Platform Presenters

Monday, February 3, 2025: Patient Voice 2025: Inequities in Access to Diagnosis, Care and Clinical Trials in Lysosomal Diseases, Speed Mentoring and Be the Catalyst Event

15:00	The Patient Voice 2025: Inequities in Access to Diagnosis, Care and Clinical Trials in Lysosomal Diseases?	This activity is supported in part by educational grants from Takeda Pharmaceuticals USA, Inc., Denali Therapeutics, Orchard Therapeutics North America, and Ultragenyx Pharmaceutical Inc.
16:30	Speed Mentoring	
18:00	Be the Catalyst Event	

WORLD*Symposium*™ 2025 Preliminary Program*
Platform Presenters

Tuesday, February 4, 2025: Basic Science
Co-Chairs: Lalitha Belur, Michael Przybilla, Dan Tagle

06:45	**Satellite Symposia**	
08:00	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and Announcements Presentation of 2025 Roscoe O. Brady Award to James M. Wilson
	James M. Wilson GEMMA Biotherapeutics (GEMMABio) Philadelphia, PA, United States	Roscoe O. Brady Award Presentation
08:30	**Françoise Piguet** Paris Brain Institute Paris, France	Development and validation of an intravenous AAV gene therapy for mucopolysaccharidosis type IIIB in mouse and dog model of the pathology
	Pratikshya Adhikari The University of North Carolina at Chapel Hill Chapel Hill, NC, United States	AAV9-based gene replacement therapy targeting the root cause for the treatment of MPS IIID in mice **2025 Young Investigator Award Recipient*
	Andres Felipe Leal Nemours Children's Health Wilmington, DE, United States	Uncovering mitochondrial disturbances in MPS IVA chondrocytes **2025 Young Investigator Award Recipient*
	Angelica Maria Herreno Pachon University of Delaware Wilmington, DE, United States	Crispr/Cas9-edited hematopoietic stem cells rescue MPS IVA fibroblasts phenotype **2025 Young Investigator Award Recipient*
	Moderated Q&A	*Piguet, Adhikari, Leal, Herreno Pachon*
09:30	**Lachlan J. Smith** University of Pennsylvania Philadelphia, PA, United States	Postnatal progression of skeletal disease in mucopolysaccharidosis type VI dogs: Preliminary findings
	Maria Fuller SA Pathology North Adelaide, Australia	Utility of signature specific biomarkers for the mucopolysaccharidoses: 8 years experience in the diagnostic laboratory
	Shih-Chang Hsueh Columbia University Irving Medical Center New York, NY, United States	A cyclic oligosaccharide structure as a novel therapeutic strategy for Krabbe disease
	Rachel Wurth Mayo Clinic Rochester, MN, United States	Characterizing and validating the small molecule signature of Krabbe disease plasma using untargeted metabolomics analysis
	Moderated Q&A	*Smith, Fuller, Hsueh, Wurth*
10:30	**Break**	
11:00	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	PS Gene-editing system corrects the CNS with blood-brain barrier penetrant ApoE-enzymes
	Ewa A. Ziolkowska Washington University School of Medicine St. Louis, MO, United States	Treatment of dysphagia associated pathologies in CLN3 deficient mice via gene therapy **2025 Young Investigator Award Recipient*
	Nathan Phan University of Minnesota Minneapolis, MN, United States	Biomarker potential of allele-specific extracellular vesicles in Gaucher disease

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Platform Presenters

Tuesday, February 4, 2025: Basic Science *(cont.)*
Co-Chairs: Lalitha Belur, Michael Przybilla, Dan Tagle

