Poster presenters are assigned to present their abstract Live (In Person) at one of eight sessions, based on the final abstract category for the abstract. All posters will be available to all attendees beginning at 3:00 PM EST on Wednesday, February 22, 2023, on the WORLD*Symposium* mobile app and will remain open throughout WORLD*Symposium* 2023. Live Q&A will only occur onsite during the assigned times as listed below.

All poster presentations will be in the Exhibit Hall (Orlando Ballroom) during assigned times. Each daily poster reception will include two separate 1-hour-long sessions. The first session will be from 3:00-4:00 PM, and the second session will be from 4:00-5:00 PM.

Basic Science Posters will be presented in two 1-hour sessions on **Wednesday, February 22** from 3:00-5:00 PM EST
Translational Research Posters will be presented in two 1-hour sessions on **Thursday, February 23** from 3:00-5:00 PM EST
Clinical Applications Posters will be presented in two 1-hour sessions on **Friday, February 24** from 3:00-5:00 PM EST
Contemporary Forum Posters will be presented in two 1-hour sessions on **Saturday, February 25** from 3:00-5:00 PM EST

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 – Wednesday, February 22, 3:00 PM - 4:00 PM

#	Author	Title	Kiosk
12	Rebecca C. Ahrens-Nicklas	Biomarkers of disease severity in multiple sulfatase deficiency	1-A
15	Gheona Altarescu	Whole blood transcriptomic profiling in the interpretation of variable phenotype presentation in Fabry disease	2-A
22	Isidro Arévalo-Vargas	Glycosphingolipids assessments of Gaucher disease patients from dried blood spots samples by LC/MS–MS	3-A
24	Marta Artola	*1,6-epi*-cyclophellitol cyclosulfamidate is a new superior lysosomal α-glucosidase stabilizer for the treatment of Pompe disease	4-A
25	Daniela Arturo Terranova	Correlation between bone manifestations and variants of the GBA gene in Gaucher disease: A systematic review	5-A
26	Frederick Ashby	Bone pathology within Sanfilippo syndrome type B mice as a novel biometric for peripheral disease correction	6-A
29	Frederick Ashby	Neural stem cell biodistribution visualization by 3D immunolabeling in Sanfilippo syndrome disease models	6-B
30	Imane Assiri	Timely diagnosis of Niemann Pick disease using thin layer chromatography with identification of SMPD1 mutations causing NPD type A/B among Moroccan population	5-B
37	Mohsen Basiri	Osteonecrosis in the era of Gaucher disease therapies	4-B
39	Mohsen Basiri	The risk of hepatocellular carcinoma is markedly increased in Gaucher disease	3-B
46	Hannah Best	Disease phenotypes present in CLN7 patient fibroblasts are improved following treatment to reduce glycosphingolipid synthesis	2-B
49	Ritika Bhat	Podocyturia correlates with reduced estimated glomerular filtration rate (eGFR) and increased albuminuria in Fabry disease (FD)	1-B
50	Constanza Bondar	Expression of caspase-3 and TGF- β1 in Fabry disease biopsies: Correlation with clinical parameters	7-A
52	Alice Brinckmann Oliveira Netto	Prenatal diagnosis of Pompe disease	8-A
62	María Castellanos	Cardiac disease in patients with mucopolysaccharidosis type II	9-A

69	Pan Chen	Beyond improving knowledge and competence: How continuing medical education closes gaps in the management of late-onset Pompe disease	10-A
70	Yun-Ru Chen	Development of a gene therapy for cardiac type Fabry disease: A gene editing strategy	11-A
71	Chihya Cheng	Evaluate the efficacy of small molecule compounds derived from drug repurposing using cardiac type Fabry disease cell model	12-A
78	Sophie Cook	Investigating lysosomal and cellular phenotypes of the lysosomal disorder cystinosis	12-B
82	Andrea Crivaro	Bone marrow adipocytes alteration in an *in vitro* model of Gaucher disease	11-B
86	Mari Davies	Acid ceramidase inhibition as a mechanism to treat lysosomal disorders	10-B
89	Francisco del Castillo	Next-generation sequencing and high-throughput enzymolome-based panel screening of suspected lysosomal disease cases identifies multiple disease-causing variants in two genes underlying different lysosomal diseases: A source of clinical and metabolic variability?	9-B
92	Chloé Dias	Microglia-derived extracellular vesicles promote neuropathology in Sanfilippo syndrome	8-B
97	Tama Dinur	Contribution of lyso-Gb1 to treatment decisions in patients with Gaucher disease	7-B
103	María Duarte	*In-vitro* evaluation of a human recombinant iduronate-2-sulfatase produces in the yeast *Komagataella phaffii*	13-A
104	Victor Duarte	*In vitro* evaluation of hydrolytic activity of two recombinant N-acetylglucosaminidases as potential therapeutic tools for mucopolysaccharidosis type IIIB	14-A
105	Hatim Ebrahim	Assessments in female with hypertrophic cardiomyopathy: Incongruous indicators of Fabry disease with valuable lessons	15-A
106	Ulla Feldt-Rasmussen	Can the use of urinary and plasma globotriaosylsphingosine and their analogues as diagnostic biomarkers be confirmed in male patients with Fabry disease?	16-A
114	Gareth Fenn	Development of a novel anthraquinone-derived fluorescent lysosomal probe	17-A
124	Maria Fuller	Signature biomarkers for diagnosis, screening, and biochemical monitoring of the mucopolysaccharidoses	18-A
131	Pilar Giraldo	Extensive femur location and homogeneous infiltration pattern are predictive of severity of bone disease: Re-assessing MRI bone marrow involvement in Gaucher disease through machine learning	18-B
137	Adenrele Gleason	Analysis of extracellular vesicles (EVs) as a biomarker for Parkinsonism in *GBA1* mutation carriers	17-B
141	Silvia Gonzalez Nieves	Substrate reduction therapy for Morquio syndrome type A: An *in vitro* evaluation	16-B
148	Anthea Guha	Inflammation in LAL deficiency is driven by substrate accumulation	15-B
163	Jacqueline Hernandez	Utilizing CRISPR/Cas9 to insert a functional copy of the SGSH gene into MPS IIIA derived induced pluripotent stem cells (iPSC)	14-B
164	Ellen Hertz	iPSC-derived neuronal models from sibling pairs with Gaucher disease discordant for Parkinson disease	13-B
165	Rachel Hickey	Newborn screening for acid sphingomyelinase deficiency in Illinois: A single center's experience	19-A
167	Llinos Honeybun	Unbiased phenotypic drug screen for CLN3 disease	20-A
183	Majdolen Istaiti	Prevalence of cancer among 867 patients with Gaucher disease from the SZMC Gaucher unit	21-A
184	Margarita Ivanova	Gender differences in circulating inflammatory, immune, & tissue growth markers associated with Fabry disease-related cardiomyopathy	22-A
185	Skyler Jackson	Understanding the functional burden of CLN3 through the eyes of patients and families	23-A
196	Neil Kasaci	Caspase inhibitors can counteract inflammasome activation and caspase-1 mediated fibrosis in Fabry disease	24-A

