WORLD*Symposium*™ 2024 Preliminary Program*
Platform Presenters

Sunday, February 4, 2024
The Patient Voice: Is Anyone Listening? and Be the Catalyst Event

4:00 PM	**The Patient Voice: Is Anyone Listening?**		
6:00 PM	**Be the Catalyst Event**		**Group Photo Schedule**
		6:15 PM	All first time attendees
		6:25 PM	2024 Young Investigator Award photo, followed by all past and present Young Investigator Award recipients
		6:35 PM	2024 Patient Advocate Leader (PAL) Award recipient photo followed by all past and present recipients
		6:45 PM	2024 Roscoe O. Brady Award recipients photo followed by all past and present recipients
		6:55 PM	Genetic Counselors
		7:05 PM	Patients and Patient Advocates
		7:15 PM	2024 WORLD*Symposium* Planning and Organizing Committee

Basic Science, Translational Research, and Clinical Application Sessions are available for CME/CNE/CEU credits. Contemporary Forum and Late-Breaking Science Sessions are not available for CME/CNE credits, however CEUs for GCs may apply.

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Platform Presenters

Monday, February 5, 2024: Basic Science
Co-Chairs: Lalitha Belur, Greg Grabowski, Michael Przybilla

6:15 AM	**Satellite Symposia**	
7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Announcements Presentation of 2024 Roscoe O. Brady Award to Elsa Shapiro
	Elsa Shapiro University of Minnesota Shapiro Neuropsychology Consulting, LLC Portland, OR, United States	Roscoe O. Brady Award Presentation: *The Contribution of Neuropsychology to Understanding Lysosomal Diseases*
8:00 AM	**Oriana Mandolfo** University of Manchester Manchester, United Kingdom	Systemic inflammation leads to neuronal loss and exacerbates behavioural deficits in a mouse model of MPS IIIA
	Helen Parker University of Manchester Manchester, United Kingdom	The meningeal immune landscape in mucopolysaccharidosis type IIIA
	Jacqueline Hernandez The Lundquist Institute at Harbor-UCLA Torrance, CA, United States	Defects in cell polarity of mucopolysaccharidosis type III (MPS III) forebrain neurons
	Travis Moore University of Montreal Montreal, QC, Canada	A small molecule drug, AVP6, rescues synaptic deficits in human iPSC-derived neurons across the mucopolysaccharidosis type III spectrum
	Moderated Q&A	*Mandolfo, Parker, Hernandez, Moore*
9:00 AM	**Yuki Shiro** Tokushima University Tokushima, Japan	CTSD integrity in the endoplasmic reticulum is required for CLN6's anti-aggregate activity **2024 Young Investigator Award Recipient*
	Ching-Chieh Chou Stanford University Stanford, CA, United States	Human transdifferentiated neurons reveal lysosomal repair deficits in Alzheimer's disease
	Salma Begum Columbia University Irving Medical Center New York, NY, United States	The psychosine and galactosylceramide brain spatial distribution and its correlation with neuropathogenic processes
	Marya S. Sabir National Institutes of Health Bethesda, MD, United States	Advancing free sialic acid storage (FSASD) disorder disease modeling: Insights from iPSC-derived neural cell types
	Moderated Q&A	*Shiro, Chou, Begum, Sabir*
10:00 AM	**Break**	
10:30 AM	**Caitlin Calhoun** CHOC Children's Research Institute Orange, CA, United States	Functional efficacy of transplanted, iPSC-derived, human neural stem cells in the brains of MPS I mice
	Bryce Binstadt University of Minnesota Minneapolis, MN, United States	Identification of inflammatory cells in dilated ascending aortas of IDUA-deficient (MPS I) mice
	Esteban Alberto Gonzalez Universidade Federal do Rio Grande do Sul Porto Alegre, Brazil	Losartan treatment in mucopolysaccharidosis type I mice: Beneficial effects on aortic structure and pathways insights **2024 Young Investigator Award Recipient*

