Indications™

Newsletter of The Lysosomal Disease Network™

December 2014 **Vol. 2, No. 1**

National Tay-Sachs and Allied Diseases Association Announces Competitive Grant Availability

By Evelyn S. Redtree, M.S.

NTSAD (see "**Indications**" September 2014) 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 disorders (such as Tay-Sachs, GM-1, Sandhoff diseases) and pediatric leukodystrophies (such as Canavan disease). Basic research and translational studies should generate strong preliminary data to enable future major funding by other third parties. Projects may be in such areas as drug delivery to the brain, new animal models designed to facilitate translational research and drug discovery, assay development for drug screening, substrate reduction, stem cells, molecular chaperones, gene therapy, and biomarkers, as well as exploring other novel therapeutic strategies.

NTSAD's updated research strategy has identified the development of Clinical Trial Readiness as a primary strategic objective. Therefore, in addition to soliciting proposals for novel research projects, the NTSAD strongly encourages applicants to submit

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In This Issue. . .

Meet Our Patient Advocacy Groups

The LDN considers itself lucky to have dedicated patient advocacy groups as members of the Network. This issue, we present with gratitude . . .

The Ryan Foundation for MPS Children

http://ryanfoundation.net/home/

The Ryan Foundation for MPS Children was founded in October 1992 by Mark and Jeanne Dant following their son Ryan's diagnosis with MPS I, at age 3. The Ryan Foundation was founded to raise funds for scientific research leading to effective treatments and a cure for MPS I. (Readers of "**Indications**" can learn more about the MPS diseases by reading the "Check Your Knowledge" article on page 5 of the first issue of "**Indications**," published December 2013. This is available for download via hyperlink in the upper-right area of the LDN homepage at: www.lysosomaldiseasenetwork.org.) Mark Dant continues to serve as volunteer

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The Lysosomal Disease Network (U54NS065768) is a part of the National Institutes of Health (NIH) Rare Diseases Clinical Research Network (RDCRN), supported through collaboration between the NIH Office of Rare Diseases Research (ORDR) at the National Center for Advancing Translational Science (NCATS), the National Institute of Neurological Disorders and Stroke (NINDS), and the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

National Tay-Sachs and Allied Diseases Association Announces Competitive Grant Availability

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proposals that address one or more of the following: understanding disease progression and natural history, as well as unmet needs from a patient perspective across the severity spectrum of any given indication; development of patient registries; translational biomarkers with clinical utility; measurable and clinically meaningful efficacy endpoints for clinical trials; clinical research networks; and newborn screening.

Grants will be awarded for an initial period of one year at $40,000 direct costs per year (10% indirect cost rate); funding for a second year is predicated by adequate progress during year 1. The application

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Meet Our Patient Advocacy Groups

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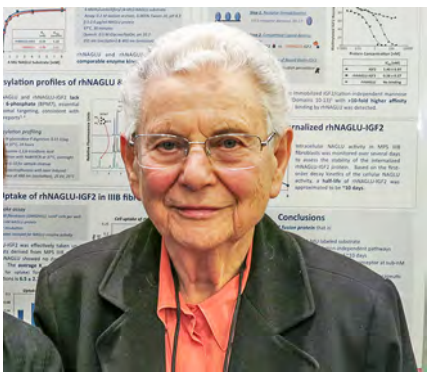
Mark Dant, co-founder and volunteer Executive Director of The Ryan Foundation

Executive Director of The Ryan Foundation, while also continuing to work to support his family in his profession as a law enforcement officer.

The Ryan Foundation's fundraising efforts began with a humble bake sale that netted $342. Following several years of progressively more successful initial fundraising efforts, a breakthrough occurred when the Foundation received $200,000.00 from an individual donor.

