Sunday, February 7, 2021

10:00 AM **2:00 PM**	**Emerging Trends:** **State-of-the-Art for Expert**	*(Registration required)*
10:00 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	Introduction and Overview of Course
10:05 AM	**Andrew P. Lieberman** Michigan Medicine University of Michigan Ann Arbor, MI United States	Lysosomal Function and Pathogenesis
10:25 AM	**Marc C. Patterson** Mayo Clinic Rochester, MN United States	Clinical Features
10:45 AM	**Amy Gaviglio** US Centers for Disease Control and Prevention (CDC) Atlanta, GA United States	Newborn Screening
11:05 AM	**Break**	
11:30 AM	**Jeanine R. Jarnes** University of Minnesota Minneapolis, MN United States	Therapies
11:50 AM	**Patroula Smpokou** Division of Rare Diseases & Medical Genetics, Office of New Drugs US Food & Drug Administration (FDA) Silver Spring, MD United States	FDA Regulatory Review
12:10 PM	**Cara O'Neill** Cure Sanfilippo Foundation Columbia, SC United States	Patient Advocate's Perspective on Experimental Therapies
12:30 PM	**Break**	
1:00 PM	**Marc C. Patterson** Mayo Clinic Rochester, MN United States	Case Studies
1:20 PM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	SARS-CoV-2 Virus
1:40 PM	**Heather Lau** New York University School of Medicine New York, NY United States	COVID-19: Clinical Impact and Management
1:59 PM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	Closing Remarks
2:00 PM	**Adjourn**	

Monday, February 8, 2021
Basic Science **Moderators: Brian Bigger & Dao Pan**

8:45 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	Welcome & Announcements Presentation of 2021 Roscoe O. Brady Award for Innovation and Accomplishment to Ellen Sidransky
9:00 AM	**Ellen Sidransky** National Human Genome Research Institute at National Institutes of Health (NIH) Bethesda, MD United States	Innovation Award Speaker Presentation
9:30 AM	**Wei Zhu** University of Minnesota Minneapolis, MN United States	Functional connectivity alterations in MPS I mouse brain at the laminar level revealed by resting-state fMRI **2021 Young Investigator Award Recipient*
9:42 AM	**Shih-hsin Kan** Children's Hospital of Orange County Orange, CA United States	iPSC-derived human neural stem cells engraft in the brains of immunocompromised MPS I mice
9:54 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	Immunogenicity, genotoxicity, and efficacy of PS gene editing in treating MPS I mice
10:06 AM	**Lalitha Belur** University of Minnesota Minneapolis, MN United States	Comparative systemic and neurologic effectiveness of intravenous and intrathecal AAV9 delivered individually or combined in a murine model of mucopolysaccharidosis type I
10:18 AM	**Live Moderated Q&A**	*Wei Zhu, Shih-hsin Kan, Chester B. Whitley, and Lalitha Belur*
10:30 AM	**Break & Exhibits**	
11:00 AM	**Tyler Harm** Iowa State University Ames, IA United States	Treatment with pentosan polysulfate improves neuropathological measures in the canine model of MPS IIIB **2021 Young Investigator Award Recipient*
11:12 AM	**Laura López de Frutos** Instituto de Investigación Sanitaria Aragón (IIS Aragón) Zaragoza, Spain	Testing new biomarkers for lysosomal diseases
11:24 AM	**Ying Sun** Cincinnati Children's Hospital Medical Center Cincinnati, OH United States	Novel mechanism of SRT and ERT on recovering the function of mitochondrial and autophagy-lysosomal pathway in Gaucher disease neuronal cell model
11:36 AM	**Paula Rozenfeld** Universidad Nacional de La Plata-CONICET, IIFP La Plata, Argentina	Gaucher disease mesenchymal stem cells showed reduced osteogenesis and increased osteoclastogenesis and adipogenesis
11:48 AM	**Live Moderated Q&A**	*Tyler Harm, Laura López de Frutos, Yin Sun, and Paula Rozenfeld*
12:00 PM	**Break, Exhibits and Satellite Symposia**	