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Monday, February 5, 2024: Basic Science *(cont.)*
Co-Chairs: Lalitha Belur, Greg Grabowski, Michael Przybilla

	Gabrielle Dineck Iop Hospital de Clínicas de Porto Alegre Porto Alegre, Brazil	Biomarker distribution in tissues of MPS I mice: Measurement of disease-specific oligosaccharides by LC-MS/MS
	Moderated Q&A	*Calhoun, Binstadt, Gonzalez, Iop*
11:30 AM	**Break and Satellite Symposia**	
1:00 PM	**Irene Serrano Gonzalo** Fundación Española Para el Estudio y Terapéutica de la Enfermedad de Gaucher y Otras Lisosomales Zaragoza, Spain	Bone involvement in Gaucher disease: Can miRNAs determine or predict the severity degree? **2024 Young Investigator Award Recipient*
	Francyne Kubaski Greenwood Genetic Center Greenwood, SC, United States	Sensitivity and specificity of serum oligosaccharide analysis for the diagnosis and treatment monitoring of patients with alpha-mannosidosis
	Maria Fuller SA Pathology North Adelaide, Australia	A multiplex lipid platform improves the laboratory diagnosis of the sphingolipidoses
	Sarah Young Duke University School of Medicine Durham, NC, United States	Measurement of glycosaminoglycans in the amniotic fluid of fetuses with mucopolysaccharidoses treated in a phase I clinical trial by in utero enzyme replacement therapy
	Moderated Q&A	*Serrano Gonzalo, Kubaski, Fuller, Young*
2:00 PM	**Jerry Fuad Harb** Children's Hospital of Orange County Orange, CA, United States	Exploring Pompe disease: Insights into the natural history of novel *Gaa*$^{c.1826dupA}$ knock-in murine model
	Chloe L. Christensen Children's Hospital of Orange County Orange, CA, United States	Restoration of acid-alpha glucosidase expression and function through efficient adenine base editing of Pompe disease variants
	Shih-hsin Kan CHOC Children's Research Institute Orange, CA, United States	Improvement of hypertrophic cardiomyopathy in *Gaa*$^{c.1826dupA}$ knock-in murine model with neonatal gene therapy
	Patricia Lam Abigail Wexner Research Institute at Nationwide Children's Hospital Columbus, OH, United States	Liver-directed AAV gene therapy corrects disease symptoms in a murine model of lysosomal acid lipase deficiency
	Moderated Q&A	*Harb, Christensen, Kan, Lam*
3:00 PM	**Poster Session**	**Exhibit Hall**
5:15 PM	**Speed Mentoring Event**	
6:15 AM	**Satellite Symposia**	
7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome & Announcements Presentation of 2024 Patient Advocate Leader (PAL) Award Announcement to Alan Finglas and 2024 Young Investigator Awards Presentation

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Tuesday, February 6, 2024: Translational Research
Co-Chairs: Amy Gaviglio, Francyne Kubaski, Dan Tagle

