WORLD*Symposium*™ 2017
Program

Monday, February 13

9:00 – 12:00	**Council Of Patient Advocates (COPA) Workshop**	WORLD Translation and WORLD Activation
1:00 – 5:00	**Emerging Trends**	State of the art for experts *(Registration required)*
1:00	**Chester B. Whitley**	Welcome and introduction
1:05	**Steven U. Walkley**	Molecular & cell biology of lysosomes
2:00	**Chester B. Whitley**	Lysosomal diseases & pathology
2:55	**Break**	Break
3:10	**Jeanine R. Utz**	Treatments for lysosomal diseases
4:05	**Marc C. Patterson**	Remarkable cases
5:00	**Adjourn**	
6:00	**Satellite Symposium**	

Tuesday, February 14

Basic Science I — Co-Chairs: R. Scott McIvor & Danuta Krotoski

6:30	**Satellite Symposium**	
7:45	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Innovation Award Announcement
7:55	**Konrad Sandhoff** LIMES Institute c/o Kekulé-Institute, Rheinische Friedrich-Wilhelms-Universitaet Bonn Bonn, Germany	Sphingolipidoses membrane lipids regulate and modify sphingolipid catabolism, its enzymes, lipid binding and transfer proteins
8:30	**Li Ou** University of Minnesota Minneapolis, MN, United States	Proteomic analysis of mucopolysaccharidosis type I mouse brain with two-dimensional polyacrylamide gel electrophoresis
8:45	**Yumiko V. Taguchi** Yale University New Haven, CT, United States	Glucosylsphingosine accelerates α-synuclein pathology in *GBA*-associated Parkinson disease
9:00	**Sharon Byers** University of Adelaide Adelaide, Australia	Cell cycle progression is disrupted in murine MPS VII growth plate chondrocytes

9:15	**Richard Roberts** Minoryx Therapeutics Mataró (Barcelona), Spain	Allosteric, non-inhibitory pharmacological chaperones for the treatment of lysosomal diseases
9:30	**Kasturi Haldar** University of Notre Dame Notre Dame, IN, United States	Chronic HDACi therapy to treat multiple lysosomal diseases
9:45	**Break and Exhibits**	
10:15	**Jeong-A Lim** National Institutes of Health Bethesda, MD, United States	Modulation of mTOR signaling as a therapeutic approach for Pompe disease
10:30	**Richard W.D. Welford** Actelion Pharmaceuticals Allschwil, Switzerland	Lucerastat, an iminosugar for substrate reduction therapy in Fabry disease: preclinical evidence
10:45	**Marshall W. Huston** Sangamo Biosciences Richmond, CA, United States	Liver-based expression of the human alpha-galactosidase A gene (GLA) in a murine Fabry model results in continuous supra-physiological enzyme activity and effective substrate reduction
11:00	**Spencer Goodman** University of California, San Diego La Jolla, CA, United States	Delivery highways: tunneling nanotubes facilitate transfer of therapeutic molecules for gene therapy treatment of cystinosis
11:15	**Shih-hsin Kan** **Los Angeles Biomedical Research** Institute at Harbor-UCLA Torrance, CA, United States	AAV5-mediated gene therapy with choroid plexus-directed α-n-acetyl-glucosaminidase expression in Sanfilippo syndrome type B mice
11:30	**Lalitha Belur** University of Minnesota Minneapolis, MN, United States	Recovery of neurologic function in mucopolysaccharidosis type I mice with existing neurocognitive dysfunction by treatment with AAV9-IDUA vector
11:45	**Lunch - On Own or Satellite Symposia**	

Basic Science II — Co-Chairs: Walter Low, Li Ou & Rashmi Gopal-Srivastava

1:00	**Lauren E. Ellis** Auburn University Auburn, AL, United States	Cardiovascular manifestations of feline Sandhoff disease after intravenous AAV gene therapy
1:15	**Shahzeb Hassan** National Institutes of Health Bethesda, MD, United States	Looking beyond the realm of traditional genetics: DNA methylation differences in discordant Gaucher disease twins
1:30	**Mia Horowitz** Tel Aviv University Ramat Aviv, Israel	The contribution of mutant GCase to the accumulation and aggregation of alpha synuclein
1:45	**Manoj K. Pandey** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Glucosylceramide partnership with immunological villain in Gaucher disease