1:00 PM	**Allen Seylani** National Institutes of Health (NIH) Bethesda, MD United States	Novel regulatory function of GCN5L1 in lysosomal tubulation and biogenesis **2021 Young Investigator Award Recipient*
1:12 PM	**Behzad Najafian** University of Washington Seattle, WA United States	Direct intercellular cross-correction of α-galactosidase-A deficiency in Fabry disease podocytes through tunneling nanotubes in a mixed cell culture model
1:24 PM	**Dau-Ming Niu** Taipei Veterans General Hospital Taipei, Taiwan	Development of a gene therapy for Fabry disease using adeno-associated viral vector mediated gene editing
1:36 PM	**Stephanie Newman** Western University London, ON Canada	AAV9-hARSA decreases sulfatide accumulation in the aged ARSA-/- mouse model for metachromatic leukodystrophy **2021 Young Investigator Award Recipient*
1:48 PM	**Live Moderated Q&A**	*Allen Seylani, Behzad Najafian, Dau-Ming Niu, and Stephanie Newman*
2:00 PM	**Break & Exhibits**	
2:30 PM	**Poster Session**	
3:30 PM	**Break & Exhibits & Networking**	
4:00 PM	**Satellite Symposia**	

Tuesday, February 9, 2021

Translational Research **Moderators: PJ Brooks & Jill Morris**

9:00 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	2021 Patient Advocate Leader (PAL) Award Announcement and Presentation to Terri L. Klein
9:15 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	2021 Young Investigator Awards Announcement and Presentation
9:30 AM	**Xin Chen** University of Texas Southwestern Medical Center Dallas, TX United States	Preclinical results in rodents strongly support clinical evaluation of scAAV9/MFSD8 as a potential gene therapy for CLN7 patients
9:42 AM	**Rachel Bailey** University of Texas Southwestern Medical Center Dallas, TX United States	Preclinical studies to support the intrathecal delivery of scAAV9/SUMF1 as a gene replacement therapy for multiple sulfatase deficiency
9:54 AM	**Li Ou** University of Minnesota Minneapolis, MN United States	PS gene editing with a novel HEXO construct to treat both Tay-Sachs and Sandhoff diseases
10:06 AM	**Su Jin Choi** Duke University Durham, NC United States	Immunosuppression with bortezomib and anti-CD20 mAb is effective in reducing neutralizing antibodies to allow repeated AAV administration in mice **2021 Young Investigator Award Recipient*
10:18 AM	**Live Moderated Q&A**	*Xin Chen, Rachel Bailey, Li Ou, and Su Jin Choi*
10:30 AM	**Break & Exhibits**	
11:00 AM	**Troy Lund** University of Minnesota Minneapolis, MN United States	Biochemical predictors of neurocognitive outcomes in Hurler syndrome
11:12 AM	**Jane Kinsella** Royal Manchester Children's Hospital Manchester, United Kingdom	Ex-vivo autologous stem cell gene therapy clinical trial for mucopolysaccharidosis type IIIA: Update on phase I/II clinical trial **2021 Young Investigator Award Recipient*
11:24 AM	**Bernhard Gentner** San Raffaele Telethon Institute for Gene Therapy Milano, Italy	Ex vivo hematopoietic stem cell gene therapy for mucopolysaccharidosis type I (Hurler syndrome)
11:36 AM	**Bryan Pukenas** University of Pennsylvania Philadelphia, PA United States	Intracisternal administration of AAV9 gene therapies to target the central nervous system
11:48 AM	**Live Moderated Q&A**	*Troy Lund, Jane Kinsella, Bernhard Gentner, and Bryan Pukenas*
12:00 PM	**Break, Exhibits and Satellite Symposia**	