197	Megan Keating	Single-centre prospective clinical and procedural analysis identifying causes of delay to haematopoietic stem cell transplant in MPSI Hurler syndrome patients	24-B
198	Ayse Kiliç	Retargeting phenylbutyrate, ursodeoxycholic acid, pyrimethamine and betaine for beta-glucocerebrosidase recovery in Gaucher disease fibroblasts resulting from homozygous p.L483P mutation	23-B
211	Lucia Lavalle	Exploring alfa-galactosidase A protein levels and its impact on Hsp60 and serum mitokines	22-B
212	Steven Le	Membrane-tethered form of NAGLU used to elucidate pathogenesis of heparan sulfate in MPS IIIB mice	21-B
213	Andrés Felipe Leal	Magnetite nanoparticles as a vehicle to transport recombinant hexosaminidase A and B through an *in vitro* model of the blood-brain barrier	20-B
214	Maria Dolores Ledesma	Sphingomyelin 16:0 is a therapeutic target for brain pathology in acid sphingomyelinase deficiency	19-B
216	Paloma Lemaitre Gómez	Evaluation of CRISPR/nCas9 for gene editing on mucopolysaccharidosis type IIIB	25-A
219	Malte Lenders	Pre-existing anti-drug antibodies in Fabry disease show less affinity and inhibitory capacity for pegunigalsidase-alfa	26-A
220	Daniel Lewi	Cherry red spot: Evidence that its identification can speed up the diagnosis of GM2 gangliosidosis	27-A
222	Jens Lichtenberg	Detecting secondary modifiers in *GBA1* -related Parkinsonism via variant calling in sibling pairs	28-A
224	Yi Lin	Earlier-onset, more severe neurodegeneration in PGRN KO mice with a decreased dose of D409V *Gba1*	29-A
228	Juan Llerena Jr.	Challenges and hurdles to cope with Pompe disease in the Brazilian Unified Health System (SUS) frame: Lessons learned from a series of 19 patients from a national Rare Disease Reference Center in the state of Rio de Janeiro, Brazil	30-A
229	Emyr Lloyd-Evans	Glycosphingolipid reduction with miglustat as a therapeutic strategy for CLN3 and other neuronal ceroid lipofuscinoses	30-B
262	Andrew Oldham	A review of the clinical progression in six late onset Pompe disease (LOPD) patients following alglucosidase alfa cessation	29-B
280	Magali Pettazzoni	Acid sphingomyelinase deficiency: Epidemiologic and genetic aspects of a French cohort 1974-2021	28-B
313	S Christy Rohani-Montez	Education needs in diagnosing rare inherited metabolic disorders: A clinician survey	27-B
369	Helen Waller-Evans	Filipin composition and imaging modality dramatically affect cellular cholesterol visualisation with implications for assessing cholesterol levels and localisation across the lysosomal disorders	26-B
LB-37	Gustavo Maegawa	Peripheral extracellular vesicle as a potential source of sounding biomarkers for Gaucher disease	25-B

Basic Science – Poster Session II – Wednesday, February 22, 4:00 PM - 5:00 PM

#	Author	Title	Kiosk
27	Frederick Ashby	Differential metabolomic pathway response to the ketogenic diet in Sanfilippo syndrome type B mice	1-A
38	Mohsen Basiri	Severe pulmonary arterial hypertension in Gaucher disease type 1	2-A
47	Hannah Best	The Batten disease associated protein CLN3 is required for the efflux of lysosomal K^+	3-A
53	Alice Brinckmann Oliveira Netto	Utilization of next generation sequencing technology for detection of mosaic variants in the IDS gene	4-A
79	Sophie Cook	Comparing the lysosomal biotoxicity of iron oxide nanoparticles for improved lysosomal purification	5-A
90	Francisco del Castillo	Identification of the first genetic variants underlying metachromatic leukodystrophy in the Moroccan population by biochemical analyses and NGS-based genetic screening	6-A
115	Gareth Fenn	Investigation of cannabidiol as a potential therapeutic in Niemann-Pick disease	6-B
230	Emyr Lloyd-Evans	Deciphering the various mechanisms of action of miglustat in the lysosomal disorders	5-B
232	Megan Loden	Raregivers™ Emotional Journey Map presented by ANGEL AID Cares	4-B
233	Mary Kate LoPiccolo	Skin α-synuclein seeding activity in adult patients with Gaucher disease type 1	3-B
238	Angela Martin Rios	Beta mannosidosis detected through deafness panel: An ultra-rare disease as a cause of a common disease	2-B
246	David Moreno Martinez	Monocyte dysfunction in Fabry disease: Preliminary results from analysing integrin and non-integrin pathways show correlation with cardiac manifestations and a differential trend with age	1-B
250	Shiny Nair	Single cell resolution of neurodegeneration in Gaucher disease	7-A
251	Behzad Najafian	The spectrum of podocyte injury in later onset (LO) variants of Fabry disease (FD)	8-A
252	Samira Najeh	Diagnosis of vitamin B deficiencies inducing inherited metabolic diseases (IMD) using GC-MS	9-A
263	Andrew Oldham	Females with late onset Pompe disease: Clinical manifestations and rate of disease progression with no treatment	10-A
272	Saida Ortolano	PBXs: New pharmacological chaperones to increase α-galactosidase A activity in Fabry disease cellular models	11-A
278	Allyson Peek	Differential expression and localization of NEU1 and NEU4 in the CNS of mouse models of GM2 gangliosidoses	12-A
279	Fernando Perretta	Fabry disease: Lyso-Gb3 normalization as a reachable therapeutic goal	12-B
281	Magali Pettazzoni	Overview of Niemann-Pick type C disease in France 1975-2020: Evolution in diagnostic strategy, molecular genetics profiles and phenotypic correlations	11-B
282	Magali Pettazzoni	LC-MS/MS quantification of three C16 sulfatide species in dried blood spots for the diagnosis and treatment monitoring of metachromatic leukodystrophy	10-B
284	Vi Pham	Optimizing cross-correction to overcome limitations of *ex vivo* gene therapy in multiple sulfatase deficiency	9-B
286	Tyler M. Pierson	Modeling CLN6 with IPSC-derived neurons and glia	8-B
292	Jackson Pountney	Quality of life in patients with Niemann-Pick disease type C and their carers	7-B
293	Luisa Prada	Pharmacological chaperones as an alternative to increase GALNS activity in mucopolysaccharidosis type IVA	13-A
305	Shoshana Revel-Vilk	Brain-derived neurotrophic factor (BDNF) associated with platelet activity and bleeding tendency in patients with Gaucher disease	14-A

306	Cecilia Riccheri	Identification of genetic variants in 79 Argentinean patients with mucopolysaccharidosis type II, Hunter syndrome	15-A
314	Christy Rohani-Montez	Case-based education on Gaucher disease significantly improves physician competence in appropriate next steps after suspecting the diagnosis	16-A
316	Paula Rozenfeld	Lyso-Gb3 determination in classic and late onset Fabry patients	17-A
329	Markus Schwarz	Results of a prospective high risk population study for the frequency of α-mannosidosis within MPS like phenotype patients in Europe and the Middle East	18-A
331	Irene Serrano Gonzalo	Study of miRNA expression profiles depending on the severity of bone involvement in patients with Gaucher disease	18-B
332	Volkan Seyrantepe	Elimination of the *B4Galnt1* gene normalizes lifespan and prevents pathology in Tay-Sachs disease mice	17-B
333	Alex J. Shamoun	Differences in organ abundance of iduronate 2-sulfatase and intravenous recombinant enzyme delivery: Potential implications for clinical response to ERT in MPS II	16-B
337	Calogera Simonaro	Joint and skeletal pathology in acid sphingomyelinase deficient mice	15-B
339	David Smerkous	Development of an online cloud-based tool for automatic measurement of foot process width (FPW) using deep learning (DL): Applications in assessment of podocyte injury in Fabry disease (FD)	14-B
341	Linda Spencer	A case of a presumed diagnosis of Morquio syndrome	13-B
343	Karolina Stepien	Iron deficiency anaemia in adult mucopolysaccharidoses	19-A
356	Emilio Vaena	Generation of an *in vitro* model for Hunter syndrome using CRISPR/Cas9 technology	20-A
361	Nato Vashakmadze	Alfa-mannosidosis: Six clinical cases	21-A
362	Dana Velasquez Rivas	LysoGb3 in children under 3 years of age with classic Fabry disease phenotype: Is there a clinical correlation?	22-A
370	Helen Waller-Evans	Measuring ion flux via lysosomal channels: Primary and secondary assays for drug discovery across the lysosomal disorders	23-A
383	Zhenting Zhang	A multifaceted evaluation of microgliosis and differential cellular dysregulations of mTOR signaling with fluctuating lysosome function in neuronopathic Gaucher disease	24-A
385	Natalya Zhurkova	Mucopolysaccharidoses in 81 patients: Neurological aspects	24-B
386	Ari Zimran	Lyso-Gb1 as a biomarker of the real-world situation in Gaucher disease: Comparative data from the Gaucher Outcome Survey (GOS) in ERT-treated and untreated patients	23-B
LB-30	Youngil Koh	Genetic alteration related to lysosome function contributes to pancreatic cancer development	22-B
LB-31	Thomas Kukar	Granulins are bioactive and ameliorate neuropathology in a mouse model of neuronal ceroid lipofuscinosis	21-B
LB-33	Andrés Felipe Leal	Assessment of an iron oxide-coupled CRISPR/nCas9 gene editing on mucopolysaccharidoses type IVA mouse model	20-B
LB-38	Gustavo Maegawa	Characterization of small molecules as potential therapeutic agents for Krabbe disease	19-B
LB-48	Maximiliano Ormazabal	Generation of Fabry disease cardiomyocyte models using CRISPR-Cas9 technology	25-A
LB-68	Nigel Wade	Examining the effects of ATP10B knockout and over-expression on lysosomal function and lipid accumulation in Gaucher disease	26-A
LB-73	Yanping Zhu	Quantifying lysosomal glycoside hydrolase activity using fluorescence-quenched substrates in physiologically relevant cellular models	27-A

