Monday, February 4, 2019

9:00 – 12:00	**Council of Patient Advocates (COPA) Workshop: WORLD*Fair*™**	The Lysosomal Disease Network (LDN) Annual Council of Patient Advocates (COPA) Meeting
1:00 – 5:00	**Emerging Trends: State-of-the-Art for Experts** *(Registration required)*	
1:00 PM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Introduction and Overview of Course
1:10 PM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Lysosomal Disease Phenotypes
1:35 PM	**Steven U. Walkley** Albert Einstein College of Medicine New York, NY, United States	Normal Lysosomal Function
2:00 PM	**Break**	
2:05 PM	**Marc C. Patterson** Mayo Clinic Rochester, MN, United States	Remarkable Cases
2:30 PM	**Steven U. Walkley** Albert Einstein College of Medicine New York, NY, United States	Lysosomal Disease Pathogenesis
2:55 PM	**Refreshment Break**	
3:10 PM	**Jeanine R. Jarnes** University of Minnesota Minneapolis, MN, United States	Current Treatments for Lysosomal Diseases
3:35 PM	**Amy Gaviglio** Minnesota Department of Health St. Paul, MN, United States	Newborn Screening
4:00 PM	**Break**	
4:05 PM	**Jeanine R. Jarnes** University of Minnesota Minneapolis, MN, United States	Future Treatments for Lysosomal Diseases
4:30 PM	**Marc C. Patterson** Mayo Clinic Rochester, MN, United States	Remarkable Cases
5:00 PM	**Adjourn**	
5:15 PM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Young Investigator Awards
5:30 PM	**Opening Reception**	Exhibit Hall
6:30 PM	**CME Satellite Symposia**	

Tuesday, February 5, 2019

6:15 AM	**Satellite Symposia**	

Basic Science I — Co-Chairs: Danilo A. Tagle & Cynthia J. Tifft

Disease Mechanisms, Pathology and Biomarkers

7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	**Welcome & Announcements** **Presentation of 2019 Roscoe O. Brady Award for Innovation and Accomplishment to Danilo A. Tagle**
7:45 AM	**Danilo A. Tagle** National Center for Advancing Translational Sciences National Institutes of Health Bethesda, MD, United States	Innovations in Rare Disease Research
8:15 AM	**Eric Joshua Garcia** National Institutes of Health Bethesda, MD, United States	Methylomic and whole transcriptome analyses reveal several potential modifier genes in GBA1-associated Parkinson disease
8:30 AM	**Vera Niederkofler** QPS Austria GmbH Grambach, Austria	CBE treatment of alpha-synuclein over-expressing and wildtype mice models Gaucher disease pathology
8:45 AM	**Annalisa Astolfi** University of Bologna Bologna, Italy	Hippo and necroptosis pathways are involved in cell growth defects in Gaucher disease
9:00 AM	**Pilar Giraldo** Institute of Health Research Aragón (IIS Aragón) Zaragoza, Spain	Strain-elastography in musculoskeletal evaluation in Gaucher disease
9:15 AM	**Amelia Ahern-Rindell** University of Portland Portland, OR, United States	Design and analysis of a CRISPR gene editing strategy in a sheep model variant of GM1-gangliosidosis
9:30 AM	**Anatalia Labilloy** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Proteomic profiling of engineered human immortalized podocyte cell model of Fabry disease
9:45 AM	**Break & Exhibits**	
10:15 AM	**Behzad Najafian** University of Washington Seattle, WA, United States	A novel method for quantification of globotriaocylceramide (GL-3) inclusions in affected podocytes in females with Fabry disease shows progressive accumulation of GL-3 in podocytes with age and no cross-correction between affected and non-affected podocytes
10:30 AM	**Jin-Song Shen** Baylor Research Institute Dallas, TX, United States	Dysregulated DNA methylation in the pathogenesis of Fabry disease

