Monday, February 10, 2020

8:00 AM – 11:30 AM	**Emerging Trends: State-of-the-Art for Experts** *(Registration required)*	
8:00 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Introduction and Overview of Course
8:10 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Lysosomal Disease Phenotypes
8:30 AM	**Steven U. Walkley** Albert Einstein College of Medicine New York, NY, United States	Lysosomal Function and Pathogenesis
9:00 AM	**Marc C. Patterson** Mayo Clinic Rochester, MN, United States	Remarkable Cases I
9:30 AM	**Break**	
9:45 AM	**Jeanine R. Jarnes** University of Minnesota Minneapolis, MN, United States	Current Treatments for Lysosomal Diseases
10:05 AM	**Amy Gaviglio** Centers for Disease Control and Prevention Atlanta, GA, United States	Newborn Screening
10:25 AM	**Jeanine R. Jarnes** University of Minnesota Minneapolis, MN, United States	Future Treatments for Lysosomal Diseases
10:45 AM	**Marc C. Patterson** Mayo Clinic Rochester, MN, United States	Remarkable Cases II
11:05 AM	**Cara O'Neill** Cure Sanfilippo Foundation Columbia, SC, United States	Patient Advocate's Perspective on Experimental Therapies
11:25 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Closing Remarks
11:30 AM	**Adjourn**	
11:30 AM	**Lunch - on own**	

Basic Science I **Co-Chairs: Jill A. Morris & Danilo A. Tagle**

Disease Mechanisms, Pathology and Biomarkers

1:00 PM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and 2020 Young Investigator Award Presentation
1:15 PM	**Soumeya Bekri** Rouen University Hospital Rouen, France	Predictive biological patterns in Fabry disease revealed by integrative omics machine learning analysis
1:30 PM	**Anastasia G. Henry** Denali Therapeutics South San Francisco, CA, United States	Brain delivery and efficacy of an intravenously-administered lysosomal enzyme using a blood-brain barrier transport vehicle
1:45 PM	**Petra Oliva** ARCHIMEDlife Vienna, Austria	Differential diagnosis of Niemann-Pick disease types A and B in cases of suspected Gaucher disease
2:00 PM	**Shaun C. Bolton** University Hospital Birmingham NHS Foundation Trust Birmingham, United Kingdom	International Niemann-Pick Disease Registry (INPDR): The characteristics of ASMD and NPC patients
2:15 PM	**Ibane Abasolo** Vall d'Hebron Institute of Research Barcelona, Spain	Extracellular vesicles increase the enzymatic activity of lysosomal proteins and improve the efficacy of enzyme replacement therapy in Fabry disease
2:30 PM	**Behzad Najafian** University of Washington Seattle, WA, United States	Podocyte globotriaosylceramide (GL-3) content in female adult patients with Fabry disease and amenable mutations reduces following 6 months of treatment with migalastat
2:45 PM	**Break**	
3:15 PM	**Weihua Tian** University of Copenhagen Copenhagen, Denmark	Long-acting glyco-design (LAGD) for improved kinetics and distribution of α-galactosidase A
3:30 PM	**Poulomee Bose** Centre Hospitalier Universitaire Sainte-Justine (CHU St. Justine) Montreal, QC, Canada	Early synaptic dysfunction in MPS IIIC
3:45 PM	**Takumi Era** IMEG, Kumamto University Kumamoto, Japan	Presynaptic dysfunction in neurons derived from Tay-Sachs-iPSCs
4:00 PM	**Sarah Kim** University of Minnesota Minneapolis, MN, United States	Quantification of cerebrospinal fluid chitotriosidase in a clinical laboratory is validated for use in diagnosis and clinical trials
4:15 PM	**Mohammad A. Hossain** Advanced Clinical Research Centre Kawasaki, Kanagawa, Japan	DNA methylation study of GLA gene and its association with autophagy and clinical severity of heterozygous Fabry disease females
4:30 PM	**Poster Reception in Exhibit Hall**	
6:30 PM	**Satellite Symposia – TBD**	