	Parisa Amirifar Duke University Medical Center Durham, NC, United States	Enzyme replacement therapy (ERT) combined with transient low-dose methotrexate (TLD-MTX) results in age- and disease-dependent immune profile changes in infantile- vs. late-onset Pompe disease patients
	Moderated Q&A	*Whitley, Ziolkowska, Phan, Amirifar*
12:00	**Break and Satellite Symposia**	
13:30	**Allan Feng** Stanford University Stanford, CA, United States	A novel murine model for neuronopathic Gaucher disease **2025 Young Investigator Award Recipient*
	Shu Xing Yale University School of Medicine New Haven, CT, United States	Apoe-Abca1 axis is involved in the pathogenesis of Gaucher disease
	Luisa Natalia Pimentel Vera Stanford University Palo Alto, CA, United States	Correction of GD1 pathology by genome edited murine hematopoietic stem cell transplantation.
	Magali Pettazzoni Lyon University Hospital Lyon, France	When technology improves diagnosis: Incidental discovery of ASMD in patients suspected with Gaucher disease
	Moderated Q&A	*Feng, Xing, Pimentel Vera, Pettazzoni*
14:30	**David Dmitrivich Smerkous** University of Washington Seattle, WA, United States	Quantification of globotriaosylceramide (GL3) in peritubular capillary endothelial cells (PTCEC) in kidney biopsies from patients with Fabry disease using machine learning
	Abdullah Hoter University of Veterinary Medicine Hannover Hannover, Germany	Cellular uptake and function of recombinant pegunigalsidase alfa in fibroblasts from Fabry patients
	Anna Reinelt University Medical Center Hamburg-Eppendorf Hamburg, Germany	Advancing cardiac disease modeling in Fabry cardiomyopathy by utilizing patient-derived induced pluripotent stem cells, heart organoids, and engineered heart tissue **2025 Young Investigator Award Recipient*
	Malte Lenders University Hospital Muenster Muenster, Germany	Biochemical amenability in Fabry disease patients under chaperone therapy - how and when to test
	Moderated Q&A	*Smerkous, Hoter, Reinelt, Lenders*
15:30	**Poster Session and Industry Expert Theater**	Exhibit Hall (Seaport Ballroom) Seaport Foyer
17:45	**Satellite Symposia**	

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Platform Presenters

Wednesday, February 5, 2025: Translational Research
Co-Chairs: Tierra Bobo, PJ Brooks, Francyne Kubaski

06:45	**Satellite Symposia**	
08:00	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and Announcements Presentation of 2025 Patient Advocate Leader (PAL) Award to Maria Kefalas, Dean Suhr and Teryn Suhr and 2025 Young Investigator Awards Presentation
08:30	**Edina Poletto** Stanford University Stanford, CA, United States	Investigational new drug-enabling studies for genome-edited hematopoietic stem cells to treat mucopolysaccharidosis type I
	Roselena S. Schuh Federal University of Rio Grande do Sul Porto Alegre, Brazil	Evaluation of off-target events after an intravenous injection of liposomal CRISPR/Cas9 complex in vivo **2025 Young Investigator Award Recipient*
	Troy Lund University of Minnesota Minneapolis, MN, United States	Changes in CSF GAG after intravenous enzyme replacement therapy
	Kim M. Hemsley Flinders University Bedford Park, Australia	Short-term daily treatment of MPS IIIA mice with rosmarinic acid is neuroprotective.
	Moderated Q&A	*Poletto, Schuh, Lund, Hemsley*
09:30	**Brian Bigger** University of Edinburgh Edinburgh, United Kingdom	Long-term HSC gene therapy in mucopolysaccharidosis type IIIB mice corrects disease with no evidence of insertional mutagenesis despite high vector copy numbers
	Betul Celik University of Delaware Newark, DE, United States	In vivo direct bone targeting lentiviral gene therapy for MPS IVA murine model
	Sampurna Saikia University of Delaware Newark, DE, United States	Immune modulation for AAV-9 gene therapy by oral administration of peptides for GALNS enables the vector re-administration in MPS IVA
	Karthikeyan Rajagopal University of Pennsylvania Philadelphia, PA, United States	In vitro development and in vivo evaluation of intra-articular GUSB mRNA therapy for mucopolysaccharidosis type VII
	Moderated Q&A	*Bigger, Celik, Saikia, Rajagopal*
10:30	**Break and Exhibits**	
11:00	**Alberto B. Burlina** University Hospital of Padua Padua, Italy	Neonatal screening for Fabry disease and long-term follow-up: The role of plasma globotriaosylsphingosine (LysoGb3) assay
	Paige Nowlin Brigham and Women's Hospital Boston, MA, United States	Enabling CNS delivery of rhGAA in GAA $^{-/-}$ mice using focused ultrasound
	Robert J. Hopkin Cincinnati Children's Hospital Medical Center Cincinnati, OH, United States	Miglustat: A first-in-class enzyme stabilizer for late-onset Pompe disease
	Aimee Donald University of Manchester Manchester, United Kingdom	Two hundred and fifty cases of "Gaucher disease type 2": A novel system of clinical categorization and evidence of genotype:phenotype correlation **2025 Young Investigator Award Recipient*
	Moderated Q&A	*Burlina, Nowlin, Hopkin, Donald*

