All printed posters will be available to WORLD*Symposium* attendees beginning 3:00 PM PST on Monday, February 5, through 5:00 PM PST on Thursday, February 8, 2024, in the Exhibit Hall. In addition, ePosters will be available for viewing to all registered attendees throughout the live meeting on the WORLD*Symposium* Mobile App, and available to On Demand registered attendees, from February 14 – March 14, 2024.

Live Q&A poster discussions will occur onsite, during the assigned times listed below. Each poster presenter has an assigned day to present their abstract in-person, based on one of four abstract categories, noted below.

All live poster presentations will be in the Seaport Ballroom (Exhibit Hall), on their scheduled date and time. Each daily poster reception is scheduled for a two-hour session from 3:00-5:00 PM PST:

Basic Science Posters will be presented on **Monday, February 5 from 3:00-5:00 PM**
Translational Research Posters will be presented on **Tuesday, February 6 from 3:00-5:00 PM**
Clinical Application Posters will be presented on **Wednesday, February 7 from 3:00-5:00 PM**
Contemporary Forum Posters will be presented on **Thursday, February 8 from 3:00-5:00 PM**
*** Late-Breaking Science Posters (poster numbers starting with an LB) will be divided between Monday, Tuesday and Thursday.

Any poster numbers not listed will not be presented as the author is unable to attend the conference.

It is the policy of WORLD*Symposium* to publish all abstracts with the list of authors exactly as the abstract was submitted to WORLD*Symposium*. The first author of the submitted abstract will be listed as the presenting author on the Preliminary Program, Agenda, and Poster List.

Basic Science – Poster Session I – Monday, February 5, 3:00 PM - 5:00 PM

#	Author	Title	Kiosk
1	Magy Abdelwahab	Is Gaucher disease in children a continuum of phenotype, a hypothesis or is it clinically evidence-based? Analysis of Egyptian and American cohorts	1-A
6	Anusha Aditya	Using iNSC patient-derived oligodendrocytes to characterize small molecules for globoid-cell leukodystrophy	1-E
10	Danna Araujo	Importance of accurate diagnosis of neuromuscular diseases for the sake of 6P medicine: Clinical case Kugelberg-Welander disease (spinal muscular atrophy type III)	2-A
11	Isidro Arevalo-Vargas	*Correlation between chitotriosidase activity in dried blood spots and plasma among Gaucher disease patients*	2-E
12	Isidro Arevalo-Vargas	Exploring lipid biomarkers in Gaucher disease: LC-MS/MS analysis of dried blood spot samples	3-A
17	Imane Assiri	Sialic acid related disorders diagnosis: The experience of the first Moroccan reference center of inherited metabolic diseases	3-E
18	Maria Athanasopoulos	Impact of ASGR1 on the hepatic expression and localization of NEU1	4-A
26	Lorenzo Badenetti	Hampered differentiation and disrupted endo-lysosomal function in a human neuronal in vitro model of mucopolysaccharidosis type II	4-E
31	Salma Begum	The psychosine and galactosylceramide brain spatial distribution and its correlation with neuropathogenic processes	5-A
35	Bryce Binstadt	Identification of inflammatory cells in dilated ascending aortas of IDUA-deficient (MPS I) mice	5-E
36	Gal Bitan	New blood-based biomarkers for Sanfilippo syndrome	6-A
40	Alessandro Burlina	Family screening after newborn screening: The “domino effect” for Fabry disease	6-E
42	Devin Burris	Dentate gyrus developmental timeline in CLN3del78/del78 (Batten disease) mice	7-A
46	Caitlin Calhoun	Functional efficacy of transplanted, iPSC-derived, human neural stem cells in the brains of MPS I mice	7-E
49	Margaret Cassidy	Patient-derived NGN2-induced neurons recapitulate biochemical phenotypes of multiple sulfatase deficiency in vitro	8-A

52	Tiffany Chen	A comparative neuropathological evaluation of knock-in murine models of Gaucher disease	8-E
56	Chloe Christensen	Restoration of acid-alpha glucosidase expression and function through efficient adenine base editing of Pompe disease variants	9-A
57	Heather Church	How many sulphatase deficiencies become multiple? The diagnostic challenges of multiple sulphatase deficiency	9-E
64	Julia Dao	Bone resorption biomarkers and proinflammatory cytokines in Gaucher disease	10-A
65	Narayan Dhimal	Studying the role of autophagy in Krabbe disease	10-E
84	Nicolas Fernandez Escobar	Argentine bone project- 11 years of experience: A study by the Argentine Gaucher Disease Diagnosis and Treatment Group	11-A
91	Maria Fuller	A multiplex lipid platform improves the laboratory diagnosis of the sphingolipidoses	11-E
93	Lidia Gaffke	Actin cytoskeleton polymerization and focal adhesion as important factors in the pathomechanism and potential targets of mucopolysaccharidosis treatment	12-A
106	Pilar Giraldo	Analysis of genotype-phenotype correlation in Gaucher disease type 1 using machine learning techniques	12-E
116	Esteban Gonzalez	*Losartan treatment in mucopolysaccharidosis type I mice: Beneficial effects on aortic structure and pathways insights*	13-A
117	Esteban Gonzalez	*Bone disease in mucopolysaccharidosis type I: Morphological, structural and biomechanical characterization, and effect of different treatment approaches*	13-E
118	Vincenza Gragnaniello	Non-compaction myocardium in an early-treated infantile-onset Pompe disease patient	14-A
122	Kylie Gray	M021 (rhGAA) has a unique glycosylation profile which enables more efficient glycogen reduction and may allow for alternative Pompe ERT dosing strategies	14-E
125	Jacky Guerrero	CRISPR/Cas9-based gene therapy for Tay-Sachs disease: Evaluation of non-viral vectors	15-A
133	Miloud Hammoud	The utilization of a combination of TLC-UHPL/MS-MS and molecular networking in the assessment and prediction of Fabry and Sandhoff diseases	15-E
134	Jerry Harb	Exploring Pompe disease: Insights into the natural history of novel Gaac.1826dupA knock-in murine model	16-A
141	Alvaro Hermida	Characterization of plasma proteomic profile in Fabry disease	16-E
142	Jacqueline Hernandez	Defects in cell polarity of mucopolysaccharidosis type III (MPS III) forebrain neurons	17-A
144	Xinying Hong	UPLC-MS/MS analysis of urinary oligosaccharides for the diagnosis of mucopolysaccharidosis and glycoproteinosis	17-E
146	Mahin Hossain	Investigating the role of SLC17A5 in myelination and neurodegeneration in a murine model of free sialic acid storage disorder	18-A
150	Marjan Huizing	Cell-based functional assays for free sialic acid storage disorder	18-E
157	Orhan Kerim Inci	Treatment with recombinant human lysosomal β-hexosaminidase reduces GM2 accumulation in Tay-Sachs disease cells	19-A
158	Gabrielle Iop	Biomarker distribution in tissues of MPS I mice: Measurement of disease-specific oligosaccharides by LC-MS/MS	19-E
159	Skyler Jackson	Rare Disease Difference Maker® program	20-A
161	Maroua Jakani	A simple, non-invasive method for the diagnosis of glycogen storage disease type III in two patients with a similar facial appearance	20-E
169	Shih-hsin Kan	Improvement of hypertrophic cardiomyopathy in Gaac.1826dupA knock-in murine model with neonatal gene therapy	21-A

