WORLD*Symposium*™ 2018 Program

Monday, February 5, 2018

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 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 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	**R. Rodney Howell** University of Miami Miami, FL, 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 & New Treatment Awards
5:30 PM	**Opening Reception**	Exhibit Hall
6:30 PM	**Satellite Symposium**	

Tuesday, February 6, 2018

6:30 AM	**Satellite Symposia**	

Basic Science I — Co-Chairs: Walter Low & Danuta Krotoski

7:45 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Innovation Award Announcement
7:55 AM	**Mark Haskins** University of Pennsylvania Philadelphia, PA, United States	"…standing on the shoulders of Giants"
8:30 AM	**Patricia Dickson** Harbor-UCLA/LABioMed Torrance, CA, United States	Neuroimaging and neuropathology reveal progressively abnormal white matter and cerebrospinal fluid volume in MPS I dogs
8:45 AM	**Roselena S. Schuh** Universidade Federal do Rio Grande do Sul Porto Alegre, Brazil	Intravenous and intranasal genome editing using the CRISPR/Cas9 system leads to long-term improvements in MPS I mice
9:00 AM	**Jillian R. Brown** TEGA Therapeutics La Jolla, CA, United States	Guanidinylated neomycin conjugation enhances intranasal enzyme replacement in the brain
9:15 AM	**Yanyan Peng** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Evaluation of a novel, non-invasive iPSC based cell therapy for neuronopathic Gaucher disease
9:30 AM	**Jenny Serra-Vinardell** National Institutes of Health Bethesda, MD, United States	Patient-derived Gaucher induced pluripotent stem cells as a tool to understand common complex disorders
9:45 AM	**Break and Exhibits**	
10:15 AM	**Mia Horowitz** Tel Aviv University Ramat Aviv, Israel	The contribution of mutant glucocerebrosidase to the aggregation of alpha synuclein
10:30 AM	**Simon Heales** Great Ormond Street Hospital, University College London London, United Kingdom	Lysosomal glucocerebrosidase inhibition is associated with perturbed dopamine turnover: a mechanistic insight into the link between Gaucher and Parkinson disease
10:45 AM	**Benjamin McMahon** National Institutes of Health Bethesda, MD, United States	The importance of astrocytes in the pathophysiology of GBA1-associated Parkinson disease
11:00 AM	**Nadav I. Weinstock** State University of New York at Buffalo Buffalo, NY, United States	GALC ablation in Schwann cells produces a demyelinating peripheral neuropathy characterized by psychosine formation but lacking globoid cells

11:15 AM	**Brian W. Bigger** University of Manchester Manchester, United Kingdom	Interleukin-1 plays a central role in behaviour abnormalities in mucopolysaccharidosis type III (MPS III)
11:30 AM	**Chelsee T. Sauni** Phoenix Nest, Inc Brooklyn, NY, United States	Pilot enzyme replacement therapy with recombinant human glucosamine (N-acetyl)-6-sulfatase in mucopolysaccharidosis type IIID mouse model
11:45 AM	**Lunch – On Own or Satellite Symposia; Exhibit Hall Open**	

Basic Science II — Co-Chairs: Brian Bigger & Jill Morris

1:00 PM	**Alexey V. Pshezhetsky** CHU Ste-Justine, University of Montreal Montreal, QC, Canada	Chaperone therapy for mucopolysaccharidosis type IIIC
1:15 PM	**Sharon Byers** University of Adelaide Adelaide, Australia	Chondrogenesis and osteogenesis are delayed by MPS IVA keratan sulphate but not normal keratan sulphate
1:30 PM	**Fabian P.S. Yu** University of Toronto Toronto, ON, Canada	Ocular pathology and visual impairment in a mouse model of acid ceramidase deficiency
1:45 PM	**Salvatore Molino** Medical College of Wisconsin Milwaukee, WI, United States	Hepatocellular dysfunction and gene expression changes in the acid ceramidase deficient mouse
2:00 PM	**Daesung Shin** State University of New York at Buffalo Buffalo, NY, United States	Temporal *Galc* deletion reveals a critical vulnerable period in the pathogenesis of Krabbe leukodystrophy
2:15 PM	**Rebecca Ahrens-Nicklas** The Children's Hospital of Philadelphia Philadelphia, PA, United States	Neuronal network dysfunction in juvenile neuronal lipofuscinosis
2:30 PM	**Hemanth R. Nelvagal** Harbor-UCLA/LABioMed Torrance, CA, United States	Early gait abnormalities relate to brainstem and spinal cord pathology in CLN1 disease
2:45 PM	**Break and Exhibits**	
3:15 PM	**Zhirui Jiang** The University of Adelaide Adelaide, Australia	MPS VII mice display reduced circulating IGF1 and disrupted cell cycle progression in the growth plate
3:30 PM	**Christina R. Mikulka** Washington University School of Medicine St. Louis, MO, United States	Eliminating cross-correction allows for cell-specific expression of the lysosomal enzyme galactocerebrosidase in the twitcher mouse
3:45 PM	**Murtaza S. Nagree** University of Toronto Toronto, ON, Canada	In vivo enrichment of transduced cells to enhance gene therapy for Fabry disease