Translational Research – Poster Session III – Thursday, February 23, 3:00 PM - 4:00 PM

#	Author	Title	Kiosk
5	Laura A. Adang	Development of disease-specific scale for multiple sulfatase deficiency	1-A
9	Patricio Aguiar	Estimation of arrhythmia risk in patients with Fabry disease using a machine learning model	2-A
10	Alia Ahmed	Long-term intravenous galsulfase in Maroteaux-Lamy syndrome further reduces urine glycosaminoglycans (GAG) after hematopoietic stem cell transplantation and improves cardiac function and endurance	3-A
19	Yin-Hsiu Chien	CK-MM as a second-tier test for Pompe disease newborn screening	4-A
28	Frederick Ashby	Genetic barcoding identifies similar transduction efficiency rankings within disease models of Sanfilippo syndrome type B and controls	5-A
32	Christiane Auray-Blais	Tandem mass spectrometry analysis of dried urine spots for newborn screening of mucopolysaccharidoses	6-A
35	Rafael Badell-Grau	Hematopoietic stem cell gene therapy for mucopolysaccharidosis type IIIC	6-B
45	Lisa Berry	Patient-reported experiences with Fabry disease monitoring and disease burden in the real-world setting: Results from a double-blind, cross-sectional survey	5-B
55	Laura Buch	Newborn screening for Pompe disease in South Carolina: Outcomes of early detection for late-onset Pompe disease phenotypes	4-B
56	Alberto Burlina	Newborn screening for Gaucher disease in Italy: Birth prevalence and outcome	3-B
60	Valeria Calbi	Blood sulfatides as disease biomarker for metachromatic leukodystrophy: Disease characterization, early diagnosis, and response to treatment	2-B
64	Betul Celik	Lentiviral gene therapy for mucopolysaccharidosis type IVA	1-B
65	Harriet Chang	A long-term course of gait assessment in the atypical CLN2 patients along with ICV cerliponase alpha treatment	7-A
84	Julia Dao	TRAP5a and TRAP5b biomarkers in Gaucher disease	8-A
87	Maria Jose de Castro Lopez	Cardiac phenotype in Hunter syndrome widening the spectrum	9-A
91	Ankit Desai	Development of high sustained IgG antibody titers against rhGAA in patients with infantile Pompe disease is associated with a shift toward Th2 immune activation	10-A
100	Aimee Donald	Sustained improvement of clinical CNS and somatic features of Gaucher disease type 3 after haematopoietic stem cell (HSC) gene therapy: A first-in-world report	11-A
101	Conan Donnelly	Development of a patient-reported data collection system for Niemann-Pick disease	12-A
123	Stephanie French	Validation of an assay to measure iduronate-2-sulfatase activity in cerebrospinal fluid to assess the efficacy of a hematopoietic stem cell (HSC) gene therapy	12-B
126	Jillian Gallagher	From mouse to sheep: Generating a sheep model and developing a gene therapy for sialidosis	11-B
128	Michael H. Gelb	Newborn screening for metachromatic leukodystrophy (MLD): An overview of ongoing and future studies	10-B
140	Antonio Gonzalez-Meneses Lopez	Personal perspectives of international caregivers and a clinican of the impact of a lack of newborn screening for MPS VII	9-B
143	Vincenza Gragnaniello	Effectiveness of corticosteroid treatment in a case of perinatal lethal Gaucher disease diagnosed by newborn screening	8-B
144	Christina Grant	Case comparison of 3 infants diagnosed with MPS I by newborn screen and impact of secondary screening	7-B

145	Emily Groopman	The ClinGen Lysosomal Diseases Gene Curation Panel: Applying a standardized curation framework to assess the clinical validity of genes for lysosomal disease	13-A
153	Andreas Hahn	Treatment of CLN1 disease with a blood-brain barrier penetrating lysosomal enzyme AGT-194	14-A
162	Kim M. Hemsley	A prohibitin-targeting drug modifies aspects of disease in a mouse model of Sanfilippo syndrome	15-A
177	Marjan Huizing	A concerted action to explore therapies for free sialic acid storage disease	16-A
178	Sarah Hurt	Elucidating expression of a corrective enzyme for MPS I through administration of an adenoviral vector targeted to endothelial cells	17-A
188	Simon Jones	Sustained biochemical engraftment and early clinical outcomes following *ex-vivo* autologous stem cell gene therapy for mucopolysaccharidosis type IIIA	18-A
192	Staci Kallish	Estimation of stroke risk in patients with Fabry disease using a machine learning model	18-B
208	Karima Lafhal	The case of glycosylation defects mimicking Wilson disease: A case report	17-B
209	Florian Lagler	Specific medical simulation trainings for parents of MPS patients: An innovative approach and its psychological benefits	16-B
217	Layzon Antonio Lemos da Silva	Screening for mucopolysaccharidoses' subtypes by tandem mass spectrometry determination of disease-specific oligosaccharides in dried blood spots	15-B
218	Malte Lenders	Assessment of neutralizing anti-drug antibodies in patients with Fabry disease and impact of dose escalation on individual titers	14-B
235	Anna Luzzi	Decreased regulatory T-cells in patients with Sanfilippo syndrome may allow the development of autoimmune disease	13-B
245	Lina Moreno Giraldo	Developmental and epileptic encephalopathy (DEE): Identification of a probably pathogenic variant of autosomal dominant inheritance in the SCN1A gene	19-A
253	Kimitoshi Nakamura	Newborn screening for Gaucher disease in Japan	20-A
264	Petra Oliva	Diagnostic approach for Fabry disease	21-A
271	Paul J. Orchard	Compassionate use of OTL-200 for patients with metachromatic leukodystrophy	22-A
295	Maximiliano Presa	Efficacy of syngeneic bone marrow transplant for the treatment of multiple sulfatase deficiency	23-A
296	Michael J. Przybilla	Treating murine Hurler syndrome utilizing small-activating RNA following bone marrow transplant	24-A
310	Eleanor Rodriguez-Rassi	Early initiation of ERT facilitated by newborn screening improves health outcomes among infantile Pompe disease patients in the US	24-B
312	Jaehyeok Roh	Potential brain penetrant N-acetylcysteine derivative can mitigate oxidative stress in Gaucher disease	23-B
317	Anne Rugari	A novel and effective process for brain and tissue banking for rare diseases	22-B
318	Michelle Saggers	Challenges and strategies with international recruitment in early phase gene therapy trials	21-B
328	Roselena S. Schuh	Nasal administration of laronidase-loaded liposomes aiming at mucopolysaccharidosis type I treatment	20-B
330	Markus Schwarz	High-risk population screening for the mucopolysaccharidoses (MPS)	19-B
338	Siyamini Sivananthan	Metachromatic leukodystrophy: A single centre review of features at diagnosis and barriers to accessing treatment	25-A
342	Richard Steet	Functional characterization of IDUA variants identified by newborn screening	26-A