172	Neil Kasaci	Lyso-Gb1 acts as a danger-associated molecular pattern (DAMP) to induce cell death and autonomous disease progession, indicating the role of additional therapeutics in Gaucher disease	21-E
173	Megan Keating	Single centre review and analysis of enzyme replacement therapy infusion associated reactions in patients with lysosomal disorders	22-A
177	Veenita Khare	Novel insights advancing the understanding of renal Fanconi syndrome in cystinosis	22-E
191	Patricia Lam	Liver-directed AAV gene therapy corrects disease symptoms in a murine model of lysosomal acid lipase deficiency	23-A
193	Steven Le	Elucidation of the pathogenesis of heparan sulfate in MPS IIIB mice using a membrane-tethered form of NAGLU	23-E
197	Malte Lenders	Identification and characterization of pre-existing anti-PEG and anti-AGAL antibodies towards pegunigalsidase-alfa	24-A
199	Daniel Lewi	Empowering the GM2 gangliosidosis community: Developing a single clinical toolkit for clinicians, patients and caregivers in the UK	24-E
207	Charis Ma	Generation of GBA1 isogenic iPSC lines to investigate the pathogenesis of GBA1-associated Parkinson disease	25-A
212	Rosa Manzoli	Molecular insights of axon guidance abnormalities in a mucopolysaccharidosis type II zebrafish model	25-E
226	Travis Moore	A small molecule drug, AVP6, rescues synaptic deficits in human iPSC-derived neurons across the mucopolysaccharidosis type III spectrum	26-A
228	David Moreno Martinez	Relationship of adhesion and chemotaxis to pathology in Fabry disease: Preliminary results of the analysis of classical monocytes from patients and a THP-1 model	26-E
239	Shiny Nair	VavCre mediated conditional deletion of Gba in mice recapitulates human Gaucher disease type 1, a platform to investigate the role of myeloid cells and altered hematopoiesis	27-A
240	Samira Najeh	The role of GC-MS in organic acidurias diagnosis in Moroccan population	27-E
243	Katherin Niño	Initial characterization of cellular models for mucopolysaccharidosis type IIIB generated by CRISPR/Cas9	28-A
247	Wataru Oboshi	Quantitative determination of glycosaminoglycans in dried blood spots for second-tier screening of mucopolysaccharidoses	28-E
253	Helen Parker	The meningeal immune landscape in mucopolysaccharidosis type IIIA	29-A
256	Allyson Peek	Nuclear localization of the NEU4 isoforms in human fibroblasts and HepG2 cell line	29-E
266	Michael Przybilla	Optimal stoichiometry of CRISPR-Cas9 components in the PS Gene-editing System	30-A
278	Estera Rintz	Molecular mechanism of resveratrol-induced autophagy in mouse model of Sanfilippo syndrome type IIIB	30-E
283	Es-Said Sabir	Molecular analysis of mucopolysaccharidosis type I in Morocco: Identification of novel mutation	31-A
284	Marya Sabir	Advancing free sialic acid storage (FSASD) disorder disease modeling: Insights from iPSC-derived neural cell types	31-E
292	Benedikt Schoser	Variability of GAA enzyme and glycogen levels in different muscle groups	32-A
299	Irene Serrano Gonzalo	Bone involvement in Gaucher disease: Can miRNAs determine or predict the severity degree?	32-E
300	Irene Serrano Gonzalo	Study of the development and implication of extracellular traps of neutrophils in vascular complications of lysosomal diseases	33-A
301	Livia Sertori Finoti	Generation and characterization of multiple sulfatase deficiency iPSC-line and neurogenin-2-induced neurons	33-E

305	Yuki Shiro	CTSD integrity in the endoplasmic reticulum is required for CLN6's anti-aggregate activity	34-A
309	Rodrigo Starosta	Decrease in psychosine in response to a ceramide galactosyltransferase inhibitor in a novel human cell model of Krabbe disease	34-E
313	Diego Suarez	GM2 gangliosidosis insights: Tay-Sachs models via CRISPR-Cas9	35-A
317	Beena Thomas	Severe form of Niemann-Pick disease type B in Iranian population: A review of the spectrum of disease in acid sphingomyelinase deficiency	35-E
321	Emilio Vaena	MPS II models for the study of joint and bone pathophysiology using CRISPR/Cas9 technology	36-A
322	Amber Van Baelen	When simultaneous detection becomes possible: A new screening method for lysosomal diseases	36-E
344	Matheus Wilke	Development of a scoring system to define lysosomal disease	37-A
351	Sarah Young	Measurement of glycosaminoglycans in the amniotic fluid of fetuses with mucopolysaccharidoses treated in a phase I clinical trial by in utero enzyme replacement therapy	37-E
LB-06	Christian Beetz	Therapeutically relevant insights from comparative characterization of GBA-associated biochemical parameters in Gaucher disease and Parkinson disease	38-A
LB-07	Laura Bell	Niemann Pick UK (NPUK) care and support team making a difference for patients and families	38-E
LB-14	Krista Casazza	Neurofilament light- A potential dynamic cross-disease fluid biomarker for CNS involvement in lysosomal diseases	39-A
LB-16	Marie-Anne Colle	Early activation of the membrane repair stabilisation pathways in Pompe disease (glycogenosis type 2)	39-E
LB-17	Udita Datta	Identification of translatable biomarkers in rodent models of Batten disease	40-A
LB-19	Conan Donnelly	Insights into Niemann-Pick disease type C - An Update from the International Niemann-Pick Disease Registry	40-E
LB-20	Fatma Eminoğlu	Long-term clinical evaluation of patients with alpha-mannosidosis: A multicenter study	41-A
LB-22	Sandra Facincone	Translation, cultural adaptation, and validation of the burden of illness survey (BBoIS) for parents or guardians of children with severe MPS II in Brazil	41-E
LB-24	Jessica Gambardella	Hypertension exaggerates left ventricular hypertrophy in Fabry patients	42-A
LB-25	Beatriz Guzman	GT-02287, a clinical stage GCase enhancer, displays neuroprotection and restores motor function in preclinical models of Parkinson disease following delayed administration	42-E
LB-27	Wendy Heywood	Fabry disease associated left ventricular hypertrophy can be determined by a machine learning based inflammatory serum panel	43-A
LB-28	Ghada Hijazi	Intrafamilial variability of c.427G>A (p.Ala143Thr) variant in the GLA gene: A case report	43-E
LB-31	Chanchala Kaddi	Head-to-head virtual comparison of alglucosidase alfa and avalglucosidase alfa efficacy in infantile-onset Pompe disease patients	44-A
LB-32	Wonki Kim	Effect on renal manifestation in Fabry disease by HM15421, a novel long-acting alpha-galactosidase A analog, in a symptomatic mouse model	44-E
LB-36	Kimmo Lehtimäki	Characterizing the CLN3Δex7/8 mouse model of Batten disease: Analysis of fine motor kinematics and retinal function	45-A
LB-37	Yi Lin	An advanced microglia-containing brain organoid platform for neuronopathic Gaucher disease modeling	45-E
LB-45	Petra Oliva	Results of a 12-month prospective high-risk population study for the frequency of α-Mannosidosis within MPS like phenotype patients in Europe and the Middle East	46-A