10:45 AM	**Siamak Jabbarzadeh-Tabrizi** Baylor Research Institute Dallas, TX, United States	Effects of genetic background on disease phenotypes in a mouse model of Fabry disease
11:00 AM	**Pasqualina Colella** Généthon, Université Evry, Université Paris Saclay Evry, France	Latent TGF-beta-binding protein *4* modulates disease severity in the knock-out mouse model of Pompe disease
11:15 AM	**Katie Harvey** Great Ormond Street Hospital London, United Kingdom	The evolving role of enzymology and metabolomics in the diagnosis of lysosomal disorders in the post genomic era
11:30 AM	**Lunch - on own or satellite symposia**	Exhibit hall is open
11:45 AM	**Satellite Symposia**	

Basic Science II — Co-Chairs: David A. Pearce & Tiina K. Urv

Developing Therapeutic Approaches in the Laboratory

1:00 PM	**Virginia Kimonis** University of California - Irvine Orange, CA, United States	Antisense oligonucleotide treatment targeting glycogen synthase (GYS1) in a mouse model of Pompe disease
1:15 PM	**Kohji Itoh** Tokushima University Tokushima, Japan	*In vivo* gene therapy for Tay-Sachs and Sandhoff diseases by utilizing AAV9 vector encoding modified *HEXB*
1:30 PM	**Kazuki Sawamoto** Nemours/Alfred I. duPont Hospital for Children Wilmington, DE, United States	Development of AAV gene therapy for Morquio syndrome type A
1:45 PM	**Saida Ortolano** Instituto de Investigación Sanitaria Galicia Sur Vigo (Pontevedra), Spain	Functional evaluation of an AAV9 based vector expressing alpha-galactosidase A for potential gene therapy of Fabry disease
2:00 PM	**Xin Chen** University of Texas Southwest Medical Center Dallas, TX, United States	Therapeutic efficacy and safety of scAAV9/AGA gene therapy in aspartylglucosaminuria mice
2:15 PM	**Murtaza S. Nagree** University of Toronto Toronto, ON, Canada	Lentiviral-modified T Rapa cells as 'micropharmacies' for lysosomal diseases
2:30 PM	**Hojun Choi** Korea Advanced Institute of Science and Technology (KAIST) Daejeon, Korea	Exosome-mediated delivery of active glucocerebrosidase to Gaucher model cells
2:45 PM	**Break & Exhibits**	
3:15 PM	**Ibane Abasolo** Vall d'Hebron Institute of Research Barcelona, Spain	Targeted nanoliposomes for the treatment of Fabry disease
3:30 PM	**Cristin Davidson** National Institutes of Health Bethesda, MD, United States	Improved disease amelioration with combination therapy for Niemann-Pick type C1 disease

3:45 PM	**Yanyan Peng** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Intravenous infusion of iPSC-derived neural progenitors expressing GCase ameliorates alpha-synuclein aggregates in a mouse model of Gaucher disease
4:00 PM	**Jeanine R. Jarnes** University of Minnesota Minneapolis, MN, United States	Chitotriosidase as a biomarker for central nervous system inflammation in the gangliosidosis diseases
4:15 PM	**Chloe L. Christensen** University of Victoria Victoria, BC, Canada	Delivery of adenine base editors to patient-derived induced pluripotent stem cells *in vitro*: A putative treatment for mucopolysaccharidosis type IIIB
4:30 PM	**Poster Reception in Exhibit Hall**	
6:30 PM	**Satellite Symposia**	