Eventually the Dants identified Emil Kakkis, M.D., Ph.D., at Harbor UCLA Research and Education Institute in Torrance, California, as a dynamic researcher who needed funding in order to make progress in his research towards enzyme replacement therapy (ERT) for MPS I. (Read the sidebar on page 4 to learn the chain of events that led the Dants to Dr. Kakkis.) Dr. Kakkis had been working with his mentor Elizabeth Neufeld, Ph.D., now Distinguished Professor Emerita, Biological Chemistry; Assistant Dean, Academic Affairs; and Emerita Member, Brain Research Institute at UCLA. (Visit Dr. Neufeld's UCLA Web page at: http://www.biolchem.ucla.edu/people/faculty/elizabeth-f-neufeld.) Dr. Neufeld had successfully achieved cloning of complementary DNA encoding α-L-iduronidase, the enzyme that is missing or deficient in MPS I patients. This led to the production of recombinant human α-L-iduronidase. The Ryan Foundation awarded Dr. Kakkis substantial research monies. This funding initially permitted Dr. Kakkis to hire key research and clinical personnel, including Merry Passage, M.S. and Barbara Lyons, R.N. These events are elucidated in a video entitled "15 Years of History, the MPS I Trial" posted on The Ryan Foundation Web site at: http://ryanfoundation.net/media/#videos.

Dr. Kakkis' work caught the attention of newly forming BioMarin Pharmaceutical company. Soon, a partnership was reached which allowed Dr. Kakkis to finish his research and get a promising new drug therapy into clinical trials. This combination of insightful leadership, industry support and dedicated work resulted, in December 1997, in the beginning of the first clinical trial of intravenous ERT for MPS I. This initial clinical trial involving 10 MPS I child patients included Ryan Dant as an intravenous ERT recipient. The 10 children came to Harbor UCLA Medical Center in Torrance,



Elizabeth Neufeld, Ph.D. at WORLD*Symposium* 2014 in San Diego, California

Photo by Victor Bloomfield

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Meet Our Patient Advocacy Groups

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California for weekly intravenous infusions of laronidase, which is the manufactured form of the enzyme α-L-iduronidase. The clinical trial period was for 52 weeks. The infused laronidase almost immediately started reversing the progression of MPS I symptoms for Ryan and other trial participants. Results were published in the New England Journal of Medicine. The free article is available online at: http://www.nejm.org/doi/full/10.1056/NEJM200101183440304#t= article.

Subsequently, in 2000, the FDA called for a second clinical trial, and 50 more MPS I children from all over North America and Europe repeated what the original ten had gone through in the previous two years. In February 2003 the Dants and several other families from both clinical trials traveled to Washington, D.C. and spoke at the FDA Hearing to approve the use of laronidase (commercially called Aldurazyme®), to treat MPS I. The panel voted ***unanimously*** to approve. Soon, children all over the world with MPS I began to improve their state of health with intravenous Aldurazyme® therapy.

Jeanne and Ryan Dant at a recent fundraising event. Ryan is now the longest-term Aldurazyme®-treated patient in the world.

The enzyme replacement therapy has relieved some, but not all, of the disease burden of MPS I for its recipients. Since the enzyme is unable to cross the blood-brain barrier due to its large molecular size, intravenous ERT has not alleviated the neuronopathic effects of MPS I. In response to this need, The Ryan Foundation has been funding research into intrathecal delivery of the enzyme. Intrathecal delivery is introduction of the replacement enzyme into the cerebrospinal fluid (CSF) of recipients, using injection into the CSF that surrounds the spinal cord. Since the spinal CSF is continuous with the brain's CSF, the enzyme then gradually circulates throughout the brain, carried by the CSF. The CSF circulation system is separate from brain's vascular system, and allows the enzyme to completely bypass the blood-brain barrier. For an overview of the blood-brain barrier, visit: http://www.brainfacts.org/brain-basics/neuroanatomy/articles/2014/blood-brain-barrier/.

Intrathecal ERT for MPS I is in clinical trial at the Los Angeles Biomedical Research Institute at Harbor UCLA Medical Center. This clinical trial, a 24-month study, is recruiting participants. (For in-depth information, visit: http://clinicaltrials.gov/show/NCT00852358). In addition to The Ryan Foundation, BioMarin Pharmaceutical company, and the LDN are also involved in supporting this research. Patricia Dickson, M.D. and Agnes Chen, M.D. are the key researchers leading this effort at Los Angeles Biomedical Research Institute. (See "LDN Research Matters" on page 5).