1:00 PM	**John Day** Stanford University Stanford, CA United States	A phase I/II open-label gene replacement clinical study for late onset Pompe Disease
1:12 PM	**Torayuki Okuyama** National Center for Child Health and Development Tokyo, Japan	Prevention of cognitive decline in patients with neuronopathic mucopolysaccharidosis type II treated by intracerebroventricular enzyme replacement therapy: 100-week results of an open-label phase 1/2 study
1:24 PM	**Julian Raiman** Birmingham Women and Children's NHS Foundation Trust Birmingham, United Kingdom	Update on safety and efficacy results for phase I/II trial of hydroxypropyl betacyclodextrin (HPâCD) administered intravenously in patients with Niemann-Pick disease type C1
1:36 PM	**Calogera Simonaro** Icahn School of Medicine at Mount Sinai New York, NY United States	Modulation of the endocannabinoid receptor CB2 as a novel treatment for the lysosomal diseases
1:48 PM	**Live Moderated Q&A**	*John Day, Torayuki Okuyama, Julian Raiman, and Calogera Simonaro*
2:00 PM	**Break & Exhibits**	
2:30 PM	**Poster Session**	
3:30 PM	**Break & Exhibits & Networking**	
4:00 PM	**Satellite Symposia**	

Wednesday, February 10, 2021

COVID-19 and Clinical Trials — **Moderators: Maurizio Scarpa & Patroula Smpokou**

8:55 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	Welcome and Keynote Speaker Introduction
9:00 AM	**Michael T. Osterholm** University of Minnesota Minneapolis, MN United States	*Keynote Address:* The COVID Pandemic: The Evolving Reality
9:30 AM	**Live Moderated Q&A**	*Michael T. Osterholm*
9:45 AM	**Heather Lau** NYU School of Medicine New York, NY United States	Impact of SARS-CoV-2 on patients with lysosomal diseases in a major NYC hospital system
9:57 AM	**Matheus Wilke** Hospital de Clinicas de Porto Alegre Porto Alegre, Brazil	Informing patients with rare diseases about COVID-19: Creation of the “Beto and the Coronavirus” booklet
10:09 AM	**Siddhee Sahasrabudhe** University of Minnesota Minneapolis, MN United States	Modeling potential interactions between oral Gaucher disease treatment and investigational COVID-19 therapies
10:21 AM	**Live Moderated Q&A**	*Heather Lau, Matheus Wilke, and Siddhee Sahasrabudhe*
10:30 AM	**Break & Exhibits**	
11:00 AM	**George Diaz** Mount Sinai School of Medicine New York, NY United States	Children treated with olipudase alfa for chronic acid sphingomyelinase deficiency show meaningful improvement on clinically relevant outcomes and an overall favorable safety profile: 1-year results of the ASCEND-Peds trial
11:12 AM	**Nicole Muschol** UKE - Universitätsklinikum Hamburg-Eppendorf Hamburg, Germany	Tralesinidase alfa (AX 250) enzyme replacement therapy for Sanfilippo syndrome type B
11:24 AM	**Emily de los Reyes** Nationwide Children’s Hospital Columbus, OH United States	Single-dose AAV9-CLN6 gene transfer slows the decline in motor and language function in variant late infantile neuronal ceroid lipofuscinosis 6: Interim results from phase 1/2 trial
11:36 AM	**Francesca Fumagalli** San Raffaele Telethon Institute for Gene Therapy (SR-TIGET), IRCCS San Raffaele Scientific Institute Milan, Italy	Lentiviral hematopoietic stem and progenitor cell gene therapy provides durable clinical benefit in early-symptomatic early-juvenile metachromatic leukodystrophy
11:48 AM	**Live Moderated Q&A**	*George Diaz, Nicole Muschol, Emily de los Reyes, and Francesca Fumagalli*
12:00 PM	**Break, Exhibits and Satellite Symposia**	

1:00 PM	**Ankit Desai** Duke University Medical Center Durham, NC United States	Transforming the clinical outcomes in CRIM-negative infantile Pompe disease identified via newborn screening: The benefits of early treatment with enzyme replacement therapy and immune tolerance induction
1:12 PM	**Melissa Wasserstein** Children's Hospital at Montefiore, Albert Einstein College of Medicine Bronx, NY United States	Adults with chronic acid sphingomyelinase deficiency show significant visceral, pulmonary, and hematologic improvements after enzyme replacement therapy with olipudase-alfa: 1-year results of the ASCEND placebo-controlled trial
1:24 PM	**Ales Linhart** General University Hospital and Charles University Prague, Czech Republic	Switching from agalsidase alfa to pegunigalsidase alfa to treat patients with Fabry disease: 1 year of treatment data from BRIDGE, a phase 3 open-label study
1:36 PM	**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
1:48 PM	**Live Moderated Q&A**	*Ankit Desai, Melissa Wasserstein, Ales Linhart, and Raphael Schiffmann*
2:00 PM	**Break & Exhibits**	
2:30 PM	**Poster Session**	
3:30 PM	**Break & Exhibits & Networking**	
4:00 PM	**Satellite Symposia**	