346	Berthold Streubel	Results from two year pilot study for identifying inherited myopathies by combining enzymatical testing with “clinical symptomatic-based next-generation sequencing”	27-A
347	Dean Suhr	The power of a rare disease count and why an overcount is not helpful	28-A
350	Keigo Takahashi	Spontaneous seizures associated with cortical interneuron loss in *Cln2*^{R207X} mice are ameliorated via gene therapy	29-A
353	Laura Tobin	Gallbladder abnormalities as an early indicator of metachromatic leukodystrophy (MLD): Use of electronic health records in a large pediatric hospital to aid early diagnosis	30-A
359	Diantha van de Vlekkert	AAV-mediated gene therapy in a mouse model of sialidosis	30-B
364	Jesus Villarrubia	Ecological study to determine the estimated prevalence of patients with acid sphingomyelinase deficiency in Spain: PREVASMD study	29-B
375	Anna-Maria Wiesinger	A precision medicine tool for high utilization and quality of individual treatment trials with immunomodulatory drugs in mucopolysaccharidosis	28-B
LB-05	Tsun Au Yeung	Measuring enzyme activities of I2S with ABG, ASM, GAA, GALC, GLA and IDUA enzyme activities using the PerkinElmer QSight® 225MD UHPLC screening system	27-B
LB-07	Michael Babcock	Development of AL00804, a novel brain penetrant glucosylceramide synthase inhibitor, to treat Gaucher disease and other neuronopathic glycosphingolipid storage diseases	26-B
LB-34	Hsiang-Yu Lin	Newborn screening for mucopolysaccharidosis type II in Taiwan and long-term follow-up of the screen-positive subjects	25-B

Translational Research – Poster Session IV – Thursday, February 23, 4:00 PM - 5:00 PM

#	Author	Title	Kiosk
6	Laura A. Adang	Developmental delay can precede neurologic regression in metachromatic leukodystrophy	1-A
102	Conan Donnelly	A global survey to investigate experiences of Niemann-Pick disease type C patients and their caregivers	2-A
267	Petra Oliva	Strategy for diagnosis of neuronal ceroid lipofuscinoses	3-A
366	Sandra Vranic	Defect-free graphene enhances enzyme delivery to fibroblasts derived from the patients with lysosomal disorders	4-A
373	Jason A. Weesner	Preclinical enzyme replacement therapy with a recombinant β-galactosidase-lectin fusion for CNS delivery and treatment of GM1-gangliosidosis	5-A
376	Anna-Maria Wiesinger	A Suspicion Index Tool (SIT) to aid diagnosis of ASMD disease: Design and first results	6-A
377	Kaylee Williams	A review of provider experiences with newborn screening for Krabbe disease in Pennsylvania	6-B
378	Shaun Wood	Identification of novel peptide sequences to fuse to iduronate-2-sulfatase for the treatment of Hunter syndrome	5-B
379	Tim Wood	Measurement of urinary glycosaminoglycans via LC-MS/MS: Comparison of HILIC and reverse phase methods and integration of MPS biomarkers	4-B
LB-01	Ibane Abasolo	Preclinical validation of nanoliposomal ERT for Fabry disease	3-B
LB-02	Caterina Abdala Villa	Utilization of biomarkers for mucopolysaccharidosis type I (MPS I) phenotype interpretation in a complex positive newborn screen case	2-B
LB-03	Maria Acosta	Search for reliable biomarkers to predict clinical outcomes: The GM1 gangliosidosis gene therapy experience	1-B
LB-06	Tsun Au Yeung	Time study for measuring I2S enzyme activity using a PerkinElmer QSight 225MD UHPLC	7-A

LB-08	Michael Babcock	Development of AL01211, an oral, non-brain penetrant glucosylceramide synthase inhibitor (GCSi), to treat Fabry disease	8-A
LB-13	Tierra Bobo	Development of self-complementary AAV-IDUA vector for treating MPS IH by compact genome engineering	9-A
LB-14	Heather Brown	Pre-pilot bloodspot newborn screening study for metachromatic leukodystrophy in Manchester, United Kingdom	10-A
LB-15	Betul Celik	The effectiveness of lentiviral vectors under different promoters in MPS IVA patient fibroblasts, HEK293T and HepG2 cells	11-A
LB-17	Gary Ngai Yin Chan	RGX-381 gene therapy for the treatment of ocular manifestations of late-infantile neuronal ceroid lipofuscinosis type 2 (CLN2 disease): Overview of nonclinical development program	12-A
LB-18	Joslyn Crowe	The burden of fear in ASMD	12-B
LB-19	Patricia I. Dickson	Intraventricular recombinant human N-acetylglucosamine-6-sulfatase corrects lysosomal storage in mucopolysaccharidosis type IIID mice	11-B
LB-20	Deborah Elstein	Lasting impact of the COVID-19 pandemic on lysosomal disease patient care: Results from a large multinational survey of healthcare professionals	10-B
LB-21	Can Ficicioglu	RGX-121 gene therapy for the treatment of neuronopathic mucopolysaccharidosis type II (MPS II): Interim analysis of data from the first in human study	9-B
LB-28	Skyler Jackson	Health insurance literacy in ASMD	8-B
LB-29	Mijeong Kim	Tacrolimus administration in combination with dexamethasone reduces neutralizing antibody formation against AAV vector and increases transgene expression in cynomolgus macaques	7-B
LB-35	Hsiang-Yu Lin	Quantification of IDUA enzymatic activity combined with observation of phenotypic change in zebrafish embryos provide a preliminary assessment of mutated *IDUA* correlated with mucopolysaccharidosis type I	13-A
LB-39	Raquel Marques	Increasing Sanfilippo syndrome awareness through children's literature and music	14-A
LB-41	Naresh Kumar Meena	AAV-mediated systemic delivery of IGF2-tagged acid α-glucosidase corrects neuromuscular pathology in murine Pompe disease	15-A
LB-42	Gabriel Miltenberger Miltenyi	Phospholipid changes and the effect of medication in Gaucher disease and Parkinson's disease	16-A
LB-43	Alexandra Monceau	Single nuclei RNA sequencing of skeletal muscle of late-onset Pompe disease reveals new molecular signatures involved in the disease progression	17-A
LB-45	Christina Ohnsman	RGX-381: First-in-human clinical trial of an investigational AAV9 gene therapy encoding TPP1 for the treatment of ocular manifestations of CLN2 Batten disease	18-A
LB-46	Huseyin Onay	Application of deep intronic sequencing and RNA sequencing for an unsolved Pompe case	18-B
LB-51	Maria Picone	Understanding pain and its impact on mental health in Fabry disease using real-world evidence from social media	17-B
LB-52	Megan Pope	Assessment of gene therapy treatment on the Pompe disease canine model	16-B
LB-55	Carly Rasmussen	Treatment-naive and post-treatment lyso-GL-1 levels in a cohort of pediatric patients with Gaucher disease	15-B
LB-57	Xiomara Rosales	Evidence from a study of CLN5$^{-/-}$ sheep supporting dose escalation in an ongoing clinical trial of NGN-101 in pediatric patients with CLN5 Batten disease	14-B
LB-60	Ida Vanessa Schwartz	New insights into lysosomal disease variants in cases of atypical parkinsonism	13-B
LB-61	Annalisa Sechi	Plasma homocystine is a potential biomarker of disease severity in Fabry disease	19-A
LB-62	Alyaa Shmara	Assessing mechanism for reduced bone mineral density in Fabry disease	20-A