8:00 AM	**Jillian Gallagher** University of Massachusetts Chan Medical School Worcester, MA, United States	Adeno-associated viral gene therapy for sialidosis using small and large animal models
	Emilie Audouard Paris Brain Institute Paris, France	Intravenous gene therapy using AAVPHP.eB for metachromatic leukodystrophy
	Patricia I. Dickson Washington University St. Louis St. Louis, MO, United States	Gene therapy with AAV-S1S3 improves disease in mucolipidosis type II mice
	Kim M. Hemsley Flinders University Adelaide, SA, Australia	Superior outcomes in neuroretina following IV versus intra-CSF AAV9 gene replacement in mice with MPS IIIA
	Moderated Q&A	*Gallagher, Audouard, Dickson, Hemsley*
9:00 AM	**Elena Gaia Banchi** Paris Brain Institute Paris, France	Development and validation of a novel AAV gene therapy for mucopolysaccharidosis type IIIB in large animal
	Betul Celik University of Delaware Wilmington, DE, United States	*Ex vivo* lentiviral gene therapy for mucopolysaccharidosis type IVA **2024 Young Investigator Award Recipient*
	Vi Pham University of Pennsylvania Philadelphia, PA, United States	Single vs. dual transgene *ex vivo* gene therapy for multiple sulfatase deficiency **2024 Young Investigator Award Recipient*
	Rafael A. Badell-Grau University of California, San Diego La Jolla, CA, United States	Gene modified hematopoietic stem cell transplantation for mucopolysaccharidosis type IIIC
	Moderated Q&A	*Banchi, Celik, Pham, Badell-Grau*
10:00 AM	**Break & Exhibits**	
10:30 AM	**Bartholomew A. Pederson** Ball State University Muncie, IN, United States	A novel siRNA targeting and delivery platform inhibits glycogen synthesis and reduces glycogen levels in skeletal and cardiac muscle in a mouse model of Pompe disease
	Luisa Natalia Pimentel Vera Stanford University Stanford, CA, United States	Genome-edited hematopoietic stem cells as a curative approach for Gaucher disease type 1 **2024 Young Investigator Award Recipient*
	Edina Poletto Stanford University Stanford, CA, United States	Clinical development of autologous genome-edited hematopoietic stem cells to treat mucopolysaccharidosis type I **2024 Young Investigator Award Recipient*
	Allisandra Rha Children's Hospital of Orange County Orange, CA, United States	Prime editing corrects the *Gaa*$^{c.1935C>}$ A pathogenic variant in infantile-onset Pompe disease mouse myoblasts
	Moderated Q&A	*Pederson, Pimentel Vera, Poletto, Rha*
11:30 AM	**Break, Exhibits & Satellite Symposia**	

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Tuesday, February 6, 2024: Translational Research *(cont.)* and Robert J. Gorlin Symposium
Co-Chairs: Amy Gaviglio, Francyne Kubaski, Dan Tagle

1:00 PM	**Sandra Vranic** University of Manchester Manchester, United Kingdom	Graphene flakes for enhanced delivery of the enzyme to the lysosomes of patient-derived fibroblasts: Bio-persistence and kinetics of substrate degradation
	Tomas Baldwin University College London London, United Kingdom	The development and application of a rapid and more informative test for autoantibodies to enzyme replacement therapies in Fabry disease **2024 Young Investigator Award Recipient*
	Sarah Hurt Washington University St. Louis Saint Louis, MO, United States	Anti-IDUA IgG alters cortical bone structure of mucopolysaccaridosis type I mice treated with intravenous enzyme replacement therapy **2024 Young Investigator Award Recipient*
	Lena Marie Westermann University Medical Center Hamburg-Eppendorf Hamburg, Germany	Analysis of drug-specific antibody response against cerliponase alfa in CLN2 patients by applying a novel two-step assay **2024 Young Investigator Award Recipient*
	Moderated Q&A	*Vranic, Baldwin, Hurt, Westermann*
2:00 PM	**Logan M. Glasstetter** National Institutes of Health Bethesda, MD, United States	A novel quantitative high-throughput screening assay identifies small-molecule therapeutic candidates for Gaucher and Parkinson disease
	Dietrich Matern Mayo Clinic Rochester, MN, United States	Newborn screening for Krabbe disease: Status quo and recommendations for improvements
	Delaney E. Wilton University of Minnesota Minneapolis, MN, United States	Is time growth? The impact of early initiation and duration of enzyme replacement therapy on growth in Hurler syndrome
	Fabiano O. Poswar Hospital de Clínicas de Porto Alegre Porto Alegre, Brazil	Safety and tolerability of losartan for the treatment of cardiovascular manifestations in mucopolysaccharidoses types IVA and VI
	Moderated Q&A	*Glasstetter, Matern, Wilton, Poswar*
3:00 PM	**Poster Session**	**Exhibit Hall**
5:15 PM	**Robert J. Gorlin Symposium**	Beyond the Blood Brain Barrier: Strategies for Treating the CNS