2:00	**Vincent Puy** Chu Amiens Amiens, France	In Sanfilippo syndrome, heparan sulfate hexasaccharides are the most pathogenic fractions involved in glia activation.
2:15	**Brian Bigger** University of Manchester Manchester, United Kingdom	Mucopolysaccharidosis IIIA storage substrate drives an innate immune neuro-inflammatory response
2:30	**Melani A. Solomon** University of Maryland College Park, MD, United States	Transcytosis alterations in lysosomal diseases
2:45	**Break and Exhibits**	
3:15	**Prakrit V. Jena** Memorial Sloan Kettering Cancer Center New York, NY, United States	Optical non-invasive detection of Niemann-Pick disease *in vitro* and *in vivo*
3:30	**Heechun Kwak** Mogam Institute for Biomedical Research Yongin, Republic of Korea	MPS II model cell line by CRISPR-Cas9 technique
3:45	**Sang-oh Han** Duke University Durham, NC, United States	Beneficial effects of carvedilol with enzyme replacement therapy in Pompe disease
4:00	**Yoseph Shaaltiel** Protalix Carmiel, Israel	Characterization of a chemically modified plant cell culture expressed human α-galactosidase-A enzyme for treatment of Fabry disease
4:15	**Kohji Itoh** Tokushima University Tokushima, Japan	A transgenic silkworm overexpressing human lysosomal enzyme as a novel resource for producing recombinant glycobiologics and its application to development of enzyme replacement therapy for lysosomal diseases
4:30	**Poster Reception in the Exhibit Hall**	
6:30	**Satellite Symposium**	

Wednesday, February 15

Translational Research I — Co-Chairs: Alessandra Biffi & Danilo Tagle

6:30	**Satellite Symposium**	
7:40	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and Announcements
7:45	**Elsa G. Shapiro** University of Minnesota Minneapolis, MN, United States	Keynote Address: Understanding and measuring neurodegeneration in childhood onset lysosomal diseases

8:15	**Patrick V. Hopkins** Missouri State Public Health Laboratory Jefferson City, MO, United States	State-wide newborn screening for four lysosomal diseases reveals high incidence rate for Pompe and Fabry diseases
8:30	**Soumeya Bekri** Normandie University Rouen University Hospital Rouen, France	Development, analytical validation and implementation of a next generation sequencing panel to assess lysosomal diseases
8:45	**Abdellah Tebani** Normandie University Rouen University Hospital Rouen, France	Metabolic phenotyping of mucopolysaccharidoses using untargeted liquid chromatography ion mobility mass spectrometry-based strategy
9:00	**R. Scott McIvor** University of Minnesota Minneapolis, MN, United States	Relative effectiveness of different routes of AAV administration for gene therapy of mucopolysaccharidosis
9:15	**Russell DeKelver** Sangamo BioSciences Richmond, CA, United States	ZFN-mediated *in vivo* genome editing results in phenotypic correction in murine MPS I and MPS II models
9:30	**Hélène F. E. Gleitz** University of Manchester Manchester, United Kingdom	Whole body correction of severe mucopolysaccharidosis type II by lentiviral-mediated stem cell gene therapy with blood-brain barrier-crossing peptides
9:45	**Break and Exhibits**	
10:15	**Kelly M. Podetz-Pedersen** University of Minnesota Minneapolis, MN, United States	Neurologic improvement in a mouse model of Hunter syndrome (mucopolysaccharidosis type II) by treatment with AAV9 vector after the development of cognitive dysfunction
10:30	**Tatiana Lobry** University of California, San Diego San Diego, CA, United States	Towards a phase I clinical trial for autologous hematopoietic stem cells transplantation in cystinosis
10:45	**Claire O'Leary** University of Manchester Manchester, United Kingdom	Gene therapy mediated correction of neurological manifestations of MPS IIIC using a novel AAV serotype
11:00	**Eun-Young Choi** National Institutes of Health Bethesda, MD, United States	Dose-ranging comparison of choroid plexus-directed versus pan-neuronal-directed recombinant AAV gene therapy in a murine model of alpha-mannosidosis
11:15	**Alejandra J. Rozenberg** University of North Carolina at Chapel Hill Chapel Hill, NC, United States	Early intrathecal gene therapy extends lifespan and improves quality of life in a mouse model for infantile neuronal ceroid lipofuscinosis
11:30	**Heather L. Gray-Edwards** Auburn University Auburn, AL, United States	Long term survival of sheep with Tay-Sachs disease after intracranial delivery of a novel bicistronic AAV gene therapy vector
11:45	**Lunch - On Own or Satellite Symposia**	