Tuesday, February 11, 2020

Basic Science II — Co-Chairs: Brian Bigger & Sarah Kim

Developing Therapeutic Approaches in the Laboratory

6:15 AM	**Satellite Symposium**	
6:15 AM	**Satellite Symposium**	
7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Announcements Presentation of 2020 Roscoe O. Brady Award for Innovation and Accomplishment to John F. Crowley
7:45 AM	**John F. Crowley** Amicus Therapeutics, Inc Cranbury, NJ, United States	Innovation Award Speaker: The Moral Obligation to Ensure Access to Medicines for All Patients in Need
8:15 AM	**Jeffrey Y. Huang** Children's Hospital of Orange County Orange, CA, United States	Longitudinal assessment and immune response to recombinant GAA in CRISPR-Cas9 generated Pompe disease knock-in mice
8:30 AM	**Maria Dolores Ledesma** Centro Biologia Molecular Severo Ochoa Madrid, Spain	Inhibition of fatty acid amide hydrolase prevents pathology in a mouse model of acid sphingomyelinase deficiency by rescuing downregulated endocannabinoid signalling
8:45 AM	**Rebecca C. Ahrens-Nicklas** The Children's Hospital of Philadelphia Philadelphia, PA, United States	Efficacy of cell-type specific rescue in a new mouse model of CLN3 disease
9:00 AM	**Vera Niederkofler** QPS Austria GmbH Grambach, Austria	Neuroinflammation in mouse models of two different lysosomal diseases
9:15 AM	**Kimmo Lehtimäki** Charles River Discovery Kuopio, Finland	Longitudinal characterization of the *Cln8*$^{mnd-/-}$ mouse model of CLN8 Batten disease fine motor performance, retinal degeneration, brain pathology, and metabolic changes
9:30 AM	**Lalitha Belur** University of Minnesota Minneapolis, MN, United States	Systemic high-level IDUA enzyme activity with correction of neurologic deficit in MPS I mice by *ex vivo* lentiviral transduction of hematopoietic stem cells
9:45 AM	**Break & Exhibits**	
10:15 AM	**Dao Pan** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	miR-143 regulates lysosomal enzyme transport across blood-brain barrier and improves CNS treatment for Hurler syndrome
10:30 AM	**Natalia Gomez-Ospina** Stanford University Stanford, CA, United States	Monocyte lineage-specific glucocerebrosidase expression in human hematopoietic stem cells: A universal genome editing strategy for Gaucher disease
10:45 AM	**Malte Lenders** University Hospital Muenster Muenster, Germany	Neutralizing anti-drug antibodies inhibit endothelial enzyme uptake and activity in Fabry disease

11:00 AM	**Zully Pulido** Pontificia Universidad Javeriana Bogotá D.C., Colombia	Recombinant hexosaminidases conjugated to magnetite nanoparticles: Alternative therapeutic treatment routes in GM2 fibroblasts
11:15 AM	**Elena V. Batrakova** University of North Carolina Durham, NC, United States	Extracellular vesicles as drug delivery vehicles for lysosomal enzyme TPP1 to treat Batten disease
11:30 AM	**Lunch - on own or satellite symposia**	Exhibit hall is open
11:45 AM	**Satellite Symposium**	
11:45 AM	**Satellite Symposium**	