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Wednesday, February 7, 2024: Clinical Applications
Co-Chairs: Marc Patterson, Lynda Polgreen, Filippo Vairo

6:15 AM	**Satellite Symposia**	
7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and Keynote Speaker Introduction
	Peter Marks Center for Biologics Evaluation and Research US Food & Drug Administration (FDA) Silver Spring, MD, United States	*Keynote Address:* Accelerating the Pace of Progress in Gene Therapy
8:00 AM	**Robert J. Hopkin** Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease: Updated results from an ongoing phase 1/2 study (STAAR)
	Simon Jones St. Mary's Hospital Manchester, United Kingdom	Clinical outcomes and sustained biochemical engraftment following *ex-vivo* autologous stem cell gene therapy for mucopolysaccharidosis type IIIA
	Francesca Fumagalli IRCCS San Raffaele Hospital Milan, Italy	Atidarsagene autotemcel (autologous hematopoietic stem cell gene therapy) preserves cognitive and motor development in early-onset metachromatic leukodystrophy with up to 12 years follow-up
	Paul Harmatz UCSF Benioff Children's Hospital Oakland, CA, United States	CAMPSIITE™ phase I/II/III: An interim clinical study update of RGX-121, an investigational gene therapy for the treatment of neuronopathic mucopolysaccharidosis type II (MPS II)
	Moderated Q&A	*Hopkin, Jones, Fumagalli, Harmatz*
9:00 AM	**Angela Schulz** University Medical Center Hamburg-Eppendorf Hamburg, Germany	Cerliponase alfa for the treatment of CLN2 disease in a patient cohort including children under 3 years of age
	Joseph Muenzer University of North Carolina Chapel Hill School of Medicine Chapel Hill, NC, United States	Interim analysis of a phase 1/2 study of weekly intravenous DNL310 (brain-penetrant enzyme replacement therapy) in mucopolysaccharidosis type II
	Yoshikatsu Eto Institute of Neurological Disease Kawasaki City, Japan	Integrated long-term efficacy and safety data on enzyme replacement therapy with pabinafusp alfa for neuronopathic mucopolysaccharidosis type II (MPS II): Updated clinical data from Japan and Brazil
	Nathalie Guffon Hôpital Femme Mère Enfant Lyon, France	Longterm efficacy of velmanase alfa treatment in patients with alphamannosidosis: Pooled data from two extension studies (up to 12 years of therapy)
	Moderated Q&A	*Schulz, Muenzer, Eto, Guffon*
10:00 AM	**Break & Exhibits**	
10:30 AM	**Derralynn Hughes** University College London, Royal Free London NHS Foundation Trust London, United Kingdom	Results from GALILEO-1, a first-in-human clinical trial of FLT201 gene therapy in patients with Gaucher disease type 1
	Arunabha Ghosh St. Mary's Hospital Manchester, United Kingdom	Safety and preliminary efficacy of LYS-GM101 gene therapy in patients with GM1 gangliosidosis: Results of a phase I/II open-label clinical trial