Thursday, February 11, 2021

Contemporary Forum **Moderators: Mark Sands & Uma Ramaswami**

8:45 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN United States	Welcome and Keynote Address Introduction
8:50 AM	**Peter Marks** Center for Biologics Evaluation and Research US Food & Drug Administration (FDA) Silver Spring, MD, United States	*Keynote Address:* Trailblazing a Regulatory Framework for Individualized Therapies
9:20 AM	**Live Moderated Q&A**	*Peter Marks*

The following session is not available for CME/CE accreditation (unless noted otherwise); CEU credits for GCs may apply.

9:30 AM	**Jeffrey Alexander** Spark Therapeutics, Inc Philadelphia, PA United States	IdeS: An enabling technology to overcome the limitation of neutralizing antibodies to AAV gene therapy
9:42 AM	**Marie-Laure Nevoret** REGENXBIO Rockville, MD United States	RGX-121 gene therapy for severe mucopolysaccharidosis type II (MPS II): Interim results of an ongoing first in human trial
9:54 AM	**Drew Tietz** Sigilon Therapeutics Cambridge, MA United States	SIG-018: Novel encapsulated non-viral cell-based therapy for MPS II
10:06 AM	**Annie Arguello** Denali Therapeutics, Inc. South San Francisco, CA United States	Iduronate-2-sulfatase transport vehicle rescues neurobehavioral and skeletal phenotypes in a mouse model of mucopolysaccharidosis type II
10:18 AM	**Live Moderated Q&A**	*Jeffery Alexander, Marie-Laure Nevoret, Drew Tietz, and Annie Arguello*
10:30 AM	**Break & Exhibits**	
11:00 AM	**Niek van Til** AVROBIO Cambridge, MA United States	Long-term hematopoietic stem cell gene therapy corrects neuromuscular manifestations in preclinical study of Pompe mice
11:12 AM	**Rachel Botham** Codexis Redwood City, CA United States	Engineering α-glucosidase to improve protein stability and cellular uptake for the potential treatment of Pompe disease
11:24 AM	**Dustin Armstrong** Valerion Therapeutics Concord, MA United States	VAL-1221: Treating Pompe disease via enhanced glycogen-targeting
11:36 AM	**Romuald Corbau** Freeline Stevenage, United Kingdom	FLT201: An AAV-mediated gene therapy for type 1 Gaucher disease designed to target difficult to reach tissues
11:48 AM	**Live Moderated Q&A**	*Niek van Til, Rachel Botham, Dustin Armstrong, and Romuald Carbau*

12:00 PM	**Break, Exhibits and Satellite Symposia**	
1:00 PM	**John Jefferies** University of Tennessee Health Science Center Memphis, TN United States	Utilization of artificial intelligence to identify undiagnosed Fabry disease patients: Development of a validated machine learning model
1:12 PM	**Deborah Marsden** Ultragenyx Pharmaceutical Inc. Novato, CA United States	Significant unmet need in infants with mucopolysaccharidosis type VII and non-immune hydrops fetalis: A summary of cases
1:24 PM	**Cristina Baricordi** AVROBIO, Inc Cambridge, MA United States	Analysis of genetically engineered stem cell product and follow up of gene therapy patients through high-throughput single cell technologies
1:36 PM	**Miganush Stepanians** PROMETRIKA, LLC Cambridge, MA United States	A survey of statistical study design and analysis methods for rare disease development programs
1:48 PM	**Live Moderated Q&A**	*John Jefferies, Deborah Marsden, Cristina Baricordi, and Miganush Stepanians*
2:00 PM	**Break & Exhibits**	
2:30 PM	**Poster Session**	
3:30 PM	**Break & Exhibits & Networking**	
4:00 PM	**Satellite Symposia**	