Barbara Wedehase, MSW, CGC, Executive Director of the National MPS Society (left) and Patricia Dickson, M.D., Chief, Medical Genetics, Department of Pediatrics, Harbor-UCLA Medical Center. The occasion was WORLD*Symposium* 2013 in Orlando, Florida. Photo by Victor Bloomfield.

Other important MPS I researchers who have received research funding from The Ryan Foundation include Mark Haskins, V.M.D., Ph.D. at the University of Pennsylvania; N. Matthew Ellinwood, D.V.M., Ph.D. at Iowa State University; Katherine Ponder, M.D. at Washington University School of Medicine in St. Louis, Missouri; Dr. Shiobahn Cashman at the University of Utah; Dr. Elizabeth Neufeld at UCLA; and Chester Whitley Ph.D., M.D. and Elsa Shapiro Ph.D. at the University of Minnesota.

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How Did the Dants Find the Right Researcher to Fund?

AWARENESS

Following Ryan Dant's diagnosis with MPS I, Mark Dant had been spending many, many hours reading everything he could on MPS in the public li- brary systems near their home in the Dallas area. Ryan was diagnosed long before the Internet came on the scene, so medical information and research could only be accessed hands-on in libraries. Mark noticed that a great deal of what he was reading was written by Dr. Elizabeth Neufeld at UCLA.

RISK-TAKING

In 1994, the National MPS Society was planning on having their Annual National Family Conference at Disney World in Orlando, but they didn't have the funding to bring MPS patients and families, as well as scientists and physicians, across the country for the Conference. The Ryan Foundation offered to fund the entire event. The Dants and their supporters got to work early that year, raising a substantial amount of money and giving it all to the National MPS Society, which ended up making the Conference possible that year.

NETWORKING

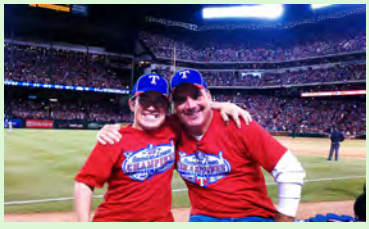
Dr. Elizabeth Neufeld presented at that Conference. The night she presented, Mark met with her at the hotel and asked her about a specific therapy that Mark had seen presented by Dr. Roscoe Brady the prior year at an international symposium about lysosomal diseases in Dusseldorf, Germany. (Jeanne Dant was working with American Airlines at the time, which afforded Mark the opportunity to fly standby worldwide for free.) Dr. Brady had just completed a clinical trial of a synthetic enzyme to treat Gaucher disease. He brought a young trial participant to the podium as a living example of how well ERT worked in the Gaucher model. Mark stopped Dr. Brady in the hotel hallway after his presentation, and showed him a video (thru a video camera viewfinder) of Ryan playing baseball. In the video, Ryan was four years old. Dr. Brady told Mark that ERT was possible in MPS I, but that Mark didn't have two things that were needed for Ryan. He said Mark didn't have enough money, because to develop a drug requires substantial amounts of it. He also said Mark didn't have enough time, since Ryan was already four, and it would take years to develop the enzyme. What Mark heard the best during that conversation was: it was possible to do ERT in MPS I.

When Mark asked Dr. Neufeld about ERT for MPS I, she replied that not only is it possible, she knew of a brilliant former student of hers who was struggling to develop enough MPS I enzyme in his lab at Harbor UCLA because of lack of funds. His name was Dr. Emil Kakkis.

BOLDNESS

The Dants called Dr. Kakkis the next day and pledged every dollar they could raise to his research. Mark recalls, "His ability and drive is what made it possible. There are few like him in the world, and that conversation with Dr. Neufeld changed everything for us because of Dr. Kakkis' will to treat those with rare disease."

REWARD



Ryan and Mark Dant at the 2010 World Series, Game 3, at The Ballpark in Arlington: Texas Rangers vs. San Francisco Giants.

LDN Research *Matters*

An Extension Study of Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I

By Evelyn S. Redtree, M.S. and Agnes H. Chen, M.D.

"An Extension Study of Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I" is the continuation of LDN Pilot Study # 6714. Agnes H. Chen, M.D. is the Principal Investigator of this extension study during the new five-year funding cycle.