Wednesday, February 6, 2019

6:15 AM	**Satellite Symposia**	

Translational Research IA — Co-Chairs: R. Scott McIvor & Mark S. Sands

Gene Therapy

7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	**2019 Patient Advocate Leader Announcement and Presentation to Mark Dant**
7:45 AM	**Steven J. Gray** University of Texas Southwestern Medical Center Dallas, TX, United States	Intrathecal and intravenous combination gene therapy in the mouse model of infantile neuronal ceroid lipofuscinosis extends lifespan and improves behavioral outcomes in moderately affected mice
8:00 AM	**Raymond Y. Wang** Children's Hospital of Orange County Children's Specialists Orange, CA, United States	Intra-articular AAV9 α-iduronidase gene therapy in the canine model of mucopolysaccharidosis type I results in rapid synovial and cartilage iduronidase expression, clearance of heparan sulfate, and high serum α-iduronidase levels
8:15 AM	**Roselena S. Schuh** Universidade Federal do Rio Grande do Sul Porto Alegre, Brazil	Newborn genome editing improves phenotype, cardiovascular, respiratory, and bone disease in mucopolysaccharidosis type I mice
8:30 AM	**Brian Bigger** University of Manchester Manchester, United Kingdom	Brain targeted stem cell gene therapy provides long-term correction of mucopolysaccharidosis type II
8:45 AM	**Giuseppe Ronzitti** Généthon, Université of Evry, Université Paris-Saclay Evry, France	Safety and efficacy evaluation of investigational liver gene transfer for secretable GAA in the treatment of Pompe disease
9:00 AM	**Jeffrey A. Medin** Medical College of Wisconsin Milwaukee, WI, United States	FACTs Fabry gene therapy clinical trial: Two year data
9:15 AM	**Kevin M. Flanigan** Center for Gene Therapy, Nationwide Children's Hospital Columbus, OH, United States	Phase 1/2 clinical trial of systemic gene transfer of scAAV9.U1a.hSGSH for MPS IIIA demonstrates 2 years of safety, tolerability, and biopotency
9:30 AM	**Cassie Bebout** Auburn University Auburn, AL, United States	Analysis of the effect of intravenous gene therapy on brain and peripheral disease in a feline model of GM1-gangliosidosis
9:45 AM	**Break & Exhibits**	

Translational Research IB — Co-Chairs: Amy Gaviglio & Priya Kishnani

Implementation and Impact of Newborn Screening

10:15 AM	**Chia-Feng Yang** Taipei Veterans General Hospital Taipei City, Taiwan	Very early treatment for infantile-onset Pompe disease contributes to better outcomes: 10-year experience of nationwide NBS in Taiwan
10:30 AM	**Barbara K. Burton** Ann & Robert H. Lurie Children's Hospital	Newborn screening for mucopolysaccharidosis type II (MPS II) in Illinois: The first year's experience

	Chicago, IL, United States	
10:45 AM	**Elizabeth Braunlin** University of Minnesota Minneapolis, MN, United States	Hematopoietic cell transplantation for severe MPS I in the first six months of life: The heart of the matter
11:00 AM	**Adam Guenzel** Mayo Clinic Rochester, MN, United States	Improved differentiation between Krabbe disease variants, carrier status, and pseudo deficiency by measurement of psychosine
11:15 AM	**Francisco J. del Castillo** Hospital Universitario Ramón y Cajal, IRYCIS Madrid, Spain	NGS-based, 107-gene resequencing panel as first-line screening test for lysosomal diseases
11:30 AM	**Lunch - on own or satellite symposia**	Exhibit Hall is open
11:45 AM	**Satellite Symposia**	

Translational Research II — Co-Chairs: Lalitha R. Belur & Philip J. Brooks

Clinical Trial Readiness: Pre-Clinical Trial Methods and Studies

1:00 PM	**Laura Adang** Children's Hospital of Philadelphia Philadelphia, PA, United States	Clinical presentation of metachromatic leukodystrophy
1:15 PM	**Cara O'Neill** Cure Sanfilippo Foundation Columbia, SC, United States	The natural history of facial features observed in Sanfilippo syndrome (MPS IIIB) using a next generation phenotyping tool
1:30 PM	**Rebecca Ahrens-Nicklas** The Children's Hospital of Philadelphia Philadelphia, PA, United States	A natural history study of multiple sulfatase deficiency
1:45 PM	**Lynda E. Polgreen** Los Angeles Biomedical Research Institute at Harbor-UCLA Torrance, CA, United States	Exploring surrogate biomarkers of skeletal and joint disease progression in mucopolysaccharidosis type I
2:00 PM	**Shunji Tomatsu** Nemours/Alfred I. duPont Hospital for Children Wilmington, DE, United States	Effect of enzyme replacement therapy on the growth of patients with Morquio syndrome type A
2:15 PM	**Ankit K. Desai** Duke University Durham, NC, United States	Changing the clinical course of infantile Pompe disease with immune modulation strategies: 12 years of experience
2:30 PM	**Simon Heales** Great Ormond Street Hospital/UCL London, United Kingdom	Urinary glucose tetrasaccharide, a useful prognostic biomarker for Pompe disease?
2:45 PM	**Break & Exhibits**	
3:15 PM	**Quoc-Hung Nguyen** University of California – San Francisco San Francisco, CA, United States.	Fetal enzyme replacement and stem cell transplantation in murine Sly syndrome targeting microglia
3:30 PM	**Igor Nestrasil** University of Minnesota Minneapolis, MN, United States	Discovery of brain MRI signatures in infants with severe form of MPS I in the pre-HSCT and post-HSCT stages
3:45 PM	**Adeline Vanderver** Children's Hospital of Philadelphia Philadelphia, PA, United States	Intrathecally administered recombinant human arylsulfatase A in patients with late-infantile metachromatic leukodystrophy: Phase 2b clinical trial design