Translational Research II **Co-Chairs: Yoshikatsu Eto & Ellen Sidransky**

1:00	**Reid Martin** BioStrategies-LC Jonesboro, AR, United States	Receptor-independent mechanisms of RTB lectin-mediated ERT delivery provide unique advantages in enzyme uptake capacity, transcytosis, and lysosomal correction
1:15	**Grzegorz Wegrzyn** University of Gdansk Gdansk, Poland	Genistein - a lysosomal stimulator for treatment of various lysosomal diseases
1:30	**Andrew D. Baik** Regeneron Pharmaceuticals Tarrytown, NY, United States	Engineering tissue specific delivery of enzymes for lysosomal disease treatment
1:45	**Edward H. Schuchman** Icahn School of Medicine at Mount Sinai New York, NY, United States	Proof-of-concept studies underlying enzyme replacement therapy for acid ceramidase deficiency
2:00	**Chanchala Kaddi** Sanofi Genzyme Cambridge, MA, United States	Quantitative systems pharmacology model of acid sphingomyelinase deficiency and the enzyme replacement therapy olipudase alfa is an innovative tool for linking pathophysiology and pharmacology
2:15	**Hung Do** Amicus Therapeutics, Inc. Cranbury, NJ, United States	Stabilized next-generation recombinant human acid alpha-glucosidase ATB200 clears accumulated glycogen and reverses cellular dysfunction to increase functional muscle strength in a mouse model of Pompe disease
2:30	**Han-Hyuk Lim** Duke University Medical Center Durham, NC, United States	Immunomodulation to enzyme replacement therapy with tolerogenic nanoparticles containing rapamycin in a murine model of Pompe disease
2:45	**Break and Exhibits**	
3:15	**Thomas Kirkegaard** Orphazyme Copenhagen, Denmark	Heat shock protein-based therapy for sphingolipidoses
3:30	**Hojun Choi** Korea Advanced Institute of Science & Technology Daejeon, Republic of Korea	Delivery of lysosomal enzymes via EXPLOR: exosome engineered for protein loading by optogenetically reversible protein interaction
3:45	**Jess G. Thoene** University of Michigan Ann Arbor, MI, United States	Microvesicle-mediated delivery of cystinosin to rabbit cornea
4:00	**Gustavo Maegawa** University of Florida Gainesville, FL, United States	Identification of psychosine-reducing small molecule agents for Krabbe disease

4:15	**N. Matthew Ellinwood** Iowa State University Ames, IA, United States	Preliminary findings of a twenty-six week or longer intracerebroventricular infusion study of BMN 250 administered once every 2 weeks in a canine model of mucopolysaccharidosis type IIIB
4:30	**Poster Reception & Presentation**	
6:30	**Satellite Symposium**	

Thursday, February 16

Clinical Trials I — Co-Chairs: Lynda Polgreen & Frits Wijburg

6:30	**Satellite Symposium**	
7:40	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and Announcements
7:45	**Richard A. Moscicki** Food and Drug Administration Silver Spring, MD, United States	Keynote Address: An FDA Perspective on Rare Disease Drug Development
8:15	**Pramod K. Mistry** Yale University School of Medicine New Haven, CT, United States	Long-term results of ENGAGE: a phase 3, randomized, double-blind, placebo-controlled, multi-center study investigating the efficacy and safety of eliglustat in adults with type 1 Gaucher disease
8:30	**Timothy M. Cox** University of Cambridge Addenbrooke's Hospital Cambridge, United Kingdom	Maintenance of quality of life in adults with type 1 Gaucher disease previously stabilized on enzyme therapy who were switched to oral eliglustat: 4 year results of the ENCORE trial
8:45	**Ari Zimran** Shaare Zedek Medical Center Jerusalem, Israel	Pharmacokinetics, safety, and efficacy of rapid infusions of velaglucerase alfa in adult patients with Gaucher disease
9:00	**Line Borgwardt** University Hospital of Copenhagen Rigshospitalet Copenhagen, Denmark	Long-term enzyme replacement therapy with velmanase alfa (human recombinant alpha-mannosidase) slows disease progression in adult patients suffering from alpha-mannosidosis
9:15	**Ana Cristina Puga** Sanofi Genzyme Cambridge, MA, United States	Olipudase alfa for the treatment of acid sphingomyelinase deficiency (ASMD): safety and efficacy in adults treated for 30 months
9:30	**Melissa Wasserstein** Children's Hospital at Montefiore Bronx, NY, United States	The New York pilot newborn screen for lysosomal diseases: 40 month data
9:45	**Break and Exhibits**	