4:00 PM	**Daphne Chen** University of North Carolina, Chapel Hill Chapel Hill, NC, United States	Identification of novel AAV capsids for treatment of lysosomal disorders
4:15 PM	**Li Ou** University of Minnesota Minneapolis, MN, United States	Metabolomics profiling of mice and patients with Sandhoff disease to identify biomarkers
4:30 PM	**Poster Reception in Exhibit Hall**	Poster presenters with First Author Last Name starting with A-L displayed
6:30 PM	**Satellite Symposium**	

Wednesday, February 7, 2018

6:30 AM	**Satellite Symposia**	

Translational Research I — Co-Chairs: Danilo Tagle & R. Scott McIvor

7:45 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Patient Advocate Leader (PAL) Award
8:00 AM	**Petra Kaufmann** National Institutes of Health Bethesda, MD, United States	Keynote Address
8:30 AM	**Natalia Gomez-Ospina** Lucile Packard Children's Hospital Stanford, CA, United States	Engineering blood stem cells for autologous transplants for lysosomal diseases: correction of mucopolysaccharidosis type I using genome-edited hematopoietic stem and progenitor cells
8:45 AM	**Yewande E.O. Pearse** Harbor-UCLA/LABioMed Torrance, CA, United States	Neural stem cells provide continuous enzyme replacement therapy and reduce neuropathology in Sanfilippo syndrome type B mice
9:00 AM	**Stuart M. Ellison** University of Manchester Manchester, United Kingdom	Pre-clinical safety and efficacy evaluation of GMP lentiviral vector in preparation for a clinical trial of hematopoietic stem cell gene therapy in MPS IIIA
9:15 AM	**Manuela Corti** University of Florida Gainesville, FL, United States	Enabling redosing of AAV by immune management in Pompe disease: preclinical to clinical studies
9:30 AM	**Shaun Brothers** University of Miami Miami, FL, United States	Novel small molecule therapy development for MPS I
9:45 AM	**Break and Exhibits**	
10:15 AM	**Nina Raben** National Institutes of Health Bethesda, MD, United States	A major advance in the search for more effective therapy for Pompe disease

10:30 AM	**Iris Alroy** Eloxx Pharmaceuticals Waltham, MA, United States	Translational read-through of CTNS nonsense mutations and attenuation of CTNS nonsense-mediated mRNA decay by ELX-02 treatment
10:45 AM	**Eugeni V. Entchev** Inventiva Daix, France	Odiparcil is a promising substrate reduction therapy in MPS VI murine model
11:00 AM	**C. Ronald Scott** University of Washington Seattle, WA, United States	A high performance assay for the detection of MPS disorders, MLD, and CTX, from newborn blood spots
11:15 AM	**Anuj Chauhan** University of Florida Gainesville, FL, United States	Contact lens based therapy for ocular cystinosis
11:30 AM	**Yedda Li** Washington University Saint Louis, MO, United States	Combination therapy increases lifespan and improves clinicobehavioral performance in the murine model of globoid cell leukodystrophy
11:45 AM	**Lunch – On Own or Satellite Symposia; Exhibit Hall Open**	