4:00 PM	**Reena V. Kartha** University of Minnesota Minneapolis, MN, United States	Preliminary N-acetylcysteine results for LDN 6722, Role of oxidative stress and inflammation in Gaucher disease type 1: Potential use of antioxidant anti-inflammatory medications
4:15 PM	**Eric K.W. Hui** ArmaGen Inc. Calabasas, CA, United States	Preclinical studies of a brain penetrating IgG trojan horse-arylsulfatase fusion protein in the metachromatic leukodystrophy mouse
4:30 PM	**Poster Reception in Exhibit Hall**	
6:30 PM	**Satellite Symposia**	

Thursday, February 7, 2019

6:15 AM	**Satellite Symposia**

Clinical Trials I — Co-Chairs: Stephen C. Groft & Anne R. Pariser

Clinical Trials for Registration

7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	*Keynote Address*: The 'new' Lysosomal Disease Network
8:00 AM	**Raphael Schiffmann** Baylor Research Institute Dallas, TX, United States	Venglustat in adult Gaucher disease type 3: Preliminary safety, pharmacology, and exploratory efficacy from a phase 2 trial in combination with imiglucerase (LEAP)
8:15 AM	**Ronald G. Crystal** Weill Cornell Medicine New York, NY, United States	Design and rationale of the LYS-SAF302 gene therapy study in mucopolysaccharidosis type IIIA (MPS IIIA) children
8:30 AM	**Myrl D. Holida** University of Iowa Hospitals and Clinics Iowa City, IA, United States	Once every 4 weeks - 2 mg/kg of pegunigalsidase alfa for treating Fabry disease; Preliminary results of a phase 3 study
8:45 AM	**Ulla Feldt-Rasmussen** Rigshospitalet, Copenhagen University Hospital Copenhagen, Denmark	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 30-month results from the randomized phase 3 ATTRACT study
9:00 AM	**Priya S. Kishnani** Duke University Durham, NC, United States	Safety and efficacy of VAL-1221, a novel fusion protein targeting cytoplasmic glycogen, in patients with late-onset Pompe disease
9:15 AM	**Paula R. Clemens** University of Pittsburgh and Department of Veterans Affairs Medical Center Pittsburgh, PA, United States	Efficacy and safety of AT-GAA (ATB200/AT2221) in ERT-switch nonambulatory patients with Pompe disease: Preliminary results from the ATB200-02 trial
9:30 AM	**Loren Pena** University of Cincinnati College of Medicine Cincinnati, OH, United States	NEO1 and NEO-EXT studies: Long-term safety of repeat avalglucosidase alfa dosing for 4.5 years in late-onset Pompe disease patients
9:45 AM	**Refreshment Break**	
10:15 AM	**Nathalie Guffon** Hôpital Femme Mère Enfant Lyon, France	The first study investigating safety and efficacy of velmanase alfa (human recombinant alpha mannosidase) in alpha-mannosidosis patients below six years of age
10:30 AM	**Kara Woolgar** Phoenix Children's Hospital Phoenix, AZ, United States	Intravenous 2-hydroxypropyl-beta-cyclodextrin for a Niemann-Pick disease type C1 infant with liver cirrhosis
10:45 AM	**Paul J. Orchard** University of Minnesota Minneapolis, MN, United States	Preliminary results demonstrate engraftment with minimal neutropenia with MGTA-456, a $CD34^+$ expanded cord blood (CB) product in patients transplanted for inherited metabolic disorders (IMD)