LB-47	cho rong Park	Effect on vascular and neurological manifestations in Fabry disease by HM15421, a novel long–acting alpha–galactosidase A analog, in a symptomatic mouse model	46-E
LB-49	Krystyna Rytel	Multi-omic analysis of iPSC-derived neurons from pairs of siblings with Gaucher disease discordant for Parkinson disease	47-A
LB-50	Roberto Sandoval	Home infusion for lysosomal diseases (LD) in Mexico: Myth or reality. Experience of the first patient in Mexico in modular infusion enzyme replacement therapy (ERT) for LD.	47-E
LB-52	Pablo Solís-Sánchez	Seizures as a novel sign of alpha-mannosidosis mimicking West and Lennox-Gastaut syndromes: A case report	48-A
LB-58	Christine Ulbricht	A systematic literature review of the healthcare resource utilization and humanistic burden of mucopolysaccharidosis type II	48-E
LB-62	Changrui Xiao	Cerebrovascular reactivity to hypo and hypercapnia in Fabry disease on fMRI	49-A
LB-63	Xiangli Zhao	Blockage of C5a/C5aR1 signaling neutralizes the aggravating effects of progranulin deficiency in Gaucher disease	49-E
LB-64	Ewa Ziólkowska	Gene therapy treats the neuromuscular consequences of CLN3 deficiency in mice	50-A

Translational Research – Poster Session II – Tuesday, February 6, 3:00 PM - 5:00 PM

#	Author	Title	Kiosk
7	Alia Ahmed	Developing biomarker trends to establish the efficacy of treatment, and to compare relative differences between different drugs	1-C
16	Mehrafarin Ashiri	Evaluation of the effects of HexM and its mannose-6-hyper-phosphorylated form (PhosHexM) in reducing GM2 ganglioside storage in a Tay-Sachs disease mouse model	1-G
20	Emilie Audouard	Cell-based device provides effective therapeutic strategy to treat metachromatic leukodystrophy	2-C
21	Christiane Auray-Blais	Glycosphingolipid evaluation for Fabry disease patients receiving migalastat after switching from enzyme replacement therapy	2-G
22	Christiane Auray-Blais	Technological update of the Provincial Neonatal Urine Screening Program in Quebec	3-C
23	Christiane Auray-Blais	Analysis by tandem mass spectrometry of lyso-Gb₃ and related analogues in dried blood spots: A convenient way to monitor patients affected with Fabry disease	3-G
25	Rafael Badell-Grau	Gene modified hematopoietic stem cell transplantation for mucopolysaccharidosis type IIIC	4-C
27	Tomas Baldwin	The development and application of a rapid and more informative test for autoantibodies to enzyme replacement therapies in Fabry disease	4-G
29	Elena Gaia Banchi	Development and validation of a novel AAV gene therapy for mucopolysaccharidosis type IIIB in large animal 	5-C
50	Betul Celik	Ex vivo lentiviral gene therapy for mucopolysaccharidosis type IVA	5-G
59	Tanya Collin-Histed	The IGA calls for greater clarity and accuracy in the use of non-comparables for the treatment of Gaucher disease	6-C
82	Asma Farjallah	Discovery of novel MPS II neuronopathic biomarkers using untargeted metabolomic approaches	6-G
90	Maurice Flurie	Cognitive and mental health challenges in Fabry disease: A real-world evidence study using social media	7-C
95	Jillian Gallagher	Adeno-associated viral gene therapy for sialidosis using small and large animal models	7-G
99	Dominique Germain	Facilitating intrafamily communication to enable earlier diagnosis of Fabry disease in relatives: Expert opinion	8-C
104	Pilar Giraldo	Descriptive study of patients with Gaucher disease type 3 in Spain and clinical response to different therapies	8-G
114	Logan Glasstetter	A novel quantitative high-throughput screening assay identifies small-molecule therapeutic candidates for Gaucher and Parkinson disease	9-C
119	Vincenza Gragnaniello	Normal cognitive outcome of transplanted mucopolysaccharidosis type I patients diagnosed by newborn screening	9-G
130	Punita Gupta	Clinical status update for prenatally diagnosed CRIM negative IOPD Patient	10-C
138	Kim Hemsley	Superior outcomes in neuroretina following IV versus intra-CSF AAV9 gene replacement in mice with MPS IIIA	10-G
139	Nadene Henderson	More frequent dosing with agalsidase beta: An update to our Fabry disease cohort	11-C
143	Rachel Hickey	Utility of routine magnetic resonance imaging in disease monitoring for acid sphingomyelinase deficiency	11-G
151	Sarah Hurt	Elucidating expression of a corrective enzyme for mucopolysaccharidoses type I through administration of an adenoviral vector targeted to endothelial cells	12-C