Translational Research II — Co-Chairs: Rashmi Gopal-Srivastava & Jim Cloyd

1:00 PM	**Thomas Wechsler** Sangamo Therapeutics Richmond, CA, United States	ZFN-mediated in vivo genome editing of hepatocytes results in phenotypic correction in murine MPS I and MPS II models
1:15 PM	**Silvere Pagant** Icahn School of Medicine at Mount Sinai New York, NY, United States	ZFN-mediated in vivo genome editing results in therapeutic levels of α-galactosidase A and effective substrate reduction in Fabry knockout mice
1:30 PM	**Cristin Davidson** Albert Einstein College of Medicine Bronx, NY, United States	Gene therapy for the treatment of Niemann-Pick disease type C1: comparison of AAV9 to a novel serotype, AAV-PHP.B
1:45 PM	**Ying Sun** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Systemic delivery of acid β-glucosidase by SapC-based nanovesicles for neuronopathic Gaucher disease therapy
2:00 PM	**Anita Grover** BioMarin Pharmaceutical Inc. Novato, CA, United States	Translational dose-response and frequency scaling for BMN 250 administered via the intracerebroventricular route: predicting a clinically effective dosing regimen from animal models of disease for the treatment of Sanfilippo syndrome type B
2:15 PM	**Ai Yin Liao** University of Manchester Manchester, United Kingdom	Induction of immune tolerance to enzyme replacement therapy in mucopolysaccharidosis type I

2:30 PM	**Derek Kelaita** ArmaGen Inc. Calabasas, CA, United States	Platform technology for treatment of the brain in lysosomal disorders
2:45 PM	**Break and Exhibits**	
3:15 PM	**Hiroyuki Sonoda** JCR Pharmaceuticals Kobe, Japan	Blood-brain barrier-penetrating iduronate-2-sulfatase reduces brain glycosaminoglycans in mouse model of mucopolysaccharidosis type II
3:30 PM	**David G. Warnock** University of Alabama Birmingham, AL, United States	Enhanced pharmacokinetics profile of pegunigalsidase alfa (PRX-102) supports once-monthly 2mg/kg dosing for the treatment of Fabry disease
3:45 PM	**Andrew Baik** Regeneron Pharmaceuticals Tarrytown, NY, United States	Next-generation antibody-guided enzyme replacement therapy in Pompe disease mice
4:00 PM	**Kelly George** Sanofi Genzyme Framingham, MA, United States	Comprehensive exploratory study to identify novel biomarkers of Pompe disease
4:15 PM	**John Sinclair** BioMarin Pharmaceutical Inc. Novato, CA, United States	Intravitreal enzyme replacement therapy attenuates retinal disease progression in a canine model of neuronal ceroid lipofuscinosis type 2 (CLN2)
4:30 PM	**Poster Reception in Exhibit Hall**	Poster presenters with First Author Last Name starting with M-Z and all Late-Breaking abstracts displayed
6:30 PM	**Satellite Symposium**	

Thursday, February 8, 2018

6:30 AM	**Satellite Symposia**	

Clinical Trials I — Co-Chairs: Ellen Sidransky & Michael Mauer

7:40 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Announcement
7:45 AM	**R. Rodney Howell** University of Miami Miami, FL, United States	Keynote Address: What innovation has changed medical care more than newborn screening?
8:15 AM	**Amy Gaviglio** Minnesota Department of Health Minneapolis, MN, United States	State of national implementation for lysosomal diseases
8:30 AM	**Stacey A. Wong** Invitae San Francisco, CA, United States	Copy number variation analysis by next-generation sequencing enhances molecular diagnostic yield of lysosomal diseases