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Platform Presenters

Wednesday, February 5, 2025: Translational Research *(cont.)*
Co-Chairs: Tierra Bobo, PJ Brooks, Francyne Kubaski

12:00	**Break, Exhibits and Satellite Symposia**	
13:30	**Patrick B. Deegan** Addenbrooke's Hospital Cambridge, United Kingdom	Algorithmic case finding approaches for Gaucher Disease type 1 in primary care records
	Pasqualina Colella Stanford University Palo Alto, CA, United States	Genome-edited autologous stem cell transplantation with enhanced brain conditioning to correct progranulin deficiency
	Akhil Kulkarni National Human Genome Research Institute, National Institutes of Health Bethesda, MD, United States	A novel AAV-based gene therapy strategy reverses lethality in a murine model of neuronopathic Gaucher disease
	Krystyna Noelle Rytel National Human Genome Research Institute, National Institutes of Health Bethesda, MD, United States	A genome-wide CRISPR activation screen to identify beta-glucocerebrosidase modifiers **2025 Young Investigator Award Recipient*
	Moderated Q&A	*Deegan, Colella, Kulkarni, Rytel*
14:30	**Keerthana Iyer** University of Pennsylvania Philadelphia, PA, United States	Porous microcarriers for sustained delivery of mRNA-lipid nanoparticles to treat joint disease in the mucopolysaccharidoses
	Michael J. Przybilla University of Minnesota Minneapolis, MN, United States	Improving blood-brain barrier penetration in Hurler syndrome using an IDUA-ApoE fusion enzyme delivered via the PS Gene Editing System
	Megan Joy Clarke Albert Einstein College of Medicine/Children's Hospital at Montefiore Bronx, NY, United States	Screenplus: An assay-based multi-tiered testing model for expanded NBS
	Dau-Ming Niu Taipei Veterans General Hospital Taipei, Taiwan	Applications of a rapid real time analysis system for whole genome/exome sequencing in newborn screening
	Moderated Q&A	*Iyer, Przybilla, Clarke, Niu*
15:30	**Poster Session and Industry Expert Theater**	Exhibit Hall (Seaport Ballroom) Seaport Foyer
17:45	**Satellite Symposia**	

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Platform Presenters

Thursday, February 6, 2025: Clinical Applications
Co-Chairs: Uma Ramaswami, Filippo Vairo, Ray Wang

06:45	**Satellite Symposia**	
08:00	**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:* Advancing the Frontier of Gene Therapy
08:30	**Connor J. Lewis** National Human Genome Research Institute, National Institutes of Health Bethesda, MD, United States	Volumetric magnetic resonance imaging and diffusion tensor imaging metrics correlate with clinical outcomes following gene therapy in GM1 gangliosidosis patients **2025 Young Investigator Award Recipient*
	Joseph Muenzer University of North Carolina Chapel Hill Chapel Hill, NC, United States	Interim analysis of the efficacy and safety of weekly intravenous tividenofusp alfa in mucopolysaccharidosis type II (MPS II): A phase 1/2 study
	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)
	Mark Thomas Royal Perth Hospital Perth, Australia	Phase 1/2 clinical trial evaluating 4D-310 in adults with Fabry disease cardiomyopathy: Interim analysis of cardiac and safety outcomes in patients with 21-42 months of follow up
	Moderated Q&A	*Lewis, Muenzer, Harmatz, Thomas*
09:30	**Derralynn Hughes** Royal Free London NHS Foundation Trust London, United Kingdom	Isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease: Updated results from an ongoing phase 1/2 study (STAAR)
	Alessandro Burlina St. Bassiano Hospital Bassano del Grappa, Italy	Reduced incidence of stroke in patients with Fabry disease treated with agalsidase beta: A matched analysis from the Fabry Registry
	Christiane Auray-Blais Université de Sherbrooke Sherbrooke, QC, Canada	Fabry disease biomarker evaluation during a five-year gene therapy clinical trial
	Aneal Khan M.A.G.I.C. Clinic Ltd Calgary, AB, Canada	Lentiviral gene therapy for Fabry disease - 5 year end of study analysis for the FACTS trial
	Moderated Q&A	*Hughes, Burlina, Auray-Blais, Khan*
10:30	**Break and Exhibits**	
11:00	**Maria Ester Me Bernardo** San Raffaele Telethon Institute for Gene Therapy Milan, Italy	Hematopoietic stem cell gene therapy for mucopolysaccharidosis type I-Hurler syndrome (OTL-203): Interim skeletal, neurological and systemic outcomes from a phase I/II study
	Francesca Fumagalli IRCCS San Raffaele Scientific Institute Milan, Italy	Atidarsagene autotemcel (autologous hematopoietic stem cell gene therapy) preserves cognition, language, and speech and slows brain demyelination and atrophy in early-onset metachromatic leukodystrophy
	Tahseen Mozaffar University of California Irvine Irvine, CA, United States	Azt845 gene replacement therapy for late-onset Pompe disease: An update on safety and preliminary efficacy data from FORTIS, a phase 1/2 open-label clinical study