Mucopolysaccharidosis I (MPS I) is a lysosomal disease caused by deficiency of α-L-iduronidase. Recombinant human α-L-iduronidase (rhIDU) is available as laronidase (Aldurazyme®) for intravenous delivery to treat physical disease due to MPS I (i.e., intravenous enzyme replacement therapy, or ERT). While partially effective for physical disease, intravenous rhIDU does not treat neurological symptoms of MPS I, because the blood-brain barrier prevents the bulk of the administered protein from entering the central nervous system (CNS).

Patients with MPS I suffer from progressive neurological disease, and this symptom may be even more prevalent with increasing lifespan due to treatment with intravenous rhIDU. In the most severe form of MPS I, children evidence a slowing in cognitive development, and fall into the range of mental retardation as defined by standard intelligence tests by age 3 years (Shapiro et al. 1995). Although patients with milder forms of MPS I may not have grossly observable problems with cognition, these patients do have learning difficulties that are apparent in school and with neuropsychological testing. Many of these patients are evidencing dementia in their teens or early adulthood.

Studies in hematopoietic stem-cell-transplanted MPS I patients have shown that central nervous system (CNS) improvement typically spans several years. The fact that cognitive outcomes may take years to show up on testing, coupled with the fact that long-term studies are not typically performed in drug development until the *post-marketing* phase, leads to a "Catch-22" in CNS drug development. In other words, companies cannot invest time and money in a pivotal study that will take several years; yet such a time-frame may be needed to see statistically-significant efficacy on CNS disease.

Brain-directed therapy such as intrathecal rhIDU is a promising approach for addressing the key remaining challenge in the treatment of MPS I: neurological disease. Determination of cognitive outcomes may require long-term follow-up of treated patients. This extension study will address this need by providing five years of treatment for MPS I patients, permitting the best chance of detecting treatment effects. In addition, long-term safety data has been completely lacking for *any* intrathecally-administered enzyme, and this study will also address that need.

The primary study site is Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center in Torrance, California. Other sites that are part of this study include the University of Minnesota and Children's Hospital Oakland Research Institute (CHORI). All of the subjects will have neuropsychological testing performed at the University of Minnesota Center for Neurobehavioral Development. Most of the subjects will have brain magnetic resonance imaging at the University of Minnesota Center for Magnetic Resonance Research. A small minority of subjects, those with ventriculoperitoneal shunts who need neurosurgical assistance for shunt reprogramming after MRI, will have their brain MRI performed at the Children's Hospital of Oakland Research Institute (which is part of the neuroimaging core for the LDN). This is due to the availability of a neurosurgeon who can reprogram the shunt after the MRI is completed. In addition, subjects who need sedation to perform the MRI will also have the study done at CHORI because of the availability of pediatric anesthesia. So that these subjects will not have to travel to three different sites for the study, subjects who need imaging at CHORI will also be consented there, and have study assessments and treatments performed there.

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LDN Research *Matters*

An Extension Study of Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I

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The safety outcomes of this study are treatment-related adverse events, and the presence and effect of an immune response to the administered rhIDU. The efficacy outcomes of this study include changes noted in the results of a neuropsychological test battery that includes tests of cognition, memory, visual-spatial ability, attention and concentration. Efficacy outcomes also include changes noted in data obtained via advanced MRI techniques that examine brain volume, white matter lesions, and connectivity; by clinical examination; and by measuring the biomarker glycosaminoglycans in patients' cerebrospinal fluid.

In summary, this study is investigating whether injecting rhIDU into the CSF of MPS I patients will treat symptoms of cognitive decline such as memory loss, language problems and learning difficulties. This is the first long-term study of this treatment approach in MPS I, and will demonstrate whether the intrathecal approach is safe and effective.

Reference Cited:

E.G. Shapiro, L.A. Lockman, M. Balthazor, W. Krivit, Neuropsychological outcomes of several storage diseases with and without bone marrow transplantation. J Inherit Metab Dis 18(4) (1995) 413-429. PMID: 7494400.