Translational Research I — Co-Chairs: Joseph J. Orsini & Amy Gaviglio

1:00 PM	**Brian Kevany** Abeona Therapeutics Cleveland, OH, United States	A novel AAV capsid with improved tropism to heart, kidney and PNS for treatment of Fabry disease
1:15 PM	**Li Ou** University of Minnesota Minneapolis, MN, United States	Liver-targeting gene editing achieves significant neurological benefits in MPS I mice
1:30 PM	**Scott Kerns** Abeona Therapeutics Cleveland, OH, United States	Combination AAV delivery to target vision loss and CNS manifestations in CLN3 disease
1:45 PM	**Halil Dundar** Gazi University Faculty of Medicine Ankara, Turkey	Triamterene-induced suppression of R227X premature termination codon in Fabry disease
2:00 PM	**Marisa Eve Pulcrano** University of California, San Francisco San Francisco, CA, United States	Translating a novel fetal therapy for lysosomal diseases into clinical care: The race for approval to treat one patient with mucopolysaccharidosis type VII
2:15 PM	**Paul J. Orchard** University of Minnesota Minneapolis, MN, United States	High dose hematopoietic stem cell transplantation leads to rapid hematopoietic and microglial recovery and disease correction in a mouse model of Hurler syndrome
2:30 PM	**Ari Zimran** Shaare Zedek Medical Center Jerusalem, Israel	Real life data on the safety and efficacy of ambroxol for patients with Gaucher disease or GBA-related Parkinson disease
2:45 PM	**Break & Exhibits**	
3:15 PM	**Michael H. Gelb** University of Washington Seattle, WA, United States	A universal newborn and diagnostic screening platform for lysosomal diseases and beyond
3:30 PM	**Melissa Wasserstein** Children's Hospital at Montefiore Bronx, NY, United States	"ScreenPlus": A comprehensive, dynamic, multi-disorder newborn screening pilot program
3:45 PM	**Ankit K. Desai** Duke University Durham, NC, United States	Benefits of prophylactic short-course immunomodulation in patients with infantile Pompe disease: Demonstration of long-term safety and efficacy in a large cohort
4:00 PM	**Dominique P. Germain** University of Versailles– St. Quentin en Yvelines (UVSQ) Montigny, France	The benefits, challenges and regional differences of family screening in rare genetic diseases: Lessons from Fabry disease

4:15 PM	**Dau-Ming Niu** Taipei Veterans General Hospital Taipei, Taiwan	Early detection of the irreversible cardiac damages in the adults with late onset Fabry disease in a large cohort study via newborn screening
4:30 PM	**Poster Reception in Exhibit Hall**	
6:30 PM	**Satellite Symposium**	
6:30 PM	**Satellite Symposium**	

Wednesday, February 12, 2020

Translational Research II — Co-Chairs: Philip J. Brooks & Ellen Sidransky

6:15 AM	**Satellite Symposium**	
6:15 AM	**Satellite Symposium**	
7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	2020 Patient Advocate Leader (PAL) Award Announcement and Presentation to Cara O'Neill
7:45 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	*Keynote Address*: Navigating Clinical Trials
8:15 AM	**Nicholas A. Bascou** University of Pittsburgh Medical Center (UPMC) Children's Hospital of Pittsburgh Pittsburgh, PA, United States	A prospective natural history study of metachromatic leukodystrophy: A 20 year study
8:30 AM	**Derralynn A. Hughes** University College London London, United Kingdom	First-in-human study of a liver-directed AAV gene therapy (FLT190) in Fabry disease
8:45 AM	**Margaret McGovern** Stony Brook School of Medicine Stony Brook, NY, United States	Prospective study of the natural history of chronic acid sphingomyelinase deficiency in children and adults: Eleven years of observation
9:00 AM	**Donna L. Bernstein** Mount Sinai School of Medicine New York, NY, United States	Lysosomal acid lipase deficiency and hematologic cancer predisposition
9:15 AM	**Jane Louise Kinsella** Royal Manchester Children's Hospital Manchester, United Kingdom	Case report of the first patient treated with ex-vivo autologous haematopoietic stem cell gene therapy transplant in mucopolysaccharidosis type IIIA
9:30 AM	**Fulvio Mavilio** Audentes Therapeutics San Francisco, CA, United States	Pre-clinical safety and efficacy findings of AT845, a novel gene replacement therapy for Pompe disease targeting skeletal muscle and heart
9:45 AM	**Break & Exhibits**	
10:15 AM	**George Karkashadze** Scientific Research Institute of Pediatrics and Child Health CCH RAoS Moscow, Russian Federation	Abnormalities in the cerebral cortex in Gaucher disease type 1: Findings from the ENIGMA storage disease working group