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	Precilla D'Souza National Human Genome Research Institute, National Institutes of Health Bethesda, MD, United States	Intravenous delivery of AAV9-GLB1 gene therapy for GM1 gangliosidosis: An interim analysis
	Carolina Fischinger Moura de Souza Hospital de Clínicas de Porto Alegre Porto Alegre, Brazil	Interim results from the first-in-human intracisternal dosing of RGX-181 investigational AAV9 gene therapy in a child with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2)
	Moderated Q&A	*Hughes, Ghosh, D'Souza, Moura de Souza*
11:30 AM	**Break, Exhibits & Satellite Symposia**	
1:00 PM	**Erin Huggins** Duke University Durham, NC, United States	Experience with enzyme replacement therapy in children with late-onset Pompe disease diagnosed via newborn screening in the United States
	Suresh Vijay Birmingham Children's Hospital Birmingham, United Kingdom	Survival achieved in infants with rapidly progressive LAL-D via sebelipase alfa ERT: Results from the International LAL-D Registry
	Roberto Giugliani Federal University of Rio Grande do Sul Porto Alegre, Brazil	Efficacy and safety data (52-week) from a phase 1/2 trial and extension study of JR-171 (lepunafusp alfa) used in enzyme replacement therapy for patients with MPS I
	Motomichi Kosuga National Center for Child Health and Development Tokyo, Japan	Efficacy and safety of combination of HSCT & ICV ERT for neuropathic mucopolysaccharidosis type II
	Moderated Q&A	*Huggins, Vijay, Giugliani, Kosuga*
2:00 PM	**Manisha Balwani** Icahn School of Medicine at Mount Sinai New York, NY, United States	Age-specific risk of Parkinson disease and Parkinsonian syndrome in patients with Gaucher disease type 1: Real-world evidence from the International Collaborative Gaucher Group Gaucher Registry
	Caroline Aimee Hastings UCSF Benioff Children's Hospital Oakland Oakland, CA, United States	Transport®NPC: open phase 3 global trial of intravenous hydroxy-propyl-beta-cyclodextrin in patients with Niemann-Pick disease type C1 (NPC1)
	Troy Lund University of Minnesota Minneapolis, MN, United States	Changes in CSF GAG after intravenous enzyme replacement therapy
	Antonio Pisani University Federico II Naples, Italy	Clinical outcomes in patients switching from agalsidase beta to migalastat: A Fabry Registry analysis
	Moderated Q&A	*Balwani, Hastings, Lund, Pisani*
3:00 PM	**Poster Session**	**Exhibit Hall**
5:15 PM	**Satellite Symposia**	
6:15 AM	**Satellite Symposia**	
7:30 AM	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and New Treatment Awards

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Thursday, February 8, 2024: Contemporary Forum
Co-Chairs: PJ Brooks, Nishitha Pillai, Uma Ramaswami

8:00 AM	**Christina Ohnsman** REGENXBIO Inc. Rockville, MD, United States	RGX-381: Interim results from the first-in-human clinical trial of an investigational gene therapy for the treatment of ocular manifestations of CLN2 Batten disease
	Shyam Ramachandran Sanofi Waltham, MA, United States	AAV-ARSA mediated gene replacement for the treatment of metachromatic leukodystrophy
	Kathleen E. Meyer Sangamo Therapeutics Brisbane, CA, United States	A 3-month gene therapy single-dose IV administration pharmacology and safety study with ST-920 (isaralgagene civaparvovec) for Fabry disease in mice
	Michael R. DiGruccio M6P Therapeutics St. Louis, MO, United States	Hyperactive GlcNAc-1-Phosphotransferase (S1S3 PTase) dramatically increases M6P levels on lysosomal enzymes for substantially improved receptor binding and cellular uptake
	Moderated Q&A	*Ohnsman, Ramachandran, Meyer, DiGruccio*
9:00 AM	**Dongkyu Jin** Novel Pharma Inc Seoul, Republic of Korea	A phase III clinical trial of GC1111 as an enzyme replacement therapy in previously untreated mucopolysaccharidosis type II (Hunter syndrome) patients: A double-blind, randomized, active-controlled (part 1) and open-labeled, historical placebo-controlled (part 2) study
	Franklin Johnson Amicus Therapeutics, Inc. Princeton, NJ, United States	Trial in progress: An open-label study (AT1001-025) to evaluate the safety and pharmacokinetics of migalastat in patients with Fabry disease and amenable *GLA* variants and severe renal impairment or end-stage renal disease treated with hemodialysis
	Taylor Fields IntraBio Oxford, United Kingdom	Results of a phase III, randomized, placebo-controlled crossover trial with N-acetyl-L-leucine for Niemann-Pick disease type C
	Tarekegn Gerberhiwot University of Birmingham Birmingham, United Kingdom	Investigating the role of miglustat in the management of a patient with Tangier disease: An n-of-1 study with alternating periods of intervention and control
	Moderated Q&A	*Jin, Johnson, Fields, Gerberhiwot*
10:00 AM	**Break & Exhibits**	
10:30 AM	**Stuart Gaffney** Chiesi Global Rare Diseases Glasgow, United Kingdom	Medical education needs to improve diagnosis of Fabry disease in the UK
	Ashley Volz BioMarin Pharmaceutical Inc. Novato, CA, United States	Skeletal dysplasia gene panel with integrated enzyme follow-up for the diagnosis of lysosomal disorders: MPS IVA case series
	Vanessa Rangel Miller Ultragenyx Pharmaceutical Inc. Novato, CA, United States	Parallel biochemical and genetic testing informs a timely and accurate diagnosis of MPS VII: Findings from 5 years of sponsored testing programs
	Raymond Y. Wang Children's Hospital of Orange County Orange, CA, United States	First in-human, intracisternal dosing of RGX-111, an investigational AAV gene therapy, for a 21-month-old child with mucopolysaccharidosis type I (MPS I): 3.5 year follow-up
	Moderated Q&A	*Gaffney, Volz, Rangel Miller, Wang*
11:30 AM	**Break, Exhibits & Satellite Symposia**	