10:15	**Angela Schulz** University Medical Center Hamburg-Eppendorf Hamburg, Germany	Long-term safety and efficacy of intracerebroventricular enzyme replacement therapy with cerliponase alfa in children with CLN2 disease: interim results from an ongoing multicenter, multinational extension study
10:30	**Francyne Kubaski** University of Delaware Newark, DE, United States	Hematopoietic stem cell transplantation for patients with mucopolysaccharidosis type II
10:45	**Troy Lund** University of Minnesota Minneapolis, MN, United States	Triple therapy for MPS IH: intrathecal iduronidase, IV-ERT, and hematopoietic cell transplant
11:00	**Agnes Chen** Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center Torrance, CA, United States	A randomized open-label clinical trial of intrathecal recombinant human alpha-l-iduronidase for cognitive decline in mucopolysaccharidosis type I
11:15	**Roberto Giugliani** Hospital de Clinicas de Porto Alegre Porto Alegre, Brazil	Intravenous infusion of iduronidase-IgG and its impact on the central nervous system in children with Hurler syndrome
11:30	**Christian J. Hendriksz** University of Pretoria Pretoria, South Africa	Elosulfase alfa treatment and changes in physical functioning and disability in Morquio syndrome type A
11:45	**Lunch - On Own or Satellite Symposia**	

Clinical Trials II		**Co-Chairs: James Cloyd & Jill Morris**
1:00	**Rossella Parini** Fondazione MBBM, Azienda Ospedaliera San Gerardo Monza, Italy	Sub-analysis of long-term elosulfase alfa treatment outcomes in adults with Morquio syndrome type A
1:15	**Marc Tardieu** Université Paris Sud Le Kremlin-Bicêtre, France	Intracerebral administration of rAAV2/5hNAGLU vector in children with MPS IIIB: results at 30 months of a phase I/II trial
1:30	**Kevin M. Flanigan** Research Institute of Nationwide Children's Hospital Columbus, OH, United States	Systemic gene transfer of scAAV9.U1a.hSGSH for MPS IIIA: tolerability and preliminary evidence for a biochemical effect
1:45	**Paul Harmatz** Children's Hospital Oakland Oakland, CA, United States	A novel, randomized, placebo-controlled, blind-start, single-crossover phase 3 study to assess the efficacy and safety of UX003 (rhGUS) enzyme replacement therapy in patients with MPS VII
2:00	**Barbara K. Burton** Northwestern University Chicago, IL, United States	Long-term benefit of sebelipase alfa over 76 weeks in children and adults with lysosomal acid lipase deficiency (LAL-D) (ARISE)

2:15	**Mark Friedman** Alexion Pharmaceuticals, Inc. New Haven, CT, United States	Effect of sebelipase alfa on survival to 3 years of age and liver function in infants with rapidly progressive lysosomal acid lipase deficiency
2:30	**Uma Ramaswami** Royal Free London NHS Foundation Trust, University College London London, United Kingdom	A randomized, phase 3B, open-label, parallel-group study of agalsidase beta in treatment-naive male pediatric patients with Fabry disease without severe symptoms (FIELD study): GL-3 clearance from kidney cells
2:45	**Break and Exhibits**	
3:15	**Ans van der Ploeg** Erasmus MC University Medical Center Rotterdam, Netherlands	Long-term efficacy of alglucosidase alfa in late-onset Pompe disease
3:30	**Dominique P. Germain** University of Versailles–St. Quentin en Yvelines (UVSQ) Montigny, France	Efficacy of migalastat in a cohort of male patients with the classic Fabry phenotype in the FACETS phase 3 study
3:45	**Derralynn Hughes** Royal Free Hospital, NHS Foundation Trust, University College London London, United Kingdom	One-year follow up of Fabry disease patients treated by IV administration of a plant derived alpha-Gal-A enzyme: safety and efficacy
4:00	**Ulla Feldt-Rasmussen** Rigshospitalet Copenhagen University Hospital Copenhagen, Denmark	Efficacy and safety of migalastat, an oral pharmacologic chaperone for Fabry disease: results from two randomized phase 3 studies, FACETS and ATTRACT
4:15	**David Warnock** University of Alabama - Birmingham Birmingham, AL, United States	PRX-102 for treating Fabry disease - immunogenicity and PK results from a phase 1-2 study
6:00	**Banquet and Award Ceremony**	