Publications arising from the LDN Pilot Study #6714, the predecessor of this continuation study:

Peer-Reviewed Journal Articles:

P.I. Dickson, A.R. Pariser, S.C. Groft, R.W. Ishihara, D.E. McNeil, D. Tagle, D.J. Griebel, S.G. Kaler, J.W. Mink, E.G. Shapiro, K.J. Bjoraker, L. Krivitzky, J.M. Provenzale, A. Gropman, P. Orchard, G. Raymond, B.H. Cohen, R.D. Steiner, S.F. Goldkind, R.M. Nelson, E. Kakkis, M.C. Patterson, Research challenges in central nervous system manifestations of inborn errors of metabolism. Mol Genet Metab 102(3) (2011) 326-338. PMCID: PMC3040279. Article available freely online at: http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3040279/.

P.I. Dickson, A.H. Chen, Intrathecal enzyme replacement therapy for mucopolysaccharidosis I: translating success in animal models to patients. Curr Pharm Biotechnol 12(6) (2011) 946-955. PMID: 21506913.

M. Vera, S. Le, S.H. Kan, H. Garban, D. Naylor, A. Mlikotic, I. Kaitila, P. Harmatz, A.H. Chen, P.I. Dickson, Immune response to intrathecal enzyme replacement therapy in mucopolysaccharidosis I patients. Pediatr Res 74(6) (2013) 712-720. PMCID: PMC3855632. Article available freely online at: http://www.ncbi.nlm.nih.gov/pmc/articles/PMC3855632/.

Published Abstracts:

A.H. Chen, P.I. Dickson, E.G. Shapiro, B. Lyons, S.H. Kan, D. Guillaume, A study of intrathecal enzyme replacement for cognitive decline in mucopolysaccharidosis I. Mol Genet Metab 99(2) (2010) S13-14.

A.H. Chen, P.I. Dickson, E.G. Shapiro, L. Rovai, B. Lyons, S.H. Kan, A. Victoroff, A study of intrathecal enzyme replacement for cognitive decline in mucopolysaccharidosis I. Mol Genet Metab 102(2) (2011) S10-11.

A.H. Chen, P.I. Dickson, E.G. Shapiro, L. Rovai, S.H. Kan, A study of intrathecal enzyme replacement for cognitive decline in mucopolysaccharidosis I. Mol Genet Metab 108(2) (2013) S29.

Book Chapter:

A.H. Chen, P.I. Dickson, Enzyme replacement therapy for cognitive decline in MPS I: past, present and future, in: Current Medical Literature - Lysosomal Storage Diseases, Volume 9, Royal Society of Medicine (Great Britain), London (2011) 9-16.

Meet the Principal Investigators

Agnes H. Chen, M.D.

Agnes H. Chen, M.D. is a pediatric neurologist at the Los Angeles County - Harbor-UCLA Medical Center, where she trained in general pediatrics and adult and child neurology. During training, she began working with Dr. Patricia Dickson on intrathecal enzyme replacement for mucopolysaccharidosis type I (MPS I).

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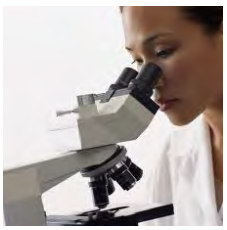
will use a two-step process. First, a one page pre-application will be submitted. From these pre-applications, investigators will be chosen to submit a full application. The application format and review criteria are downloadable as a .pdf document at: http://www.ntsad.org/index.php/resources/library/research/251-ntsad-2014-2015-request-for-proposal-guidelines/file. The **deadline for submission** of pre-applications is 5:00 p.m. (EST) on Friday **January 2, 2015**.

The *pre-applications* will be reviewed by sub-committees of NTSAD's Scientific Advisory Committee and Corporate Advisory Council. Selected applications will be invited to submit full applications for further peer review and funding consideration. Grant awards are based on proposal evaluation by a Research Evaluation Subcommittee of NTSAD's Scientific Advisory Committee. The grant awards will be announced in April 2015. The funding period begins approximately on July 1, 2015, pending the Institution's agreement to NTSAD's Research Grant Policies. These policies include intellectual property clauses and related terms to protect the interests of families. The policies also outline the system for making the grant payments in installments, following approved research-progress reports. This process ensures that the highest quality science, and projects with the most meaningful outcomes are awarded. In addition, the process holds the researchers accountable for following through appropriately with their proposed project.



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Your Submissions Welcome!