10:30 AM	**Erik A. Lykken** University of Texas (UT) Southwestern Medical Center Dallas, TX, United States	Combination intrathecal and intravenous gene therapy reveals a dominant role for treatment age in determining survival and behavioral outcomes in the mouse model of infantile neuronal ceroid lipofuscinosis
10:45 AM	**Jacinthe Gingras** Homology Medicines Bedford, MA, United States	HMI-202: Investigational gene therapy for treatment of metachromatic leukodystrophy (MLD)
11:00 AM	**Umut Cagin** Genethon Évry, France	Liver expression of secretable GAA rescues advanced Pompe disease at the biochemical, functional, and transcriptional level in Gaa-/- mice
11:15 AM	**Carlos J. Miranda** Freeline Therapeutics Stevenage, United Kingdom	One-off liver directed AAV gene therapy achieves long term uptake of acid beta-glucocerebrosidase by macrophages of affected tissues in Gaucher disease
11:30 AM	**Lunch - on own or satellite symposia**	Exhibit Hall is open
11:45 AM	**Satellite Symposium**	
11:45 AM	**Satellite Symposium**	

Clinical Trials I — Co-Chairs: Stephen C. Groft & Tiina K. Urv

Clinical Trials for Registration

1:00 PM	**John Mitchell** Montreal Children's Hospital Montreal, QC, Canada	Farber disease (acid ceramidase deficiency) natural history study: Prospective and retrospective clinical data
1:15 PM	**Manisha Balwani** Icahn School of Medicine at Mount Sinai Hospital New York, NY, United States	Clinical manifestations of lysosomal acid lipase deficiency (LAL-D): The international LAL-D Registry
1:30 PM	**Christoph Schwering** University Medical Center Hamburg-Eppendorf Hamburg, Germany	Development of the “Hamburg best practice guidelines for ICV-enzyme replacement therapy (ERT) in CLN2 disease” based on 5 years treatment experience in 48 patients
1:45 PM	**George Diaz** Icahn School of Medicine at Mount Sinai New York, NY, United States	Preliminary data from first clinical trial of enzyme replacement therapy with olipudase alfa in pediatric patients with chronic visceral and neurovisceral acid sphingomyelinase deficiency
2:00 PM	**Nuthana Prathivadi Bhayankaram** Royal Manchester Children's Hospital Manchester, United Kingdom	Umbilical cord blood transplant is the preferred stem cell source in children with MPS IH (Hurler syndrome) undergoing hematopoietic stem cell transplantation
2:15 PM	**Kevin M. Flanigan** Nationwide Children's Hospital Columbus, OH, United States	Interim results of Transpher A, a multicenter, single-dose, phase 1/2 clinical trial of ABO-102 gene therapy for Sanfilippo syndrome type A (mucopolysaccharidosis type IIIA)

