed by the FDA to provide patients with earlier access to safe and effective medical devices. The EAP would seek to speed the product development timeline as well as reduce the time for premarket review. To be eligible for participation in the program, the medical device must meet specific criteria. **More**.

FDA and NORD to Co-Sponsor Public Meeting on Immune Responses to Enzyme Replacement Therapies

The FDA and NORD are co-sponsoring a 1-day public workshop on June 9 in Silver Spring, MD, to discuss the role of immune tolerance induction in patients receiving replacement biological products. Attendance is free and preregistration is encouraged. **More**.

FDA to Host Public Meeting on Inborn Errors of Metabolism Patient-Focused Drug Development

FDA will host a public meeting on patient-focused drug development for neurological manifestations of inborn errors of metabolism on June 10. The purpose will be to obtain patient input on the impact of the neurological manifestations of inborn errors of metabolism on daily life and patient views on treatment options. **More**.

Recent Orphan Drug Approvals

Alprolix, coagulation factor IX (recombinant), Fc fusion protein, has been approved for use in adults and children with hemophilia B. Alprolix is manufactured by Biogen Idec, Inc. **Read the press release**.

Cyramza (ramucirumab) has been approved to treat patients with advanced stomach cancer or gastroesophageal junction adenocarcinoma, a form of cancer located in the region where the esophagus joins the stomach. Cyramza is marketed by Eli Lilly. **Read the press release**.

Sylvant (siltuximab) has been approved to treat patients with multicentric Castleman's disease (MCD). This is first FDA-approved drug to treat patients with MCD. Sylvant is marketed by Janssen Biotech Inc. **Read the press release**.

Recent Orphan Drug Designations

Kite Pharma Inc. has received orphan drug designation for an autologous engineered T cell product for the treatment of diffuse large B cell lymphoma.

Boehringer Ingelheim has received orphan drug designation for volasertib for the treatment of acute myeloid leukemia. This medication previously received a breakthrough therapy designation.

Emergent BioSolutions has received orphan drug designation for its BioThrax vaccine to treat patients who have been exposed or are believed to have been exposed to anthrax.

Tolero Pharmaceuticals, Inc. has received orphan drug designation for Alvocidib for the treatment

of patients with acute myeloid leukemia.

Bayer HealthCare has received orphan drug designation for Ciprofloxacin Dry Powder for Inhalation (Ciprofloxacin DPI) for the treatment of non-cystic fibrosis bronchiectasis.

News from NORD Member Organizations

Alpha-1 Foundation

Longtime Alpha-1 advocate Karen Erickson has joined the Alpha-1 Foundation as their Associate Executive Director of Community Engagement. **More**.

American Partnership for Eosinophilic Disorders (APFED)

Registration is open for APFED's 12th Annual Patient Conference on Eosinophilic Gastrointestinal Disorders, which will take place July 11-12 in Denver, CO. A limited number of travel grants is available to help eligible families offset conference-related costs. **More**.

ECD Global Alliance

The 2nd International Erdheim-Chester Disease (ECD) Medical Symposium will be held Sept 18 at the NIH in Bethesda, MD, followed by the 2nd Annual International ECD Patient and Family Gathering on Sept. 19-20. **More**.

Ehlers-Danlos National Foundation (EDNF)

The EDNF 2014 Annual Learning Conference will be held in Houston, TX, July 10-12. **More**.

National Ataxia Foundation (NAF)

Funding is available for FY 2015 NAF Research Grants, Young Investigator Awards, Fellowship Awards, and Pioneer SCA Translational Research Awards. Letters of intent are required. **More**.

National Hemophilia Foundation (NHF)

The NHF will hold its 66th Annual Meeting, “Nothing About Us Without Us” Sept 18-20 in Washington, DC. **More**.

NephCure Foundation

The Foundation has announced that the 10th International Podocyte Conference will take place June 4-6 in Freiburg, Germany. The goal of the 2014 conference is to share the latest clinically relevant research on kidney podocytes (cells that filter blood in the glomerulus of the kidney). **More**.