Friday, February 12, 2021

Late-Breaking Science **Moderators: Roberto Giugliani & Elizabeth Braunlin**

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

9:30 AM	**Cathal S. Mahon** Denali Therapeutics Inc. South San Francisco, CA United States	Molecular architecture determines brain delivery of transferrin receptor targeted iduronate 2 sulfatase in a mouse model of mucopolysaccharidosis type II
9:42 AM	**Jennifer Clarke Matthews** Sanofi Framingham, MA United States	Murine models of lysosomal diseases exhibit differences in brain protein aggregation and neuroinflammation
9:54 AM	**Miles C. Smith** University of Minnesota Minneapolis, MN United States	Ex vivo lentiviral transduction of hematopoietic stem cells in mucopolysaccharidosis type II (MPS II) mice achieves high levels of systemic iduronate-2-sulfatase (IDS) enzyme activity and normalization of glycosaminoglycans (GAGs)
10:06 AM	**Juana I. Navarrete** Hospital Central Sur PEMEX Mexico City, Mexico	Use of biomarkers to follow up positive lysosomal diseases in newborn screening
10:18 AM	**Live Moderated Q&A**	*Cathal Mahon, Jennifer Clarke Matthews, Miles Smith and Juana Navarrete*
10:30 AM	**Break & Exhibits**	
11:00 AM	**Stephanie Cherqui** University of California, San Diego La Jolla, CA United States	Hematopoietic stem cell gene therapy for cystinosis: Updated results from a phase I/II clinical trial
11:12 AM	**Priya Kishnani** Duke University Medical Center Durham, NC United States	Efficacy and safety results of the avalglucosidase alfa phase 3 COMET trial in late-onset Pompe disease patients
11:24 AM	**Mark Thomas** Royal Perth Hospital Perth, Australia	AVR-RD-01, an investigational lentiviral gene therapy for Fabry disease: Overview of clinical data from phase 1 and phase 2 studies
11:36 AM	**Kevin M. Flanigan** Nationwide Children's Hospital Columbus, OH United States	Updated results of Transpher A, a multicenter, single-dose, phase 1/2 clinical trial of ABO-102 gene therapy for Sanfilippo syndrome type A (MPS IIIA)
11:48 AM	**Live Moderated Q&A**	*Stephanie Cherqui, Priya Kishnani, Mark Thomas, and Kevin Flanigan*
12:00 PM	**Break, Exhibits and Satellite Symposia**	
1:00 PM	**Michaël Hocquemiller** LYSOGENE Neuilly sur seine, France	CNS-specific reductions of heparan sulfate and secondary storage biomarkers in Sanfilippo syndrome type A patients treated with the investigational gene therapy LYS-SAF302
1:12 PM	**Maria J. de Castro** Hospital Clínico Universitario de Santiago de Compostela Santiago de Compostela, Spain	Updated results of Transpher B, a multicenter, single-dose, phase 1/2 clinical trial of ABO-101 gene therapy for Sanfilippo syndrome type B (MPS IIIB)

1:24 PM	**Anna I. Bakardjiev** Denali Therapeutics South San Francisco, CA United States	Intravenous ETV:IDS (DNL310) significantly reduces cerebrospinal fluid heparan sulfate in an open label Ph1/2 study in MPS II patients
1:36 PM	**Elizabeth M. Berry-Kravis** Rush University Medical Center Chicago, IL United States	Evidence for long-term efficacy of intrathecal adrabetadex for the treatment of neurological decline in patients with Niemann-Pick disease, type C1
1:48 PM	**Live Moderated Q&A**	*Michaël Hocquemiller, Maria de Castro, Anna Bakardjiev, and Elizabeth Berry-Kravis*
2:00 PM	**Break & Exhibits**	
2:30 PM	**Poster Session**	
3:30 PM	**Virtual Networking Event**	