2:30 PM	**Frits Wijburg** Amsterdam UMC Amsterdam, Netherlands	Phase 2-3 gene therapy trial using adeno-associated virus vector for patients with mucopolysaccharidosis type IIIA
2:45 PM	**Break & Exhibits**	
3:15 PM	**Kim L. McBride** Nationwide Children's Hospital Columbus, OH, United States	Safety, tolerability and preliminary evidence of biopotency in Transpher B, a multicenter, single-dose, phase 1/2 clinical trial of ABO-101 gene therapy for Sanfilippo syndrome type B (mucopolysaccharidosis type IIIB)
3:30 PM	**Raymond Y. Wang** Children's Hospital of Orange County (CHOC) Children's Specialists Orange, CA, United States	Long-term safety and efficacy of vestronidase alfa, rhGUS enzyme replacement therapy, in subjects with mucopolysaccharidosis type VII
3:45 PM	**Julia B. Hennermann** University Medical Center Mainz Mainz, Germany	Puberty, fertility and pregnancy in patients with mucopolysaccharidosis and mucolipidosis: A multicentre cross-sectional study
4:00 PM	**Torayuki Okuyama** National Center for Child Health and Development Tokyo, Japan	Therapy for MPS II with an intravenous blood-brain barrier-crossing enzyme (JR-141): 26-week results from a phase 3 study in Japan suggesting significant efficacy against central nervous system and systemic symptoms
4:15 PM	**Marc Patterson** Mayo Clinic Rochester, MN, United States	Efficacy and safety of arimoclomol in patients with Niemann-Pick disease type C: Results from a double-blind, randomized placebo-controlled trial with a novel treatment
4:30 PM	**Poster Reception in Exhibit Hall**	
6:30 PM	**Satellite Symposium**	

Thursday, February 13, 2020

Clinical Trials II — Co-Chairs: Yoshikatsu Eto & Priya S. Kishnani

Clinical Outcomes

6:15 AM	**Satellite Symposium**	
6:15 AM	**Satellite Symposium**	
7:25 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome
7:30 AM	**Peter Marks** Center for Biologics Evaluation and Research U.S. Food and Drug Administration Silver Spring, MD, United States	*Keynote Address:* The Shift from Personalized to Individualized Therapies
8:00 AM	**Samuel Gröschel** University Children's Hospital Tübingen, Germany	Effect of intrathecal recombinant human arylsulfatase A enzyme replacement therapy on structural brain MRI in children with metachromatic leukodystrophy

8:15 AM	**Francesca Fumagalli** San Raffaele Telethon Institute for Gene Therapy (SR-TIGET), IRCCS San Raffaele Scientific Institute Milano, Italy	Lentiviral hematopoietic stem and progenitor cell gene therapy (HSPC-GT) for metachromatic leukodystrophy (MLD): Clinical outcomes from 33 patients
8:30 AM	**Emily de los Reyes** Nationwide Children's Hospital Columbus, OH	Single-dose AAV9-CLN6 gene transfer stabilizes motor and language function in CLN6-type Batten disease: Interim results from the first clinical gene therapy trial
8:45 AM	**David G. Warnock** University of Alabama Birmingham, CA, United States	Pegunigalsidase alfa, a novel PEGylated ERT, evaluated in Fabry disease patients with progressing kidney disease, RCT study design
9:00 AM	**Christoph Wanner** University of Würzburg Würzburg, Germany	Rationale and design of the MODIFY study: A phase 3 multicenter, double-blind, randomized, placebo-controlled, parallel-group study to determine the efficacy and safety of lucerastat oral monotherapy in adult subjects
9:15 AM	**Raphael Schiffmann** Baylor Research Institute Dallas, TX, United States	Venglustat combined with imiglucerase positively affects neurological features and brain connectivity in adults with Gaucher disease type 3
9:30 AM	**Pramod K. Mistry** Yale University School of Medicine New Haven, CT, United States	Individual patient responses to eliglustat in treatment-naïve adults with Gaucher disease type 1: Final data from the phase 3 ENGAGE trial
9:45 AM	**Break**	
10:15 AM	**David Kronn** New York Medical College Valhalla, NY, United States	Mini-COMET study: Safety, immunogenicity, and preliminary efficacy for repeat avalglucosidase alfa dosing in patients with infantile-onset Pompe disease (IOPD) who were previously treated with alglucosidase alfa and demonstrated clinical decline
10:30 AM	**Mazen M. Dimachkie** University of Kansas Medical Center Kansas City, KS, United States	NEO1 and NEO-EXT studies: Long-term safety and exploratory efficacy of repeat avalglucosidase alfa dosing for 5.5 years in late-onset Pompe disease patients
10:45 AM	**Stephanie Austin** Duke University Durham, NC, United States	Extended treatment with VAL-1221, a novel protein targeting cytoplasmic glycogen, in patients with late-onset Pompe disease
11:00 AM	**Paul Harmatz** University of California - San Francisco (UCSF) Benioff Children's Hospital Oakland, CA, United States	A new randomized placebo controlled study to establish the safety and efficacy of velmanase alfa (human recombinant alpha-mannosidase) enzyme replacement therapy for the treatment of alpha-mannosidosis
11:15 AM	**Angela Schulz** University Medical Center Hamburg-Eppendorf Hamburg, Germany	Cerliponase alfa for the treatment of CLN2 disease in an expanded patient cohort including children younger than three years: Interim results from an ongoing clinical study
11:30 AM	**Lunch - on own or satellite symposia**	
11:45 AM	**Satellite Symposium**	
11:45 AM	**Satellite Symposium**	