152	Sarah Hurt	Anti-IDUA IgG alters cortical bone structure of mucopolysaccaridosis type I mice treated with intravenous enzyme replacement therapy	12-G
153	Suleiman Igdoura	A new method for the purification of bioviable NEU1 sialidase for enzyme replacement therapy for sialidosis	13-C
156	Jackie Imrie	Metachromatic leukodystrophy: Working together for optimal patient support	13-G
160	Shailly Jain	Phenotypic findings related to the c.1139+1 G > A variant in four individuals with MPS IVA	14-C
171	Audrey Kao	A long-term observation of ophthalmology assessment in atypical CLN2 patients receiving ICV cerliponase alfa treatment	14-G
174	Pamela Kell	Application of a pentasaccharide biomarker to access treatment efficacy of gene therapy for GM1 gangliosidosis	15-C
175	Johnny Kenth	Novel approach for tracheal resection in Morquio syndrome type A with end-stage critical airway obstruction: A UK case series	15-G
183	Uma Ramaswami	Changes in enzymatic activity of alpha galactosidase A in patients with Fabry disease treated with an oral chaperone therapy	16-C
184	Akhil Kulkarni	Assessment of a novel gene therapy strategy for neuronopathic Gaucher disease	16-G
185	KeriAnn Kuperman	Emerging challenges in lysosomal newborn screening: A multi-state cohort analyzing the benefits and harms of uncertain prognoses	17-C
186	Joanne Kurtzberg	Neurodevelopmental outcomes of hematopoietic stem cell transplantation for infantile Krabbe disease diagnosed through newborn screening	17-G
194	Andres Leal	Assessment of an iron oxide-coupled CRISPR/nCas9 gene editing in a mucopolysaccharidosis type IVA mouse model: An update and future perspectives	18-C
195	Chris Lee	Brain targeted AAV-GALC gene therapy reduces psychosine and extends lifespan in a mouse model of Krabbe disease	18-G
202	Charles Lourenco	What lies beneath: Next generation sequencing unraveling Niemann-Pick disease type C in adults	19-C
209	May Christine Malicdan	Collaborative research efforts drive therapeutic advancements for free sialic acid storage disorder (FSASD)	19-G
214	Tristan Martineau	Urine filter paper high-risk screening test for early detection of lysosphingolipidoses using tandem mass spectrometry	20-C
217	Dietrich Matern	Urine glycosaminoglycans: Data from the CAP/ACMG proficiency testing program	20-G
219	Molly McPheron	Challenges in management of late-onset Pompe disease (LOPD) identified through Indiana newborn screening	21-C
221	Naresh Kumar Meena	Intravital imaging of muscle damage and response to therapy in a model of Pompe disease	21-G
237	Sylvia Mutua	Validation of GMFC-MLD scale as a measure of gross motor function in metachromatic leukodystrophy	22-C
238	Steven Nadler	Nonclinical studies in non-human primates on ABX1100: A centyrin:Gys1 siRNA conjugate for the treatment of Pompe disease	22-G
242	Igor Nestrasil	Quantitative brain morphometry identifies cerebellar, cortical, and subcortical gray and white matter atrophy in late-onset Tay-Sachs disease	23-C
251	Saida Ortolano	Morphological hallmarks of classical Fabry disease: An ultrastructural study in a large Spanish family	23-G
252	Katrina Paleologos	A parent's journey to consent: An analysis of the number and mode of attempts used to gain consent from the first 11,000 ScreenPlus participants	24-C
255	Bartholomew Pederson	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	24-G

257	Vi Pham	Single vs. dual transgene ex vivo gene therapy for multiple sulfatase deficiency	25-C
259	Luisa Pimentel Vera	Genome-edited hematopoietic stem cells as a curative approach for Gaucher disease type 1	25-G
261	Edina Poletto	Clinical development of autologous genome-edited hematopoietic stem cells to treat mucopolysaccharidosis type I	26-C
263	Fabiano Poswar	Safety and tolerability of losartan for the treatment of cardiovascular manifestations in mucopolysaccharidoses types IVA and VI	26-G
264	Carlos Prada	Hips don't lie: Utility of imaging in individuals with Gaucher disease	27-C
271	Christina Quitmann	Hip dysplasia in Hurler syndrome: A retrospective analysis of longitudinal data from neonatal hip screening to long-term follow-up	27-G
275	Suraj Ramchand	Using machine learning to distinguish Fabry disease from hypertrophic cardiomyopathy with ECG and ECHO data	28-C
277	Allisandra Rha	Prime editing corrects the Gaa c.1935C>A pathogenic variant in infantile-onset Pompe disease mouse myoblasts	28-G
281	Jaehyeok Roh	Repeated-dose oral n-acetylcysteine in Gaucher disease: Pharmacokinetics summary	29-C
285	Sampurna Saikia	Immune tolerance to GALNS enhances the therapeutic efficacy of AAV gene therapy	29-G
290	Katie Sapp	Newborn screening for lysosomal disorders: Three years of experience in Indiana	30-C
294	Roselena Schuh	Intra-articular administration of nonviral vectors aminh at mucopolysaccharidosis type I mice gene editing	30-G
296	Ida Vanessa Schwartz	Metab-Latam: Sharing scientific knowledge about lysosomal disorders in Latin America	31-C
297	Annalisa Sechi	Efficacy of miglustat treatment in a patient with SCARB2 associated action myoclonus renal failure syndrome	31-G
302	Anjana Sevagamoorthy	Development of a rigorous approach for retrospective natural history studies in leukodystrophies	32-C
307	Garima Shrivastava	New insights into the use of bortezomib-based immunomodulation in the setting of high sustained antibody titers in Pompe disease	32-G
310	Connolly Steigerwald	CLN2 disease resulting from a novel homozygous deep intronic splice variant in TPP1 discovered using long-read sequencing	33-C
315	Emilie Théberge	Chest pain subtype prevalence in the British Columbia cohort of the Canadian Fabry Disease Initiative	33-G
316	Emilie Théberge	Retrospective review of recent ASA prescribing practices for primary prevention of major adverse cardiovascular events in the Canadian Fabry Disease Initiative cohort	34-C
324	Nato Vashakmadze	Alpha-mannosidosis in children and adults: 17 clinical cases	34-G
328	Sandra Vranic	Graphene flakes for enhanced delivery of the enzyme to the lysosomes of patient-derived fibroblasts: Bio-persistence and kinetics of substrate degradation	35-C
330	Hua Wang	Unveiling a novel disease entity of lysosomal disorder family MBTPS1-related spondyloepimetaphyseal dysplasia with elevated lysosomal enzymes: A case series review	35-G
341	Lena Marie Westermann	Analysis of drug-specific antibody response against cerliponase alfa in CLN2 patients by applying a novel two-step assay	36-C
342	Anna-Maria Wiesinger	Parental perception of treatment options for mucopolysaccharidosis: A survey to bridge the gap for personalized medicine	36-G
343	Anna-Maria Wiesinger	Development of a suspicion index tool to aid diagnosis of ASMD disease	37-C
349	Teresa Hoi-Yee Wu	A UK-based pre-pilot newborn screening study for metachromatic leukodytrosphy identified a late infantile case	37-G