Meet Our Patient Advocacy Groups

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The critical importance of The Ryan Foundation's dedicated fundraising efforts and search for its researcher-beneficiaries is made clear by the results obtained. The Dants have not concerned themselves with needless duplication of the services provided by the National MPS Society (see "**Indications**" April 2014). Rather, they have maintained their narrow focus on facilitating groundbreaking research into effective treatment and a cure. They are providing global leadership by showing how private individuals can effectively circumvent the slow progress in rare diseases' treatments – slow progress that results, in part, from the traditional research-funding paradigm. The traditional model of research funds flowing from government agency, to the researcher's associated university or other institution, and to researcher, is obviously correct when the government is the funder. The Ryan Foundation has shown there is another path that is equally valid and can be fruitful. For this the rare disease community, not just the MPS disease community, owes The Ryan Foundation a huge debt of gratitude.

Meet the Principal Investigators

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She investigated glycosaminoglycan storage in different brain regions of the MPS I dog, and evaluated patients that were participating in the first clinical trial of intrathecal enzyme replacement for MPS I. That experience during her training led to her interest in the central nervous system manifestations of lysosomal storage diseases and research into potential treatments. In 2007, Dr. Chen became Assistant Professor of Pediatrics and Neurology, and continued her close collaboration with Dr. Dickson. During the LDN's first

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Meet the Principal Investigators

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NIH funding cycle, Dr. Chen was Co-PI of LDN Pilot Study #6714, "A Study of Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I" working with Dr. Patricia Dickson as the PI.

Agnes H. Chen, M.D.

Dr. Chen is currently Principal Investigator of the continuation study of LDN pilot study #6714, entitled "An Extension Study of Intrathecal Enzyme Replacement for Cognitive Decline in Mucopolysaccharidosis I" (see page 5). She is also Associate Professor, Department of Pediatrics, David Geffen School of Medicine at UCLA. She continues her busy clinical practice in general pediatric neurology and has research activities in other lysosomal diseases, including mucopolysaccharidosis type IIIA (Sanfilippo Syndrome type A), Niemann-Pick disease type C, and metachromatic leukodsytrophy. For more about Dr. Chen, visit: http://harborpeds.org/academics/faculty/agneschen. A list of Dr. Chen's selected publications can be found there.

Genzyme Announces the 2014 Genzyme PAL Award Winners

On October 1, 2014 Genzyme, a Sanofi Company, announced the 2014 winners of its Patient Advocacy Leadership (PAL) Awards program. There were 45 applicants, and proposals were received from patient organizations in 23 countries. Proposals were received from the Krabbe, Gaucher, MPS, MLD, Fabry, Pompe, Tay-Sachs, Sandhoff, and Niemann-Pick disease communities, as well as several rare disease patient organizations that represent the lysosomal disease community. Nine patient organizations were selected by the External Review Committee to receive a PAL Award. The successful applicants came from Australia, Brazil, Croatia, Japan, the Netherlands, Peru, the United Kingdom, the United States, and a joint proposal from Canada and the United States.

One of the PAL Award winners is the Fabry Support and Information Group (FSIG), whose PAL project is the production of a video. This video will be designed in such way as to allow it to be adapted by different patient organizations to educate across various lysosomal diseases and diverse cultures around the world. The purpose of the video is to teach the underlying mechanisms of lysosomal diseases in an easily understandable, entertaining format. (For more information about the FSIG, read the February 2014 issue of "**Indications**," available via hyperlink on the upper-right area of the LDN's homepage at: http://www.lysosomaldiseasenetwork.org/.)

For the complete list of 2014 PAL Award winners, visit: http://www.genzymeadvocacyawards.com/award-winners.aspx.

Calling all Rare Diseases Research Manuscripts

The online *Journal of Rare Disorders* (www.journalofraredisorders.com) has issued a call for manuscripts. To wit, "The Journal of Rare Disorders invites you to submit original research, open-label studies, review articles, regulatory issues, case reports, and editorials. We are also planning **a special focus issue on lysosomal storage disorders** for 2015. There is never a fee to publish and all articles are available online without charge." All articles are chosen on the basis of scientific rigor. The deadline for submission of articles for the special lysosomal diseases issue is March 30, 2015. The *Journal of Rare Disorders* also welcomes announcements and news items of importance to the rare diseases community. Please submit your press releases and announcements to Cindy Jablonowski, Publisher: cindy@scicomgroup.com. The new Editor-in-Chief of the *Journal of Rare Disorders* is James Cloyd, PharmD, the Administrative Director of the Lysosomal Disease Network. The guidelines for authors are available at: http://www.journalofraredisorders.com/ForAuthors.htm.