The following session is not available for CME/CE accrediation; CEU credits for GCs may apply.

Contemporary Forum — Co-Chairs: R. Scott McIvor & Anne R. Pariser

1:00 PM	**Dwight Koeberl** Duke University School of Medicine Durham, NC, United States	A phase 1 study of gene therapy with ACTUS-101 in late-onset Pompe disease
1:15 PM	**Sean M. Armour** Spark Therapeutics, Inc. Philadelphia, PA, United States	Preclinical development of *SPK-3006*, an investigational liver-directed AAV gene therapy for the treatment of Pompe disease
1:30 PM	**Alissa Brandes** Prevail Therapeutics New York, NY, United States	Gene therapy PR006 increased progranulin levels and improved lysosomal related phenotypes in model systems
1:45 PM	**Birgitte Volck** AVROBIO, Inc. Cambridge, MA, United States	AVR-RD-01 lentiviral gene therapy reduces Gb3 substrate in endothelial cells of renal peritubular capillaries in a previously untreated classic Fabry disease male patient
2:00 PM	**Lin Liu** M6P Therapeutics St. Louis, MO, United States	A new platform technology for next generation lysosomal enzyme replacement and potential gene therapy in the treatment of lysosomal diseases
2:15 PM	**Manolo Bellotto** Gain Therapeutics Lugano, Switzerland	Brain penetrant structurally targeted allosteric regulators for treating GLB1-related disorders
2:30 PM	**Julie C. Ullman** Denali Therapeutics South San Francisco, CA, United States	Novel FACS based method demonstrates CNS cell-type distribution and efficacy of a BBB penetrant ERT in a mouse model of MPS II
2:45 PM	**Break**	
3:15 PM	**William Casey Hallows** Codexis Redwood City, CA, United States	Engineering α-galactosidase A (GLA) to improve protein stability, efficacy and reduced immune response for the treatment of Fabry disease
3:30 PM	**Nicholas France** E-Scape Bio, Inc San Francisco, CA, United States	Sphingosine-1-phosphate receptor type 5 (S1P5) agonism: A potential new mechanism for the treatment of neuronopathic features of Niemann-Pick disease type C and neurodegenerative sphingolipidoses
3:45 PM	**Linda Ingemann** Orphazyme A/S Copenhagen N, Denmark	Rescue of NPC1 protein and effect on biomarkers by arimoclomol treatment in Niemann-Pick disease type C
4:00 PM	**Emanuela Izzo** BioMarin Pharmaceutical Inc. Novato, CA, United States	Utility of gene panel testing in children with seizure onset after 2 years of age: Results from a European and Middle Eastern epilepsy genetic testing program
4:15 PM	**R. Scott McIvor** Immusoft Corporation Seattle, WA, United States	Iduronidase-transposed human B lymphocytes correct enzyme deficiency and glycosaminoglycan storage disease in immunodeficient MPS I mice
4:30 PM	**Adjourn**	
4:30 -5:30 PM	**Networking Reception**	
5:00 -7:00 PM	**Lysosomal Disease Network (LDN) Annual Meeting**	

***Agenda subject to change*