352	Emily Yu	The impact of multiple sulfatase deficiency on children and families: A caregiver's perspective	38-C
LB-02	Kannan Alpadi	Targeted next generation sequencing for newborn screening of lysosomal disorders	38-G
LB-04	Swathi Ayloo	AAV-GBA1 mediated gene replacement for the treatment of Gaucher disease and GBA-PD	39-C
LB-05	Michael Babcock	Phase 1 healthy volunteer studies of AL01211 an oral, non-brain penetrant glucosylceramide synthase inhibitor, to treat Fabry disease and type 1 Gaucher disease	39-G
LB-10	Elizabeth Braunlin	Cardiac factors influencing pulmonary function in mucopolysaccharidosis (MPS): Report from a new registry	40-C
LB-12	Martino Calamai	Versatile flow cytometry assay for improving pre-diagnosis and pharmacological follow-up of patients affected by LDs	40-G
LB-23	Nima Fattahi	Outcomes of ERT/SRT monotherapy and combination therapy in neuronopathic Gaucher disease mouse model suggest transfer of peripheral ERT to the brain	41-C
LB-26	Zackary Herbst	Tandem mass spectrometric enzyme activity assay for simultaneous detection of Tay-Sachs and Sandhoff diseases in dried blood spots for newborn screening	41-G
LB-29	Monika Izdebski	Further characterization of GLA variants of uncertain significance	42-C
LB-30	Brett Johnson	Aligning the Pompe disease community to harness patient consented data to accelerate drug development	42-G
LB-33	Ashley Lahr	One center's experience of infants identified with variants of unknown significance during newborn screening for lysosomal diseases	43-C
LB-34	Kyle Landskroner	Nizubaglustat regulates GM1 ganglioside neuronal health in a human brain organoid model of GM1 gangliosidosis	43-G
LB-44	Petra Oliva	Results of prospective newborn screening for metachromatic leukodystrophy in Germany and Austria	44-C
LB-46	Allison Paltzer	Further clinical and biochemical phenotype of the GLA p.A143T variant	44-G
LB-48	Laith Refaei	Acid sphingomyelinase deficiency (ASMD) patient journey: Results from interviews with patients with ASMD highlight the challenges of diagnosis and care	45-C
LB-55	Sophie Thomas	Study to understand level of functioning and quality of life of children with Infantile lysosomal acid lipase deficiency, compared to children without the condition	45-G
LB-56	Shunji Tomatsu	An AAV Based Clinical Trial for mucopolysaccharidosis type IVA (2022-BGTC-005)	46-C
LB-60	Sarah Viall	Five years of newborn screening for Fabry disease and Gaucher disease in Oregon	46-G
LB-61	Knut Wittkowski	Updates on an alpha-cyclodextrin clathrate as a safe (not ototoxic) and convenient (oral) treatment of lysosomal diseases (LDs, incl. Batten and Niemann-Pick diseases)	47-C

Clinical Applications – Poster Session III – Wednesday, February 7, 3:00 PM - 5:00 PM

#	Author	Title	Kiosk
3	Jonathan Acevedo	Impact of anakinra on disordered movement in Sanfilippo syndrome	1-B
4	Maria Acosta	Comparing GM1 gangliosidosis patients treated and untreated with gene therapy: Longitudinal white matter changes in GM1 using differential tractography	1-F
5	Laura Adang	A phase 2 study assessing TAK-611 150 mg intrathecal weekly in patients with late-infantile metachromatic leukodystrophy (NCT03771898; SHP611-201; EMBOLDEN) compared to matched historical control data from children with late-infantile MLD (GLIA-MLD)	2-B
8	Julia Alton	Exploring the experience of females living with Fabry disease in North America	2-F
9	Carolina Aranda	Hypersensitivity reactions and enzyme replacement therapy: Outcomes and safety of rapid desensitization in 5,132 infusions	3-B
14	Charlotte Aries	Experiences and challenges of pregnancies in a patient with mucolipidosis type III	3-F
28	Manisha Balwani	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	4-B
32	John Bernat	Assessment of immunogenicity from the pegunigalsidase alfa clinical trial program: Integrated analysis of de novo and treatment-boosted anti-drug antibodies	4-F
38	Alexander Broomfield	Review of craniocervical monitoring and surgical intervention in paediatric mucopolysaccharidosis type VI in UK	5-B
41	Alessandro Burlina	Stroke in patients with Fabry disease: A Fabry Registry analysis of natural history data from patients stratified by disease phenotype	5-F
43	Barbara Burton	Somatic outcomes in a phase 1/2 study of weekly intravenous DNL310 (brain-penetrant enzyme replacement therapy) in mucopolysaccharidosis type II	6-B
44	Barry Byrne	NEO1/NEO-EXT studies: Long-term muscle quantitative magnetic resonance imaging and functional efficacy in adults with late-onset Pompe disease (LOPD) on avalglucosidase alfa treatment	6-F
47	Maria Camprodon-Gomez	Bone involvement outcomes in Spanish patients with Gaucher disease type 1 treated with eliglustat: Sub-study International Collaborative Gaucher Group (ICGG)	7-B
51	Magdalena Cerón-Rodriguez	Pediatric Fabry disease in Mexico: Genotype-phenotype relationship	7-F
54	Jae Yeong Cho	Fabry disease in female monozygotic twins with a complex intronic haplotype mutation: A case report	8-B
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60	Nicola Condon	Barriers and facilitators to physical activity and exercise in late-onset Pompe disease (LOPD): A qualitative study exploring patients' perspectives and experiences	9-B
61	James Cooper	PPCS, the biomarker with the best clinical utility for Niemann-Pick disease type B?	9-F
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71	Alícia Dornelles	Efficacy and safety of enzyme replacement therapy with alglucosidase alfa for the treatment of patients with infantile-onset Pompe disease: A systematic review and metanalysis	11-B
72	Lucy Dougherty De Miguel	Diagnostic strategies for hydrocephalus detection in MPS patients	11-F
76	Consuelo Durand	Quality of life follow-up of patients with mucopolysaccharidosis type VII in Argentina	12-B

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109	Roberto Giugliani	A natural history study of late-infantile and juvenile GM1 and GM2 gangliosidoses (PRONTO): Baseline clinical data	18-B
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112	Roberto Giugliani	Vestronidase alfa for the treatment of mucopolysaccharidosis type VII (MPS VII): Updated results from a novel, longitudinal, multi-center disease monitoring program (DMP)	19-F
113	Roberto Giugliani	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	20-B
115	Ozlem Goker-Alpan	Characteristics and management of US pediatric patients with Gaucher disease from the Gaucher Outcome Survey	20-F
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121	Christina Grant	Cardiac findings in neuronopathic MPS I: A case series	21-F
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124	Giuseppina Grillo	Fabry App: The value of a portable technology in recording patient monitored information in patients with Fabry disease	22-F
126	Nathalie Guffon	Alpha-mannosidosis international caregiver and patient survey: Changes in mobility, pain or discomfort, and patients' self-care over time	23-B
127	Nathalie Guffon	Long term efficacy of velmanase alfa treatment in patients with alpha mannosidosis: Pooled data from two extension studies (up to 12 years of therapy)	23-F