LB-63	Mahsa Taherzadeh	Severe neuronal demyelination in Sanfilippo disease	21-A
LB-64	Lucas Tricoli	Improved gene therapy for metachromatic leukodystrophy	22-A
LB-66	Arjan van der Flier	In vivo brain glycogen measurements in the Pompe disease mouse using GlycoCEST/ nOe MRI demonstrating CNS efficacy of a novel anti-mouse TfR-targeted GAA enzyme replacement therapy	23-A
LB-70	Chester B. Whitley	The PS Gene-editing (PSG) System for treatment of lysosomal diseases	24-A
LB-71	Matheus Wilke	Glucosylsphingosine (lyso-Gb1) correlates with IgG levels in a cohort of Gaucher disease patients	24-B
LB-74	Ryan Colburn	1:18,702. A robust projection of Pompe disease prevalence at birth based on over 11.7 million newborns screened	23-B
LB-56	Mark Roberts	Late-onset Pompe disease (LOPD) patients treated with avalglucosidase alfa show favorable results compared to cipaglucosidase alfa plus miglustat: Indirect treatment comparison	22-B

Clinical Applications – Poster Session V – Friday, February 24, 3:00 PM - 4:00 PM

#	Author	Title	Kiosk
1	Magy Abdelwahab	Kyphoscoliosis in Egyptian patients with chronic neuronopathic Gaucher disease over 16 years: Part of disease phenotype or a marker of disease severity	1-A
4	Carlos Acosta Rodríguez Bueno	The impact of vitamin D in patients with lysosomal diseases on ERT	2-A
11	Huma Ahmed	Respiratory impairments in patients suffering from Fabry disease	3-A
14	Nadia Ali	Pregnancy with Morquio syndrome type A: What are patients' perspectives and has enzyme replacement therapy (ERT) changed them?	4-A
21	Gabriela Araújo	Who cares for the caregivers? Lysosomal disease patients' caregivers health status in Brazil	5-A
23	Charlotte Aries	Addressing neurodegeneration in sialidosis type II with hematopoietic stem cell transplantation: A three year interim report	6-A
43	Michal Becker-Cohen	Prodromal Parkinsonian features in carriers of GBA1 variant(s) compared to controls	6-B
44	John Bernat	Long-term safety and efficacy of pegunigalsidase alfa administered every 4 weeks in patients with Fabry disease: Two-year interim results from the ongoing phase 3 BRIGHT51 open-label extension study	5-B
57	Barbara K. Burton	Safety profile of idursulfase administered at home in patients with mucopolysaccharidosis type II (MPS II) enrolled in the Hunter Outcome Survey	4-B
59	Barry J. Byrne	Long-term follow-up of cipaglucosidase alfa/miglustat in ambulatory patients with Pompe disease: An open-label phase I/II study (ATB200-02)	3-B
61	Valeria Calbi	Lentiviral haematopoietic stem cell gene therapy for metachromatic leukodystrophy: Results in 5 patients treated under nominal compassionate use	2-B
68	Hui-An Chen	Immune modulation therapy for enzyme replacement therapy-treated Pompe disease patients	1-B
74	Harriet Clayton	Barriers to participation in clinical trials	7-A
75	Tanya Collin-Histed	A collaborative approach to address the unmet needs of patients with neuronopathic Gaucher disease type 2 and type 3, the creation of GARDIAN, a patient-led, patient-owned global registry	8-A
80	Timothy Cox	Fine-motor functioning and its relevance to activities of daily living in adults with late-onset GM2 gangliosidosis: Analysis of baseline data of AMETHIST trial	9-A

93	Jordi Diaz-Manera	Avalglucosidase alfa improves motor and respiratory function in late-onset Pompe disease: A COMET win-ratio analysis	10-A
95	Jordi Diaz Manera	AT845 gene replacement therapy for late onset Pompe disease: An update on safety and preliminary efficacy data from FORTIS, a phase I/II open-label clinical study	11-A
109	Chae Sung Lee	Safety and tolerability of agalsidase beta infusions shorter than 90 minutes in patients with Fabry disease: Evidence from a Japanese post-marketing study	12-A
111	Larissa Faqueti	A Brazilian patient with late infantile metachromatic leukodystrophy treated with lentiviral hematopoietic stem-cell gene therapy: A report from prenatal diagnosis to early treatment	12-B
113	Ulla Feldt-Rasmussen	Real world evidence study of Danish patients with Fabry disease: A 20-year longitudinal retrospective analysis of prospectively collected data	11-B
117	Rachel Fisher	Seriously different versions of mucopolysaccharidosis type VII	10-B
118	Kevin Flanigan	Interim results of transpher A, a multicenter, single-dose clinical trial of UX111 gene therapy for Sanfilippo syndrome type A (mucopolysaccharidosis IIIA)	9-B
121	Luise Förster	Schindler disease type III: Clinical presentation of a patient carrying the homozygous missense variant c.973G>A (p.E325K) in the NAGA gene	8-B
125	Francesca Fumagalli	Long-term clinical outcomes of atidarsagene autotemcel (autologous hematopoietic stem cell gene therapy [HSC-GT] for metachromatic leukodystrophy) with up to 11 years follow-up	7-B
130	Pilar Giraldo	ELIKIDS: Baseline characteristics from the eliglustat substrate reduction therapy trial in children with Gaucher disease type 1 or type 3	13-A
132	Roberto Giugliani	Long-term catch-up growth in children with acid sphingomyelinase deficiency treated with olipudase alfa enzyme replacement therapy in the ASCEND-Peds trial	14-A
134	Roberto Giugliani	Vestronidase alfa for the treatment of mucopolysaccharidosis type VII (MPS VII): Updated results from a novel, longitudinal, multicenter Disease Monitoring Program (DMP)	15-A
136	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 type VI: Study update	16-A
138	Ozlem Goker-Alpan	The survival as relates to the clinical spectrum, molecular variants and chaperone response in acute neuronopathic Gaucher disease	17-A
139	Adina Gomez Paredes	Fabry disease: Experience with two families in the state of Sinaloa, Mexico	18-A
146	Nathalie Guffon	Safety and compliance of home infusion of velmanase alfa for the treatment of alpha-mannosidosis in the clinical trial and real-world settings	18-B
149	Punita Gupta	Infantile Tay-Sachs disease: The need for improved prenatal screening	17-B
152	Luca Hagenah	The relationship between the expanded neuronal ceroid lipofuscinosis 2 (CLN2) clinical rating scale for motor function (CLN2 CRS-MX) and GAITRite® parameters	16-B
157	Paul Harmatz	Interim results of a phase 1/2 study of JR-171 (lepunafusp alfa), a novel brain-penetrant enzyme replacement therapy for MPS I	15-B
168	Justin Hopkin	Acid sphingomyelinase deficiency: Burden of disease and real world impact of enzyme replacement therapy on pediatric patients and caregivers	14-B
169	Robert J. Hopkin	STAAR, a phase I/II study of isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease: Dose escalation phase results	13-B
170	Dafne Horovitz	Transition to home infusion in Pompe disease in Brazil: Safety of the regimen during the COVID-19 pandemic and thereafter	19-A
171	Alison Howie	Development of a core outcome set for mucopolysaccharidoses (MPS) in children: Results from Delphi surveys and a consensus workshop	20-A
174	Erin Huggins	GSD II and two: A case series of Pompe disease with coexisting genetic diagnoses	21-A