11:00 AM	**Joseph Muenzer** University of North Carolina, Chapel Hill Chapel Hill, NC, United States	CHAMPIONS: A phase 1/2 clinical trial with dose escalation of SB-913 ZFN-mediated in vivo human genome editing for treatment of MPS II (Hunter syndrome)
11:15 AM	**Paul Harmatz** UCSF Benioff Children's Hospital Oakland Oakland, CA, United States	EMPOWERS: A phase 1/2 clinical trial of SB-318 ZFN-mediated in vivo human genome editing for treatment of MPS I (Hurler syndrome)
11:30 AM	**Lunch - on own or satellite symposia**	
11:45 AM	**Satellite Symposia**	

Clinical Trials II — Co-Chairs: Yoshikatsu Eto, Jill Morris & Marc C. Patterson

Clinical Outcomes

1:00 PM	**Maureen Cleary** Great Ormond Street Hospital London, United Kingdom	ICV-administered tralesinidase alfa (BMN 250; NAGLU-IGF2) is well-tolerated and reduces heparan sulfate accumulation in the CNS of subjects with Sanfilippo syndrome type B (MPS IIIB)
1:15 PM	**Maria L. Escolar** Children's Hospital of Pittsburgh Pittsburgh, PA, United States	Long-term neurodevelopmental outcomes of hematopoietic stem cell transplantation for late-infantile Krabbe disease
1:30 PM	**Karolina M. Stepien** Salford Royal NHS Foundation Trust Salford, United Kingdom	Hormonal dysfunction in adult patients with mucopolysaccharidosis type I post haematopoietic stem cell transplantation
1:45 PM	**Ashish Gupta** University of Minnesota Minneapolis, MN, United States	Allogeneic hematopoietic stem cell transplant improves outcomes in fucosidosis
2:00 PM	**Mark Roberts** Salford Royal NHS Foundation Trust Salford, United Kingdom	Preliminary patient-reported outcomes and safety of AT-GAA (ATB200/AT2221) in patients with Pompe disease from the ATB200-02 trial
2:15 PM	**Edwin Chavez-Cintora** BioMarin Pharmaceutical Inc. Novato, CA, United States	Insights into Sanfilippo syndrome provided by the ConnectMPS worldwide online registry
2:30 PM	**Troy Lund** University of Minnesota Minneapolis, MN, United States	Predicting intelligence in MPS IH with biomarkers
2:45 PM	**Refreshment Break**	
3:15 PM	**Brianna Glase** National Institutes of Health Bethesda, MD, United States	Robust clinical outcome measures for patients with juvenile onset GM1-gangliosidosis
3:30 PM	**Angela Schulz** University Medical Center Hamburg-Eppendorf Hamburg, Germany	Persistent treatment effect of cerliponase alfa in children with CLN2 disease: A 3 year update from an ongoing multicenter extension study

3:45 PM	**Tama Dinur** Shaare Zedek Medical Center, Hadassah Medical Center, The Hebrew University Jerusalem, Israel	Long-term follow-up of 103 untreated adult patients with type 1 Gaucher disease
4:00 PM	**Uma Ramaswami** Royal Free London NHS Foundation Trust University College London London, United Kingdom	Migalastat: Single centre experience of adult patients with Fabry disease from the Royal Free London NHS Foundation Trust, UK
4:15 PM	**Christian J. Hendriksz** Steve Biko Academic Hospital Pretoria, South Africa	Evidence-based, expert-agreed recommendations for the management of patients with MPS IVA/VI: Recommendations to replace the specific missing enzyme
4:30 PM	**Adjourn**	
4:30 PM	**Networking Reception in Foyer**	
5:30 PM	**Council of Research Experts (CORE) Meeting**	The Lysosomal Disease Network (LDN) annual Council of Research Experts (CORE) meeting for NIH-funded investigators