128	Paul Guijt	Progress in the IGA/IWGGD project on home therapy for Gaucher disease	24-B
129	Paul Guijt	Guidelines on self-management of Gaucher disease	24-F
131	Seo-Yeon Gwak	Impact of enzyme replacement therapy on cardiac function in patients with Fabry disease cardiomyopathy: A prospective multimodality imaging study using diastolic stress echocardiography and cardiac MRI	25-B
132	Andreas Hahn	Baby-COMET: Safety of avalglucosidase alfa after repeat dosing in treatment-naïve participants with infantile-onset Pompe disease (IOPD)	25-F
136	Caroline Hastings	Transport®NPC: open phase 3 global trial of intravenous hydroxy-propyl-beta-cyclodextrin in patients with Niemann-Pick disease type C1 (NPC1)	26-B
145	Robert Hopkin	Isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease: Updated results from an ongoing phase 1/2 study (STAAR)	26-F
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148	Derralynn Hughes	Results from GALILEO-1, a first-in-human clinical trial of FLT201 gene therapy in patients with Gaucher disease type 1	27-F
149	Derralynn Hughes	Tolerability of pegunigalsidase alfa across the clinical program: Integrated analysis of infusion-related reactions by dosing regimens	28-B
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167	Deepthy Joseph	Rare complication in late-onset Pompe disease: A case report	29-B
168	Ana Jovanovic	Evaluation of long-term renal outcomes in Fabry disease: A single centre analysis	29-F
170	Ilkka Kantola	Aging Fabry disease patients	30-B
176	Johnny Kenth	Novel instruments for evaluating postoperative outcomes in MPS IVA patients: A confluence of AI for thematic quality of life analysis and Bayesian respiratory function assessment	30-F
178	Priya Kishnani	Real-world experience of switching treatments from alglucosidase alfa to avalglucosidase alfa for patients enrolled in the Pompe Registry	31-B
179	Priya Kishnani	Using a novel measuring device to objectively evaluate the clinical benefit on ptosis in infantile-onset Pompe disease (IOPD): Results from 145 weeks' avalglucosidase alfa dosing in Mini-COMET participants previously treated with alglucosidase alfa	31-F
180	Motomichi Kosuga	Efficacy and safety of combination of HSCT & ICV ERT for neuropathic mucopolysaccharidosis type II	32-B
181	David Kronn	Mini-COMET study: Safety and efficacy data after avalglucosidase alfa dosing for ≥145 weeks in participants with infantile-onset Pompe disease (IOPD) who had demonstrated clinical decline or sub-optimal response while receiving alglucosidase alfa	32-F
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223	Ankit Mehta	Tolerability of pegunigalsidase alfa across the clinical program: Integrated analysis of infusion-related reactions by prior enzyme replacement therapy	37-B
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230	Elizabeth Morris	Filling the gap: Creation of a lysosomal disorders specific training programme for healthcare professionals	38-B
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232	Tahseen Mozaffar	Effect size analysis of cipaglucosidase alfa plus miglustat versus alglucosidase alfa in ERT-experienced adults with late-onset Pompe disease in PROPEL	39-B
233	Joseph Muenzer	Interim analysis of a phase 1/2 study of weekly intravenous DNL310 (brain-penetrant enzyme replacement therapy) in mucopolysaccharidosis type II	39-F
234	Nicole Muschol	A phase I/II clinical trial of JR-441 for treatment of Sanfilippo syndrome type A (MPS IIIA)	40-B
235	Nicole Muschol	A Delphi consensus approach to monitoring and integrated care coordination of patients with alpha-mannosidosis	40-F
244	Samantha Nishimura	15 year evaluation of prodromal Parkinson features in a cohort with homozygous and heterozygous GBAI mutations	41-B
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249	Paul Orchard	Design of a multi-center randomized active controlled phase 3 clinical trial (HURCULES) evaluating the safety and efficacy of OTL-203 in patients with MPS IH versus standard of care with allogeneic hematopoietic stem cell transplantation	42-B
250	Paul Orchard	Compassionate use of lentiviral gene therapy for metachromatic leukodystrophy	42-F
254	Sneha Patel	Patient-reported experiences during the SARS-CoV-2 (COVID-19): Results from a cross-sectional survey	43-B
258	Nishitha Pillai	Frequent GAG monitoring to evaluate the efficacy of enzyme replacement therapy in MPS IVA	43-F
260	Antonio Pisani	Clinical outcomes in patients switching from agalsidase beta to migalastat: A Fabry Registry analysis	44-B
262	Juan Politei	Prevalence of migalastat-amenable mutations in patients with Fabry disease from Brazil, Argentina and Colombia	44-F
265	Shaney Pressley	Off-label use of agalsidase beta and immune tolerance induction: The challenges of treatment initiation in young Fabry disease patients	45-B
270	Anika Quillin	Optimizing detection of early gastrointestinal symptoms in young children with Fabry disease	45-F
272	Deepa Rajan	Nerve conduction velocity studies in infantile Krabbe disease demonstrate long term continued progression of peripheral demyelinating neuropathy after hematopoietic stem cell transplantation (HSCT)	46-B
279	Mark Roberts	Baseline demographics of the UK Early Access to Medicines Scheme registry for cipaglucosidase alfa plus miglustat in enzyme replacement therapy-experienced adults with late-onset Pompe disease	46-F

280	David Rogers	Intravitreal enzyme replacement therapy to prevent retinal disease progression in children with neuronal ceroid lipofuscinosis type 2 (CLN2): Unilateral injection results	47-B
282	Ashwin Roy	Longitudinal changes in transthoracic echocardiography and biochemical markers in Fabry disease	47-F
286	Armaan Saith	Eliglustat substrate reduction therapy in Gaucher disease patients with cardiac comorbidities	48-B
287	Armaan Saith	Digenic disorders in patients with Gaucher disease: Implications for clinical management and study of modifier genes	48-F
288	Armaan Saith	Eliglustat substrate reduction therapy in pediatric patients with Gaucher disease	49-B
289	Luz Maria Sanchez	First experience with olipudase alfa for ASMD type B in Mexico	49-F
291	Maurizio Scarpa	Tailored diagnostic decision tree resulting from machine learning to improve early diagnosis of ASMD	50-B
293	Benedikt Schoser	Minimal clinically important differences in six-minute walking distance in late-onset Pompe disease	50-F
295	Angela Schulz	Cerliponase alfa for the treatment of CLN2 disease in a patient cohort including children under 3 years of age	41-H
306	Sofia Shrestha	Dose matters: Need for higher dose of avalglucosidase alfa in late-onset Pompe disease patients with disease progression on standard dose	42-D
308	Grace Stafford	IOPD entering adulthood: Lessons from two decades of ERT experience	42-H
311	Karolina Stepien	Service evaluation of respiratory tract dysfunction in adult Morquio syndrome (MPS IVA): A single tertiary multi-disciplinary centre experience	43-D
312	Karolina Stepien	Case series on patients with delayed diagnosis of mild/moderate alpha-mannosidosis	43-H
314	Bernhard Suter	NGLY1 deficiency natural history study	44-D
319	Antonio Toscano	COMET post hoc analysis: Efficacy of long-term avalglucosidase alfa in subgroups of participants with late-onset Pompe disease	44-H
320	Antonio Toscano	Switching treatment to cipaglucosidase alfa plus miglustat positively affects motor function and quality of life in patients with late-onset Pompe disease	45-D
326	Suresh Vijay	Survival achieved in infants with rapidly progressive LAL-D via sebelipase alfa ERT: Results from the International LAL-D Registry	45-H
332	Hua Wang	Transition from agalsidase beta to migalastat in a female Fabry disease patient with recurrent stroke-like episodes	46-D
333	John Wang	Atypical neurological presentation in Gaucher disease type 3C	46-H
335	WM Subadra Wanninayake	High sensitivity troponin T and I to evaluate Fabry disease cardiomyopathy	47-D
336	Melissa Wasserstein	Olipudase alfa enzyme replacement therapy reverses interstitial lung disease in adults with acid sphingomyelinase deficiency: Long-term pulmonary outcomes of the ASCEND trial	47-H
337	Melissa Wasserstein	Impact of homozygous p.Arg610del genotype on disease burden and treatment response in adults with acid sphingomyelinase deficiency in the ASCEND trial of olipudase alfa	48-D
338	Michael West	Switch from enzyme replacement therapy to pharmacologic chaperone: Improvement in advanced Fabry nephropathy	48-H
339	Michael West	FollowME Fabry Pathfinders Registry: Renal effectiveness in a cohort of patients on migalastat treatment for at least three years	49-D
340	Michael West	Initiation of pharmacologic chaperone therapy for Fabry disease in the Canadian Fabry Disease Initiative (CFDI) registry is not associated with reduction of kidney function	49-C