176	Derralynn Hughes	FollowME Fabry Pathfinders registry: Renal effectiveness in a multi-national, multi-center cohort of patients on migalastat treatment for at least three years	22-A
182	Majdolen Istaiti	Ambroxol therapy for patients with Gaucher disease type 1, either poor responders to enzyme replacement therapy/substrate reduction therapy (ERT/SRT) or untreated	23-A
186	Jeanine R. Jarnes	Updated interim safety, biomarker, and efficacy data from Imagine-1: A phase 1/2 open-label, multicenter study to assess the safety, tolerability, and efficacy of a single dose, intra-cisterna magna (ICM) administration of PBGM01 in subjects with type I (early onset) and type IIA (late onset) infantile GM1 gangliosidosis (GM1)	24-A
189	Simon Jones	Atidarsagene autotemcel, a European post-regulatory approval model for delivery of autologous hematopoietic stem cell gene therapy products via a network of qualified treatment centers (QTCs)	24-B
190	Ana Jovanovic	Clinical characteristics of female patients enrolled in the FollowME Fabry Pathfinders registry	23-B
194	Peter Karachunski	Median nerve ultrasound: A screening tool in aiding diagnosis of carpal tunnel syndrome in MPS I patients	22-B
202	Priya S. Kishnani	Efficacy and safety of avalglucosidase alfa in participants with late-onset Pompe disease after 145 weeks of treatment during the COMET trial	21-B
206	Sandra Kyosen	Genetics-based infusion centers: Report of 16 years experience of one Brazilian center	20-B
225	Katie Linden	Implementation and clinical utility of stability and severity scoring systems in the Fabry disease cohort in Northern Ireland	19-B
226	Ramona Lindschau	Wolman disease presenting with secondary hemophagocytic lymphohistiocytosis	25-A
231	Caryn Lobel	FDrisk: Development of a validated risk assessment tool for Fabry disease utilizing electronic health record data	26-A
239	Ana Maria Martins	Changes in quality of life reflecting neurobehavioral improvements observed by caregivers/physicians of patients with neuronopathic mucopolysaccharidosis: An interview-based survey from Brazil following clinical trials with pabinafusp alfa	27-A
241	Eugen Mengel	Natural history of acid sphingomyelinase deficiency (ASMD) among European patients during childhood and adolescence: A retrospective observational study	28-A
242	Pramod K. Mistry	Changes in hematologic and visceral manifestations over time following imiglucerase initiation in Gaucher disease type 1 and type 3 pediatric patients in the ICGG Gaucher Registry	29-A
248	Joseph Muenzer	Interim analysis of key clinical outcomes from a phase 1/2 study of weekly intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II	30-A
249	Nicole Muschol	Real-world clinical profiles of patients with alpha-mannosidosis: Baseline evaluations from the SPARKLE registry	30-B
258	Antonio Ochoa-Ferraro	Cipaglucosidase alfa and miglustat under the Early Access to Medicines Scheme (EAMS): A single centre experience	29-B
288	Nishitha Pillai	Hematopoietic cell transplantation for mucopolysaccharidosis type I in the presence of decreased cardiac function	28-B
344	Karolina Stepien	Biochemical and clinical outcomes in childhood-onset and adult-onset cases with Gaucher disease type I	27-B
319	Norio Sakai	Qualitative analysis of patient characters, interviews on the burden of neuronopathic Gaucher disease in Japan	26-B
LB-26	Justin Hopkin	Patient versus caregiver burden in ASMD	25-B

Clinical Applications – Poster Session VI – Friday, February 24, 4:00 PM - 5:00 PM

#	Author	Title	Kiosk
94	Jordi Diaz-Manera	Nonparametric analysis of forced vital capacity in the COMET trial demonstrates superiority of avalglucosidase alfa vs alglucosidase alfa	1-A
110	Yoshikatsu Eto	Real-world data of enzyme replacement therapy with pabinafusp alfa for neuronopathic MPS-II: Updated clinical data from Japan	2-A
133	Roberto Giugliani	Long-term neurodevelopmental changes in subjects with MPS II following long-term treatment with pabinafusp alfa: An integrated analysis from pre- and post-approval clinical trials in Brazil and Japan	3-A
135	Roberto Giugliani	Long-term outcomes in patients with Fabry disease who were treated with agalsidase alfa for more than nineteen years: The Fabry Outcome Survey	4-A
147	Nathalie Guffon	Baseline characteristics of a real-world population with alpha-mannosidosis: Insights from the SPARKLE registry	5-A
203	Priya S. Kishnani	Mini-COMET study: Effects of 97 weeks of avalglucosidase alfa dosing on ptosis in participants with infantile-onset Pompe disease who were previously treated with alglucosidase alfa	6-A
207	Sandra Kyosen	Rapid disease progression after enzyme replacement therapy interruption in a patient with cholesteryl ester storage disorder	6-B
254	Aya Narita	Development and evaluation of Gaucher disease-specific patient reported outcome measurement in Japan	5-B
259	Antonio Ochoa-Ferraro	At home enzyme replacement therapy for patients with lysosomal disorders: A single-centre experience	4-B
268	Cara O'Neill	Development of consensus guidelines for the clinical care of individuals with Sanfilippo syndrome	3-B
269	Neslihan Onenli Mungan	Taliglucerase-alfa experience with 34 Gaucher disease patients from Turkey	2-B
274	Olaf Pachcinski	Dysregulation of invariant natural killer T cells levels in a large cohort of Fabry disease patients	1-B
277	Marc Patterson	Evaluation of the long term effect of arimoclomol in NPC	7-A
287	Paivi Pietila Effati	Long-term effectiveness of enzyme replacement therapy in Fabry disease with the p. Arg227Ter (R227*) mutation	8-A
289	Nishitha Pillai	Resolving variants of unknown significance and pseudodeficiency alleles	9-A
290	Guillem Pintos Morell	Early initiation of agalsidase alfa treatment improves clinical outcomes in male patients with classical Fabry disease: A Fabry Outcome Survey (FOS) analysis	10-A
291	Fabiano Poswar	Ventricular vascular coupling in mucopolysaccharidosis types IVA and VI: Data from the baseline assessment of a phase II clinical trial	11-A
311	David L. Rogers	Intravitreal enzyme replacement therapy to prevent retinal disease progression in children with neuronal ceroid lipofuscinosis type 2 (CLN2): Interim safety report	12-A
320	Annamaria Sapuppo	Avalglucosidase alfa in patients with infantile-onset Pompe disease (IOPD): The Italian real world experience	12-B
327	Benedikt Schoser	Characteristics of patients who have switched from alglucosidase alfa to avalglucosidase alfa: Baseline data from the Pompe Registry	11-B
334	Ali Sheikh	The biochemical and radiological evaluation of bone health in patients with mucopolysaccharidosis	10-B