Calendar of Upcoming Events

National MPS Society Annual National Family Conference, December 18 - 21, 2014 at Disney's Contemporary Resort in Orlando, FL, USA. Visit: https://mpssociety.org/event-registration/?regevent_action=register&event_id=147

Lysosomal Disease Network's 11th Annual WORLD*Symposium*™, February 9 - 12, 2015 at the Hyatt Regency Orlando Hotel in Orlando, Florida. Visit: www.worldsymposia.org

Fabry Support & Information Group's 2015 FSIG Expert Fabry Conference, February 13 - 15, 2015 in Orlando, Florida. Details will become available as they are finalized. Visit: http://www.fabry.org/FSIG.nsf/Pages2/HomePage

2015 American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting, March 24 - 28, 2015 at the Salt Palace Convention Center, and the Salt Lake Marriott Downtown at City Creek, and the Hilton Salt Lake Downtown Hotel in Salt Lake City, Utah. Exhibit dates: March 25 - 27, 2015. Visit: http://ww4.aievolution.com/acm1501/

National Tay-Sachs and Allied Diseases 37th Annual Family Conference, April 16 - 19th, 2015 at Hyatt Regency Reston, 1800 Presidents Street, Reston, Virginia. Visit: http://www.ntsad.org/index.php/event-listings/family-conference/2015-conference

American Society of Gene & Cell Therapy 18th Annual Meeting, May 13 - 16, 2015, in New Orleans, Louisiana. ASGCT homepage: http://www.asgct.org/

The Calliope Joy Foundation's *An Evening with Jim & Jill Kelly for Hunter's Hope* to benefit a National Care Network for Leukoydstrophies, Friday, May 15, 2015 at the Rittenhouse Hotel, Philadelphia, PA. VIP Reception starts 6:00 p.m.; Dinner 7:00 - 10:00 p.m. Event Chair, Maria Kefalas: mkefalas67@gmail.com. Visit: http://www.thecalliopejoyfoundation.org/

The annual "Michael, Marcia & Christa Parseghian Scientific Conference" for Niemann-Pick type C disease research will be held June 11 -13, 2015 at the University of Notre Dame. Researchers will gather for three days to discuss the advances in NP-C research. This yearly meeting helps to form collaborations and determine the future direction of NP-C research. Visit: http://www.parseghian.org/events.html or call: (520) 577-5106.

Batten Disease Support and Research Association Family Conference 2015, July 9 - 12, 2015 at The Eaglewood Resort and Spa, 1401 Nordic Road, Itasca, Illinois. Visit: www.eaglewoodresort.com, and http://bdsra.org/

United Leukodystrophy Foundation's 2015 Annual Scientific Meeting and Family Conference, July 15 - 18, 2015 at Embassy Suites Downtown Old Market in Omaha, Nebraska. Visit: http://ulf.org/conferences

Cystinosis Research Network 2015 Family Conference, July 16 - 18, 2015 at Doubletree by Hilton Chicago Magnificent Mile, Chicago, IL. Visit: https://cystinosis.org/

Glycoproteinoses: Fourth International Conference on Advances in Pathogenesis and Therapy (a combined scientific and family conference), July 23 - 26th, 2015, in St. Louis, Missouri, USA. Visit: www.ISMRD.org

The Association for Glycogen Storage Disease 2015 Family/Professional Conference, September 18 - 19, 2015 in Oklahoma City, OK. For updates, visit: http://www.agsdus .org/

Disappointed that your conference or meeting is not listed here? The remedy is easy:

LDNed@umn.edu

Bright Ideas

There is an approximately 56-minute video of a July 30, 2014 public meeting of the **Rare Disease Congressional Caucus** available online at: http://jonahsjustbegun.org/#prettyPhoto. In the video, **Dr. Emil Kakkis** begins speaking at 31:40.

Indications™

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