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	Reena Sharma Salford Royal Hospital Salord, United Kingdom	Results from GALILEO1, a first in human clinical trial of FLT201 AAV-gene therapy in adult patients with Gaucher disease type 1
	Moderated Q&A	*Bernardo, Fumagalli, Mozaffar, Sharma*
12:00	**Break, Exhibits and Satellite Symposia**	
13:30	**Roberto Giugliani** Federal University of Rio Grande do Sul Porto Alegre, RS, Brazil	Rainbow study: Phase 2 study of nizubaglustat as an investigational treatment for Niemann-Pick disease type C and GM2 gangliosidosis
	Tatiana Bremova-Ertl University of Bern Bern, Switzerland	Long-term findings of N-acetyl-L-leucine for Niemann-Pick disease type C
	Orna Staretz Chacham Soroka Medical Center Be'er Sheva, Israel	Trappsol® Cyclo™: open label treatment in the transportnpc™ sub-study in patients under the age of 3 diagnosed with Niemann -Pick disease type c1
	Benedikt Schoser Ludwig-Maximilians-University Munich, Germany	Clinically important improvements in 6-minute walk distance (6MWD) and forced vital capacity (FVC) in adults with late-onset Pompe disease (LOPD) switching from alglucosidase alfa (alg) to cipaglucosidase alfa plus miglustat (cipa+mig) in the PROPEL study
	Moderated Q&A	*Giugliani, Bremova-Ertl, Staretz Chacham, Schoser*
14:30	**Maurizio Scarpa** University Hospital of Udine Udine, Italy	Children with chronic acid sphingomyelinase deficiency treated with olipudase alfa for 4+ years show improvements or normalization in multiple disease manifestations: Final results of the ASCEND-Peds trial
	Melissa Wasserstein The University Hospital for Albert Einstein College of Medicine Bronx, NY, United States	Final results of the ASCEND trial: Continued or sustained improvements in hepatosplenomegaly, respiratory outcomes, and lipid profile after 4 years of olipudase alfa enzyme replacement therapy in adults with acid sphingomyelinase deficiency
	Pilar Giraldo Hopital Quirónsalud Zaragoza and Spanish Foundation for Gaucher Disease and other Lysosomal Disorders (FEETEG) Zaragoza, Spain	Efficacy of eliglustat administered with and without imiglucerase in pediatric participants with Gaucher disease type 1 or type 3: The ELIKIDS study
	Pramod K. Mistry Yale University School of Medicine New Haven, CT, United States	Long-term outcomes of imiglucerase treatment in children with Gaucher disease type 1 or type 3 starting therapy before the age of 2 years
	Moderated Q&A	*Scarpa, Wasserstein, Giraldo, Mistry*
15:30	**Poster Session and Industry Expert Theater**	Exhibit Hall (Seaport Ballroom) Seaport Foyer
17:45	**4th Annual Robert J. Gorlin Symposium**	The Situation Room: Gene Therapy in the Real World

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Platform Presenters

Friday, February 7, 2025: Contemporary Forum and Late-breaking Science
Co-Chairs: Elizabeth Braunlin, Roberto Giugliani, Cynthia Tifft