347	Michelle Wood	Baseline physiotherapy composite profile assessment (PCPA) as a predictor of functional outcomes in infantile-onset Pompe disease (IOPD) treated with enzyme replacement therapy in a UK paediatric centre	50-D
348	Kara Woolgar	Utilization of intravenous methylprednisolone to reduce neutralizing agalsidase-beta antibodies in two male pediatric Fabry disease patients	50-H
350	Shoji Yano	Pathohistological study of intracranial Gaucheroma causing deafness in a patient with Gaucher disease type 3: Effects of substrate reduction therapy	48-C
354	Ari Zimran	Real-world experiences with taliglucerase alfa home infusions for patients with Gaucher disease: A global cohort study	49-G

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13	Christian Argueta	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	1-D
24	Khashayar Azimpour	Estimating Fabry disease prevalence and treatment rates in the United States: An epidemiological analysis	1-H
30	Karen Bean	Cost-effectiveness framework by tandem mass spectrometry (TMS) for newborn screening of metachromatic leukodystrophy (MLD) in the United States (US)	2-D
33	Elizabeth Berry-Kravis	Real-world data collection in Niemann-Pick disease type C: Data from expanded access program with arimoclomol	2-H
34	Akhil Bhalla	DNL310 normalizes primary storage substrates, corrects biomarkers of lysosomal dysfunction and reduces biomarkers of neuronal injury (neurofilament light chain) in MPS II: 2-year interim analysis of a phase 1/2 study	3-D
37	Nidal Boulos	*Audiology assessment of participants in CAMPSIITETM, a phase I/II/III study of RGX-121 in neuronopathic MPS II*	3-H
39	Alberto Burlina	Unexpected high frequency of Krabbe disease detected by newborn screening in Italy	4-D
45	Umut Cagin	Influence of background enzyme replacement therapy on baseline biomarker profiles of patients with Fabry disease	4-H
48	Liching Cao	Anti-AAV6 antibody assay for patient enrollment supporting ST-920 phase 1/2 study for Fabry disease	5-D
62	Ana Crespo	Unveiling Fabry disease journey to diagnosis: An analysis based on two United States claims databases	5-H
67	Michael DiGruccio	Hyperactive GlcNAc-1-Phosphotransferase (S1S3 PTase) dramatically increases M6P levels on lysosomal enzymes for substantially improved receptor binding and cellular uptake	6-D
68	Hung Do	An innovative gene therapy approach to produce novel GALC variant with enhanced protein stability and enzyme activity with high levels of mannose 6-phosphate for Krabbe disease	6-H
70	Patricia Dorling	The burden of Fabry disease in the United States: Results from the Adelphi Fabry Disease Specific Programme	7-D
73	Alexandra Dumitriu	Pre- and post-diagnosis journey among commercially insured male and female Fabry disease patients in the United States	7-H
74	Alexandra Dumitriu	Fabry disease pre-treatment patterns in United States claims data	8-D
75	Alexandra Dumitriu	Real-world evidence study finds no new-onset diabetes or drug-related hyperglycemia in Pompe disease patients treated with avalglucosidase alfa	8-H
77	Stefan Ebner-Benke	Characterization of in vitro drug screening models for Gaucher disease based on patient-derived human fibroblasts	9-D
79	Eileen Elliott	Development of a first-in-class autologous B cell therapy for the treatment of Fabry disease	9-H
80	Natalie Engmann	DNL310 phase 1/2 case study demonstrates properties of raw, standard and growth scale scores for adaptive behavior scales	10-D
83	Vivian Fernandez	Caregiver perspectives on their MPS II journey	10-H
88	Taylor Fields	Results of a phase III, randomized, placebo-controlled crossover trial with N-acetyl-L-leucine for Niemann-Pick disease type C	11-D
89	Steffen Fischer	Ganglioside quantification in dried blood spots: A potential biochemical tool to diagnose and monitor metabolic and neurodegenerative diseases	11-H

94	Stuart Gaffney	Medical education needs to improve diagnosis of Fabry disease in the UK	12-D
97	Michael Gelb	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	12-H
98	Tarekegn Gerberhiwot	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	13-D
102	Aidan Gill	Sleep disturbance is an important feature of neurological lysosomal disease: For patients and families	13-H
103	Aidan Gill	Swallowing disturbance is an important feature of neurological lysosomal disease: For patients and families	14-D
107	Roberto Giugliani	A phase 2, randomized, double-blind, placebo-controlled, multicentre study to evaluate the efficacy and safety of pentosan polysulfate sodium in treating subjects with mucopolysaccharidosis VI: Study update	14-H
137	Andrew Hedman	Optimized dual promoter AAV gene therapy for lysosomal β-glucocerebrosidase with high M6P content for treatment of neuronopathic Gaucher disease	15-D
140	Christian Hendriksz	No patient should be left behind: Pilot project to expand lysosomal disorder diagnostics in Africa	15-H
154	Toshiaki Ikeda	A global phase III study of pabinafusp alfa (JR-141) for neuronopathic mucopolysaccharidosis type II: Updated study design	16-D
155	Atsushi Imakiire	Recovery of retinal function in MPS II mice by treatment with pabinafusp alfa	16-H
163	Dongkyu Jin	Therapeutic potential of intracerebroventricular recombinant human heparan-N-sulfatase enzyme replacement therapy in MPS IIIA mice	17-D
164	Dongkyu Jin	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	17-H
165	Franklin Johnson	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	18-D
192	Catherine Lawrence	Ethical decision-making for early access to investigational medicines in rare disease	18-H
196	Mindy Leffler	Objective assessment of functional abilities in lysosomal diseases using structured at-home video recordings	19-D
198	Mikhail Levit	Olink® proteomic profiling of biofluids from patients with Gaucher disease type 3 to understand disease pathology	19-H
203	Nicole Lyn	Assessing the impact of pain on health-related quality of life (HRQoL) in patients with Fabry disease	20-D
204	Nicole Lyn	Comparing treatment options for Fabry disease: Feasibility assessment for network meta-analysis (NMA)	20-H
205	Nicole Lyn	Measuring the intangible cost of lysosomal disorders: Fabry disease, Gaucher disease type 3, and GM2 gangliosidosis	21-D
208	Irina Maksimova	Different monitoring patterns in treated and untreated patients with Fabry disease: Analysis of a United States claims database	21-H
210	Betsy Malkus	Performance measures and patient/caregiver-reported assessments collected in a longitudinal, multi-center disease monitoring program (DMP) of patients with mucopolysaccharidosis type VII (MPS VII)	22-D
220	Renata Medinaceli Quintela	Use of wearable sensor technology to identify digital biomarkers for monitoring gait parameters in children and adolescents with GM1 and GM2 gangliosidoses	22-H