Oxalosis and Hyperoxaluria Foundation (OHF)

The OHF has announced that the 2014 Patient Meeting & Walk and 11th International Primary Hyperoxaluria Workshop will be held June 27-29 in Chicago. These events are open to anyone affected by oxalosis, primary hyperoxaluria & other hyperoxaluria conditions - patients, relatives, friends & clinicians. **More**.

Patient and Family Resources

Connecting Kids to Coverage

Enrollment of children and teens for free or low-cost health insurance coverage through Medicaid and the Children's Health Insurance Program (CHIP) is ongoing. Visit **http://www.insurekidsnow.gov/**for state-by state resources.

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.

Klinefelter's Syndrome/47 XXY, Triple X Syndrome/Trisomy X/47 XXX, and 47 XYY

The Department of Experimental Psychology at the University of Oxford is seeking children ages 5-16 years with an extra sex chromosome who live in the UK to study language, reading and communication problems. The study involves a 2 hour assessment of reading, speaking, reasoning and language tasks at home or school; a test to measure blood flow in the two halves of the brain; a saliva sample from children and parents for DNA testing; and parental completion of 3 questionnaires about the child's development. For more information contact: Louise Atkins at**louise.atkins@psy.ox.ac.uk** or +44(0)1865 271386.

Rare Disease Diagnosis

International Consensus (ICON) Document Published to Encourage Early Diagnosis of Congenital Immunodeficiencies

Early diagnosis of congenital immunodeficiencies is necessary to provide appropriate treatment that benefits affected children. This ICON paper outlines features of the most common congenital immunodeficiencies including innate immunodeficiencies, phagocytic deficiencies, complement deficiencies, disorders of T cells and B cells (combined immunodeficiencies), antibody deficiencies, and immunodeficiencies associated with syndromes. **More**.

Dental, Oral and Craniofacial Manifestations of Rare Bone Diseases

A recent paper published by the International and American Associations for Dental Research focuses on dental, oral and craniofacial manifestations of rare bone diseases. Four key

physiologic processes are discussed: bone/tooth formation (fibrous dysplasia), extracellular matrix production (osteogenesis imperfecta), mineralization (familial tumoral calcinosis/hyperostosis hyperphosphatemia syndrome, hypophosphatemic rickets, and hypophosphatasia), and bone resorption (Gorham-Stout disease). **More**.

Treatment

Acetazolamide Helps Improve Vision in Idiopathic Intracranial Hypertension (IIH)

The NIH-funded IIH Treatment Trial found that acetazolamide (Diamox), an inexpensive glaucoma drug, was effective in preserving and restoring vision for women with IIH. This medication is commonly prescribed for IIH, but this study provides much-needed evidence of its benefit. **More**.

Upcoming Meetings and Webcasts

Emerging Therapies for Rare Diseases

The Center for Orphan Disease Research and Therapy at the University of Pennsylvania Perelman School of Medicine will hold their First Annual Symposium on Rare Disease, "Emerging Therapies for Rare Diseases" on May 2 in Philadelphia. Registration is free of charge. **More**.

Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (DACHDNC)

The next meeting of the DACHDNC will be May 29-30 in Rockville, MD. The meeting is open to the public with in-person attendance limited to space availability, but the meeting can also be viewed via webinar. All participants, in-person and web, must register by May 2. **More**.

Symposium on Regulatory Challenges for Orphan Medicines

This symposium will take place on June 18 at 3:30 p.m during the DIA 2014 50th Annual Meeting in San Diego, and focus on the regulatory environment and trends for orphan products around the world. **More**.

Miscellaneous

ACMG Updates Recommendation on "Opt Out" for Genome Sequencing Return of Results

The American College of Medical Genetics and Genomics (ACMG) issued new guidelines that make it possible for patients who have genome sequencing to opt out of receiving information about gene alterations not related to the indication for testing. The new recommendation also states that patients should be informed of the opt-out option and its implications by the ordering clinician before the test is ordered. **More**.