8:45 AM	**Lynda E. Polgreen** Harbor-UCLA/LABioMed Torrance, CA, United States	Open-label, single arm, pilot study of intravenous laronidase following allogeneic transplantation for Hurler syndrome
9:00 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MI, United States	Final results of the first-in-human open-label study of intravenous SBC-103 in children with mucopolysaccharidosis type IIIB
9:15 AM	**Nicole Muschol** University Medical Center Hamburg-Eppendorf Hamburg, Germany	ICV-administered BMN 250 (NAGLU-IGF2) is well tolerated and reduces heparan sulfate accumulation in the CNS of subjects with Sanfilippo syndrome type B (MPS IIIB)
9:30 AM	**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: two year results from an ongoing multicenter extension study
9:45 AM	**Break**	
10:15 AM	**Joseph Muenzer** University of North Carolina at Chapel Hill Chapel Hill, NC, United States	Efficacy and safety of intrathecal idursulfase in pediatric patients with mucopolysaccharidosis type II and early cognitive impairment: design and methods of a controlled, randomized, phase II/III multicenter study
10:30 AM	**Roberto Giugliani** Hospital de Clínicas de Porto Alegre, Universidade Federal do Rio Grande do Sul Porto Alegre, Brazil	Safety and clinical efficacy of AGT-181, a brain penetrating human insulin receptor antibody-iduronidase fusion protein, in a 26-week study with pediatric patients with mucopolysaccharidosis type I
10:45 AM	**Caroline Sevin** Bicêtre Hospital Le Kremlin-Bicetre, France	Intracerebral gene therapy in children with metachromatic leukodystrophy: results of a phase I/II trial
11:00 AM	**Kevin M. Flanigan** Nationwide Children's Hospital The Ohio State University Columbus, OH, United States	A phase 1/2 clinical trial of systemic gene transfer of scAAV9.U1a.HSGSH for MPS IIIA: safety, tolerability, and preliminary evidence of biopotency
11:15 AM	**Sophie Olivier** Lysogene Paris, France	Five years of clinical data in a direct to CNS gene therapy trial to address the severe lethal neurological manifestations of MPS IIIA
11:30 AM	**Torayuki Okuyama** National Center for Child Health and Development Tokyo, Japan	Novel blood-brain barrier delivery system to treat CNS in MPS II - first clinical trial by anti-transferrin receptor antibody fused enzyme therapy
11:45 AM	**Lunch – On Own or Satellite Symposia**	

Clinical Trials II — Co-Chairs: Uma Ramaswami & Stephen Groft

1:00 PM	**Raymond Wang** Children's Hospital of Orange County Orange, CA, United States	Sustained efficacy and safety of vestronidase alfa (rhGUS) enzyme replacement therapy in patients with MPS VII
1:15 PM	**Franklin K. Johnson** Amicus Therapeutics, Inc. Cranbury, NJ, United States	First-in-human preliminary pharmacokinetic data on a novel recombinant acid α-glucosidase, ATB200, co-administered with the pharmacological chaperone, AT2221, in patients with late-onset Pompe disease
1:30 PM	**Paul Harmatz** UCSF Benioff Children's Hospital Oakland Oakland, CA, United States	Global treatment responder analysis demonstrates clinically relevant effect of velmanase alfa long term enzyme replacement therapy for alpha mannosidosis, in a phase III randomized placebo controlled trial
1:45 PM	**Caren Swift** Baylor Research Institute Dallas, TX, United States	Ten years of migalastat treatment in a patient with Fabry disease: a case report
2:00 PM	**Julia B. Hennermann** University Medical Center Mainz Mainz, Germany	Pharmacokinetics, pharmacodynamics, and safety of moss agalactosidase A in patients with Fabry disease
2:15 PM	**Derralynn Hughes** Royal Free Hospital University College London London, United Kingdom	Clinical outcomes in Morquio syndrome type A treated with elosulfase alfa: results from the managed access agreement in England
2:30 PM	**Simon A. Jones** Central Manchester University Hospitals NHS Foundation Trust Manchester, United Kingdom	Effect of sebelipase alfa on survival to 3 years of age and liver function in infants with rapidly progressive lysosomal acid lipase deficiency: results from two studies
2:45 PM	**Break**	
3:15 PM	**Livia D. Paskulin** Universidade Federal do Rio Grande do Sul Porto Alegre, Brazil	Taliglucerase-alpha and Gaucher disease type 1: a five-year follow-up
3:30 PM	**David J. Kuter** Massachusetts General Hospital Boston, MA, United States	Open-label expanded access study of taliglucerase alfa in patients with Gaucher disease requiring enzyme replacement therapy
3:45 PM	**Joel Charrow** Northwestern University Feinberg School of Medicine Chicago, IL, United States	Long-term stability in randomized and non-randomized patients in the phase 3 randomized, double-blind EDGE trial of once- versus twice-daily dosing of eliglustat in patients with Gaucher disease type 1

4:00 PM	**Heather A. Lau** New York University New York, NY, United States	Long-term treatment response based on severity of Gaucher disease type 1 at baseline after 8 years of treatment with oral eliglustat: final efficacy and safety results from a phase 2 clinical trial in treatment-naïve adult patients
4:15 PM	**M. Judith Peterschmitt** Sanofi Genzyme Cambridge, MA, United States	Evaluation of glucosyl ceramide synthase (GCS) inhibition for GBA-associated Parkinson's disease
4:30 PM	**Networking Reception in Foyer**	