224	Kathleen Meyer	A 3-month gene therapy single-dose IV administration pharmacology and safety study with ST-920 (isaralgagene civaparvovec) for Fabry disease in mice	23-D
225	John Mitchell	Co-developing The Canadian MPS Registry: A longitudinal rare disease patient registry	23-H
229	Hiroki Morioka	Enzyme replacement therapy with a blood brain barrier-penetrating antibody-fused alpha-L-iduronidase prevents bone deformities in a mouse model of mucopolysaccharidosis type I	24-D
236	Monika Musial-Siwek	*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*	24-H
241	Pooja Nandi	Cardiac biomarkers in Fabry disease	25-D
268	Ruth Pulikottil-Jacob	Psychometric validation of Pompe Disease Symptom Scale (PDSS) and Pompe Disease Impact Scale (PDIS) in patients with late-onset Pompe disease: A real-world evidence study	25-H
269	Ruth Pulikottil-Jacob	Content validation of the modified Friedreich Ataxia Rating Scale-Activities of Daily Living (mFARS-ADL) for use with Gaucher disease type 3 patients: A qualitative study	26-D
273	Shyam Ramachandran	AAV-ARSA mediated gene replacement for the treatment of metachromatic leukodystrophy	26-H
274	Shyam Ramachandran	Evaluation of a novel AAV capsid with widespread CNS and peripheral biodistribution in non-human primates	27-D
276	Vanessa Rangel Miller	Parallel biochemical and genetic testing informs a timely and accurate diagnosis of MPS VII: Findings from 5 years of sponsored testing programs	27-H
303	Shirin Sharghi	Histological characterization of the 6neo mouse model of Pompe disease	28-D
304	Seung-Yub Shin	Development of YH35995A, a novel highly potent and BBB-penetrating GCS inhibitor for the treatment of Gaucher disease	28-H
318	Sophie Thomas	The impact of Fabry disease symptoms on patient's quality of life (QOL) and mental health: A qualitative interview study in the UK	29-D
323	Arjan van der Flier	Anti-human-TfR-GAA efficiently clears CNS and muscle glycogen in a translatable hTfR-KI/ Pompe disease mouse model	29-H
325	Bastien Vidal	Creation of a versatile therapeutic platform using autologous gene-engineered B cell protein factories to make durable therapies for patients with genetic diseases	30-D
327	Ashley Volz	Skeletal dysplasia gene panel with integrated enzyme follow-up for the diagnosis of lysosomal disorders: MPS IVA case series	30-H
329	Mark Walzer	A study to evaluate seroprevalence of antibodies to AAV8 and biomarkers in patients with late-onset Pompe disease: Rationale and study design	31-D
346	Christopher Wingrove	Exploring the journey of patients with Fabry disease in Brazil	31-H
353	Natalya Zhurkova	Lysosomal acid lipase deficiency	32-D
LB-03	Roberto Araujo	A paradigm shift in collecting RWD for GM2 gangliosidosis	32-H
LB-08	Lisa Berry	Unanticipated diagnosis of mucopolysaccharidosis type IIIB in the setting of normal intelligence	33-D
LB-09	Niels Brandt-Jacobsen	Annual echocardiography among all patients with Fabry disease, or time for personalized clinical workout?	33-H
LB-11	Barbara Burton	Enzyme replacement therapy is associated with improved endurance in MPS IVA patients at different ages	34-D
LB-13	Candace Cameron	A natural history study of Sanfilippo syndrome type D: Retrospective data review	34-H
LB-15	Nan Chen	A phase 4, open label, multicenter study of the safety and efficacy of agalsidase beta in Chinese patients with Fabry disease	35-D

LB-18	Alejandra de la Torre Fernandez	Malignancies in Gaucher disease: 20 year follow-up	35-H
LB-21	Maria Escolar	Reklaim, a novel phase IB clinical trial of FBX101 (AAVrh10.galc) intravenously administered after UCBT for the treatment of infantile Krabbe disease	36-D
LB-35	Heather Lau	Reduction of heparan sulfate (HS) exposure in cerebrospinal fluid (CSF) correlates with improved long-term cognitive function in patients with mucopolysaccharidosis type IIIA (MPS IIIA) following treatment with UX111 gene therapy	36-H
LB-38	Aytan Mammadbayli	Progressive myoclonus epilepsy in patient with KCTD7 mutation.	37-D
LB-39	Ana Maria Martins	The impact of lepunafusp alfa (JR 171) on the disease burden in MPS I: Patient reported outcomes	37-H
LB-40	Wladimir Mauhin	Burden of illness in acid sphingomyelinase deficiency type B and type A/B: A multinational retrospective chart review study of 270 patients	38-D
LB-41	Daisy Ng-Mak	Motor function limitations and impacts among patients with GM1 and GM2 gangliosidoses in the United States: A qualitative study	38-H
LB-42	Kaila Niehaus	A 4 year old with Niemann-Pick disease type C2: Clinical course and treatment	39-D
LB-43	Albina Nowak	Resilience in patients with Fabry disease and its association with disease course, psychosocial factors and quality of life: A multicentre cross-sectional study	39-H
LB-53	Thanyachai Sura	Rare diseases (RD): The importance of early detection and treatment in Ramathibodi Hospital Thailand	40-D
LB-54	Mark Thomas	Phase 1/2 clinical trial evaluating 4D-310 in adults with Fabry disease cardiomyopathy: Interim analysis of cardiac and safety outcomes in patients with 10-32 months of follow-up	40-H
LB-59	Jaime Vengoechea	Switch back to alglucosidase alpha from AVA due to IRR: No cross-reactivity	41-D