340	Barbara Soberon	Consequences of late diagnosis and treatment in patients with Gaucher disease type 1: Experience of the Argentine group	9-B
345	Karolina Stepien	Long-term clinical and biochemical outcomes in adult mucopolysaccharidosis type I Hurler Scheie and Scheie patients	8-B
349	Angela Sun	Mucopolysaccharidosis type VII treated in the first year of life: A case report	7-B
352	Cynthia Tifft	AMETHIST: Baseline characteristics from a phase 3 trial of venglustat, a novel brain-penetrant glucosylceramide synthase inhibitor, in GM2 gangliosidoses and related diseases	13-A
357	Àngel Valls-Villalba	Bone disease in eliglustat treated Gaucher disease type 1 patients: A real-world experience in a tertiary referral hospital	14-A
358	Amber Van Baelen	Innovation in the diagnostic field: A new multiplex analysis for lysosomal diseases in dried blood spots	15-A
360	Todd Vanyo	Lyso-Gl3 levels in patients during transition from agalsidase beta to migalastat	16-A
365	Jesus Villarrubia	Value contribution of olipudase alfa therapy for the treatment of non-central nervous system manifestations of acid sphingomyelinase deficiency (ASMD) by multi-criteria decision analysis (MCDA)	17-A
368	Eric Wallace	First results of a head-to-head trial of pegunigalsidase alfa vs. agalsidase beta in Fabry disease: 2 year results of the phase 3 randomized, double-blind, BALANCE study	18-A
371	Raymond Y. Wang	RGX-111 gene therapy for the treatment of severe mucopolysaccharidosis type I (MPS I): Interim analysis of data from the first in human study	18-B
372	Melissa P. Wasserstein	Plasma lyso-sphingomyelin as a biomarker for acid sphingomyelinase deficiency: Correlations with baseline disease and response to olipudase alfa treatment in clinical trials	17-B
374	Michael West	Early therapy in Fabry disease: Outcomes from the Canadian Fabry Disease Initiative (CFDI) registry	16-B
LB-09	Allison Bannick	Outcomes of individuals with symptomatic Pompe disease transitioned from alglucosidase alfa to avalglucosidase alfa-ngpt	15-B
LB-12	Lisa Berry	Fabry disease coinciding with pathogenic variant autosomal dominant hereditary transthyretin amyloidosis - which is the red herring?	14-B
LB-27	Justin Hopkin	The impact of olipudase alfa on QoL in pediatric ASMD patients with neurologic disease	13-B
LB-32	Ashley Lahr	Clinical profiles of forty-five women diagnosed with Fabry disease	19-A
LB-40	Angela Martin Rios	Hypogonadism in a cohort of male patients with Pompe disease	20-A
LB-47	Neslihan Onenli Mungan	Patient or relative attitudes towards diagnosis and treatment processes in lysosomal diseases: Insights from a patient/relative survey in Turkey	21-A
LB-54	James Radke	Using virtual education programs to change clinical practice in lysosomal diseases	22-A
257	Sofia Nordin	Disease burden, treatment patterns and healthcare resource utilization associated with Pompe disease in Sweden: A real-world evidence study	23-A
LB-58	Ashwin Roy	Retrospective assessment of adults with implantable loop recorders in Fabry disease	24-A
LB-59	Benedikt Schoser	Long-term efficacy and safety of cipaglucosidase alfa/miglustat in ambulatory patients with Pompe disease: A phase III open-label extension study (ATB200-07)	24-B
LB-67	Victor Vasquez	Early results in a patient with CLN2 undergoing early treatment with cerebroventricular enzyme replacement therapy	23-B
LB-69	Lena Marie Westermann	Brain volumetric analysis in CLN2 patients receiving standard of care ICV-ERT with cerliponase alfa	22-B

Contemporary Forum – Poster Session VII – Saturday, February 25, 3:00 PM - 4:00 PM

#	Author	Title	Kiosk
2	Walter Acosta	Lectin-mediated delivery to tissues and organs affected by the development of anti-drug antibodies in MPS I mouse model	1-A
7	Laura A. Adang	Prospective telemedicine natural history study of multiple sulfatase deficiency	2-A
8	Mathews Adera	The Guard1 clinical trial - A first in-human, phase 1/2 study evaluating AVR-RD-02, a hematopoietic stem cell (HSC) gene therapy for Gaucher disease: Preliminary safety, pharmacodynamic and clinical efficacy results from the subjects observed for up to 24 months post-infusion	3-A
13	Jose Alcantara Rodriguez	COMPASS, a double-blinded randomized phase 2/3 study of the efficacy and safety of intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II	4-A
17	Lilu Guo	Biomarkers profiling in patients with mucopolysaccharidosis and correlation with other primary biomarkers	5-A
18	Allyson Anding	*In vivo* models for the assessment of glycogen accumulation and depletion in various tissues	6-A
31	Ewald Auer	Motor deficits in homozygous 6^{neo} mice as model of Pompe disease	6-B
33	Olulade Ayodele	A retrospective chart review of supportive care in patients with mucopolysaccharidosis type II in the United States	5-B
34	Olulade Ayodele	A targeted literature review on the manifestations and clinical burden in patients with Fabry disease	4-B
40	Julie Batista	Progression of left ventricular mass (LVM) is associated with clinical events in Fabry disease: Analyses from Fabry Registry	3-B
41	Isabela Batsu	Two years of venglustat combined with imiglucerase shows continued positive effects on neurological features and brain connectivity in adults with Gaucher disease type 3	2-B
42	Peter Bauer	At a Glance: The largest Niemann-Pick type C1 cohort with 602 patients diagnosed over 15 years	1-B
48	Akhil Bhalla	DNL310 normalizes primary storage substrates and biomarkers of lysosomal dysfunction in neuronopathic MPS II: 2-year interim analysis of a phase 1/2 study	7-A
51	Natalia Boukharov	Characterization of the G3Stg/*Gla* KO Fabry disease mouse model pathology to improve preclinical to clinical translation	8-A
54	Jillian Brown	Development and validation of enzyme replacement therapy for GM1 gangliosidosis	9-A
63	Jeff Castelli	Indirect treatment comparison of three enzyme replacement treatments for late-onset Pompe disease: A network meta-analysis with patient-level and aggregate data	10-A
66	Shun-Chiao Chang	Birth prevalence of metachromatic leukodystrophy: A systematic literature review	11-A
73	Yoonjin Cho	Quantifying and modelling disease progression trajectory for natural history of MPS II	12-A
76	Nils Confer	A novel imaging technology to improve subcutaneous nodule quantification in Farber disease	12-B
77	Souvik Modi	Lysosomal dysfunction in Alzheimer's disease and Parkinson disease	11-B
81	Bruce Crawford	Gender heterogeneity in the diagnosis and treatment journey for patients with Fabry disease: A Japanese database analysis	10-B
83	Christine í Dali	Association between NPC severity score domains and corresponding items of the performance-based Scale for the Assessment and Rating of Ataxia (SARA)	9-B
85	Sean Daugherty	Early development of a locus specific database for *GUSB* , the gene associated with mucopolysaccharidosis type VII: Hints of a higher predicted prevalence	8-B