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1:00 PM	**R. Scott McIvor** University of Minnesota Minneapolis, MN, United States	First-in-human clinical trial of genetically engineered B cells: Application to the treatment of mucopolysaccharidosis type I
	Shababa T. Masoud Denali Therapeutics South San Francisco, CA, United States	ETV:SGSH, a brain-penetrant enzyme transport vehicle for SGSH, improves lysosomal and microglial morphology, degeneration and cognitive behavior in MPS IIIA mice
	Atsushi Imakiire JCR Pharmaceuticals Co., Ltd. Kobe, Japan	Recovery of retinal function in MPS II mice by treatment with pabinafusp alfa
	Monika Musial-Siwek Be Biopharma Cambridge, MA, United States	Development of an *ex vivo* precision gene engineered B cell medicine that produces highly active and sustained levels of acid sphingomyelinase for the treatment of Neimann-Pick disease
	Moderated Q&A	*McIvor, Masoud, Imakiire, Musial-Siwek*
2:00 PM	**Dustin Armstrong** Parasail LLC Quincy, MA, United States	A clinical candidate (VAL-1221) capable of treating multiple glycogen storage diseases including Lafora and other neurological polyglucosan disorders
	Arjan van der Flier Sanofi Cambridge, MA, United States	Anti-human-TfR-GAA efficiently clears CNS and muscle glycogen in a translatable hTfR-KI/Pompe disease mouse model
	Christian Argueta Takeda Development Center Americas, Inc. Cambridge, MA, United States	Baseline levels of neurofilament light chain in the cerebrospinal fluid correlate with clinical outcomes in patients with MPS II from a phase 2/3 clinical trial (NCT02055118) and extension study (NCT02412787) of intrathecal idursulfase
	Michael H. Gelb University of Washington Seattle, WA, United States	Second-tier glycosaminoglycan analysis in dried blood spots by the endogenous non-reducing end method provides the best approach for reducing false positives in newborn screening of all sub-types of mucopolysaccharidoses
	Moderated Q&A	*Armstrong, van der Flier, Argueta, Gelb*
3:00 PM	**Poster Session**	**Exhibit Hall**
5:15 PM	**Satellite Symposia**	

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Friday, February 9, 2024: Late-Breaking Science
Co-Chairs: Rebecca Ahrens-Nicklas, Elizabeth Braunlin, Roberto Giugliani

6:45 AM	**Satellite Symposia**	
8:00 AM	**Late-breaking Session**	4 abstracts: 12 min each
	Moderated Q&A	
9:00 AM	**Late-breaking Session**	4 abstracts: 12 min each
	Moderated Q&A	
10:00 AM	**Break**	
10:15 AM	**Late-breaking Session**	4 abstracts: 12 min each
	Moderated Q&A	
11:15 AM	**WORLD*Symposium* 2024 Adjourns**	

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