06:45	**Satellite Symposia**	
08:00	**Chester B. Whitley** University of Minnesota Minneapolis, MN, United States	Welcome and New Treatment Awards
08:30	**Heather Ann Lau** Ultragenyx Pharmaceutical Inc Novato, CA, United States	Treatment with UX111 gene therapy rapidly reduced heparan sulfate (HS) exposure in cerebrospinal fluid (CSF) and improved long-term cognitive function in children with mucopolysaccharidosis type IIIA (MPS IIIA)
	Daniel Virga Regeneron Pharmaceuticals Tarrytown, NY, United States	Development of a durable gene therapy for targeting CNS and visceral pathologies in acid sphingomyelinase deficiency
	Eugen Mengel SphinCS Hochheim, Germany	Efficacy results from a 12-month double-blind randomised trial of arimoclomol for treatment of Niemann-Pick disease type C- presenting are scored 4-domain NPC clinical severity scale
	Tyler Picariello Dyne Therapeutics Waltham, MA, United States	The FORCE™ platform delivers acid alpha-glucosidase to muscle as well as central nervous system and resolves pathology in Pompe disease mice
	Moderated Q&A	*Lau, Virga, Mengel, Picariello*
09:30	**Frances M. Platt** University of Oxford Oxford, United Kingdom	Insights into the mechanism of action of a acetyl-leucine as a therapeutic for lysosomal diseases
	John A. Bernat University of Iowa Health Care Iowa City, IA, United States	Extending the interval between pegunigalsidase alfa infusions in patients with Fabry disease: Five-year interim results from the ongoing BRIGHT51 study
	Late-Breaking Abstract	2 Speakers To Be Announced (TBA) with Moderated Q&A
	Moderated Q&A	*Platt, Bernat*
10:30	**Break**	
11:00	**Late-Breaking Abstract**	4 Speakers To Be Announced (TBA) with Moderated Q&A
12:00	**Break and Satellite Symposia**	

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Platform Presenters

Friday, February 7, 2025: Rapid Fire Competition
Co-Chairs: Rebecca Ahrens-Nicklas, Amy Gaviglio, Marc Patterson

13:30	**Melissa A. Calton** 4D Molecular Therapeutics Emeryville, CA, United States	Non-clinical evaluation of 4D-310 in combination with rituximab/sirolimus: A translational study to support adoption of a novel prophylactic immunomodulation regimen in clinical trials in adults with Fabry disease
13:40	**Shiny Nair** Yale University School of Medicine New Haven, CT, United States	Molecular cell atlas of the brain in neuronopathic Gaucher disease
13:50	**Tippi C. MacKenzie** University of California San Francisco San Francisco, CA, United States	Interim results from a first in human phase 1 clinical trial of in utero enzyme replacement therapy for lysosomal disorders
14:00	**Dawn A. Laney** Emory University School of Medicine Atlanta, GA, United States	Development and validation of an automated predictive scoring system to identify patients at increased risk for Fabry disease using Japanese electronic cardiac failure data
14:10	**Emily Eshraghian** University of Minnesota Minneapolis, MN, United States	Long term enzyme replacement therapy after hematopoietic stem cell transplant results in immune tolerance and improved biochemical outcomes
14:20	**Slawomir Wantuch** Orchard Therapeutics London, United Kingdom	Correction of glycogen accumulation in muscle, heart and CNS in a pre-clinical model of hematopoietic stem cell gene therapy for Pompe disease
14:30	**Michael H. Gelb** University of Washington Seattle, WA, United States	Massively parallel biochemical annotation of VOUS for lysosomal disorders
14:40	**Jennifer Goldstein** University of North Carolina at Chapel Hill Chapel Hill, NC, United States	Pseudodeficiency: A poorly defined and misunderstood term in an era of precision medicine
14:50	**Miles Clark Greenberg** University of Minnesota Minneapolis, MN, United States	Heparan sulfate reduction in cerebrospinal fluid is associated with long-term cognitive outcomes in Hurler syndrome.
15:00	**Rapid Fire Abstract**	3 Speakers To Be Announced (TBA)
15:10	**Rapid Fire Abstract**	
15:20	**Rapid Fire Abstract**	
15:30	**WORLDSymposium 2025 Adjourns**	

**The Preliminary Program is subject to change without notice. Any updates to the program will be posted on the website: worldsymposia.org*

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