96	Michael DiGruccio	Highly phosphorylated β-glucocerebrosidase (M011) that targets central nervous system neurons as a potential treatment for neuronopathic Gaucher disease type 2 and 3	7-B
99	Hung Do	Highly phosphorylated β-glucocerebrosidase (M011) has much broader tissue targeting and superior substrate reduction with potential for alternative dosing strategies for the treatment of Gaucher disease type I	13-A
108	Maria L. Escolar	First-in-human phase 1/2 trial of intravenous FBX-101 following hematopoietic stem cell transplantation increases GALC activity, supports brain development, and improves motor function in patients with infantile Krabbe disease: RESKUE clinical trial	14-A
112	Naima Fdil	Isolated lactosylceramide storage: Is it the consequence of a specific protein deficiency?	15-A
116	Taylor Fields	Collaboration between patient advocacy and industry to create a master protocol to investigate the novel therapy acetyl-L-leucine for three ultra-rare neurodegenerative diseases: Niemann-Pick type C, the GM2 gangliosides and ataxia-telangiectasia	16-A
127	Michael Gelb	A glimpse into the feasibility of next generation sequencing for newborn screening of lysosomal and other diseases with second-tier biochemical assays as part of the screening process	17-A
129	Jacinthe Gingras	Gene therapy for metachromatic leukodystrophy: Lead candidate optimization	18-A
142	Russell Gotschall	M021: rhGAA with optimal glycosylation profile containing very high levels of bis-phosphorylated N-glycans clears accumulated glycogen and rapidly normalizes muscle strength in treated Pompe disease mice	18-B
150	Beatriz Guzman	A structurally targeted allosteric regulator of GCase restores enzyme activity, reduces microgliosis, and improves fine locomotor skills in the CBE model of neuronopathic Gaucher disease	17-B
151	Nagy Habib	Drugging transcription factors with small activating RNAs: A novel approach for enhancing bone marrow therapy for monogenic rare diseases	16-B
154	Kohtaro Hamauchi	International online survey of fucosidosis: Key symptoms and the family experience	15-B
155	Miloud Hammoud	The role of combination between TLC and UHPL/MS-MS in the diagnosis and prognosis of metachromatic leukodystrophy	14-B
158	Guido Hartmann	Cellular disease models of Gaucher and Niemann Pick type C diseases	13-B
160	Andrew Hedman	Novel dual promoter AAV gene therapy platform ensures production of therapeutic soluble lysosomal enzymes with high M6P content to enable broad cellular uptake and cross correction *in vivo*	19-A
161	Walter Heine	Venglustat, a novel brain-penetrant glucosylceramide synthase inhibitor, for Gaucher disease type 3: Phase 3 LEAP2MONO trial design	20-A
166	Stephan Hold	Combined assay methodology for the analysis of enzyme activities and biomarker concentrations for Fabry, Gaucher, Krabbe, Niemann Pick types A/B, and Pompe disease	21-A
173	Yinyin Huang	Using single nuclear RNAseq to assess impact of AAV-ARSA gene therapy on oligodendrocyte populations	22-A
179	Atsushi Imakiire	Life-span extension in Krabbe disease mice by treatment with a transferrin receptor-targeted galactocerebrosidase	23-A
180	Asuka Inoue	Nonclinical pharmacodynamics, pharmacokinetics and safety profiles of anti-human transferrin receptor antibody-fused N-sulfoglucosamine sulfohydrolase for mucopolysaccharidosis type IIIA	24-A
181	Rizwana Islam	Preventing Fabry disease progression in a symptomatic mouse model with a recombinant adeno-associated virus (rAAV) based gene therapy	24-B
187	Rachel Johansson	Development of NtBuHA as a small molecule therapeutic for CLN1 Batten disease: An update on research efforts	23-B

191	Chanchala Kaddi	Mechanism-guided analysis of avalglucosidase alfa efficacy on key biomarkers and clinical endpoints in late-onset Pompe disease patients	22-B
193	Ilkka Kantola	Start age of enzyme replacement therapy (ERT) did not effect ECG parameters in Fabry disease patients (FD) treated by ERT for 5 years	21-B
199	Ed Kim	Systematic literature review on the burden of illness, health-related quality of life, and treatment options in pediatric and adult patients with alpha mannosidosis	20-B
201	Kwi Hye Kim	*In vitro* pharmacology study using retina organoids and retina-on-a-chip of CLN2 patient-derived induced pluripotent stem cells	19-B
205	Monica Kumar	Safety and clinical outcomes in children with acid sphingomyelinase deficiency were not impacted by a change in the olipudase alfa drug manufacturing process: ASCEND-Peds 1-year subanalysis	25-A
215	Randolph Leiser	Quantitative systems pharmacology (QSP) analysis predicts that olipudase alfa treatment of patients with extreme acid sphingomyelinase deficiencies can result in meaningful debulking of visceral tissue sphingomyelin	26-A
221	Daniel Lewi	Empowering the GM1 and GM2 community with a co-created education resource about a natural history study	27-A
223	Jolanda Liefhebber	Efficacy of AAV5-GLA gene therapy in manifest Fabry disease mice	28-A
227	Lin Liu	Novel AAV gene therapy produces beta-glucocerebrosidase with high levels of M6P to enable cellular uptake and cross-correction in the CNS as a potential treatment for type 2/3 Gaucher disease	29-A
236	Alasdair MacCulloch	Quality of life with late-onset Pompe disease: Qualitative interviews and general public utility estimation	30-A
243	Meera E. Modi	Building a better translational model of neuropathic Gaucher disease	30-B
255	Sarah Neuhaus	Phase 1/2 study update of an AAV9-based gene therapy for Gaucher disease type 2 (PROVIDE trial)	29-B
260	Christina Ohnsman	Loss of visual function associated with photoreceptor degeneration in CLN2 disease	28-B
265	Petra Oliva	The ratio of Niemann-Pick disease type A/B to cases of Gaucher disease varies by country	27-B
308	Camille Rochmann	Describing the natural history and burden of illness in Gaucher disease type 3 patients using a cluster from Optum's de-identified market clarity data (2007-2020)	26-B
LB-10	Kenneth Berger	Switching therapy from alglucosidase alfa to avalglucosidase alfa in patients with late onset Pompe disease (LOPD): Longitudinal assessment of respiratory function from the COMET trial	25-B

Contemporary Forum – Poster Session VIII – Saturday, February 25, 4:00 PM - 5:00 PM

#	Author	Title	Kiosk
3	Walter Acosta	Correction of bone pathology in MPS I mouse model using lectin-mediated delivery	1-A
67	Shun-Chiao Chang	The natural history and burden of illness of metachromatic leukodystrophy: A systematic literature review	2-A
200	Ed Kim	Systematic literature review on the epidemiology, diagnosis, and natural history of alpha mannosidosis	3-A
244	Meera E. Modi	Disease pathology in the iduronate-2-sulfatase KO mouse: Opportunities for novel endpoints for second generation enzyme replacement strategies	4-A
247	Hideto Morimoto	Intravenous treatment with pabinafusp alfa dose-dependently prevents neurological impairment and bone deformities in a mouse model of mucopolysaccharidosis type II	5-A

256	Sarah Neuhaus	Design and outcome measures for a phase 1/2 dose-finding study to evaluate systemic administration of an AAV9-based gene therapy for peripheral manifestations of Gaucher disease	6-A
261	Christina Ohnsman	Characterization of retinal degeneration phenotype in classic CLN2 disease using OCT biomarkers and an *in vitro* retinal model	5-B
266	Petra Oliva	A MLD newborn screening pilot-study for metachromatic leukodystrophy in Germany: Results of the first 12 months	4-B
270	Midori Ono	Assessing the impact on caregivers burden for patients with Gaucher disease in Japan	3-B
275	Francis Pang	The cost-effectiveness of OTL-200 for the treatment of metachromatic leukodystrophy (MLD) in the US	2-B
276	Shivakumar D. Pattada	RTB-lectin facilitates the distribution of enzymes across the blood-brain-barrier and correction in the MPS IIIA mouse model	1-B
283	Richard Pfeifer	Using IVIM/SAGA as screening tools during lentiviral vector lead selection for detection of clinically translatable insertional transformational risk	7-A
285	Dawn Phillips	Daily living skills on the Vineland Adaptive Behavioral Scale version 2 (VABS-II) in neuronopathic mucopolysaccharidosis type II (MPS II)	8-A
294	Maria Praggastis	Delivering therapeutic lysosomal enzyme to the CNS: A rapid, large-scale *in vivo* screen identifies optimal characteristics of BBB-crossing TFRC antibodies	9-A
297	Ana C. Puhl	Developing treatments for rare diseases on a shoestring: The Batten disease (CLN1) enzyme replacement therapy experience	10-A
298	Aurora Pujol	Towards a substrate reduction therapy for metachromatic leukodystrophy	11-A
299	Ruth Pulikottil-Jacob	Survival of patients with acid sphingomyelinase deficiency (ASMD) in the United States (US): A retrospective real-world study	12-A
300	Harpreet Ram	The Global Alliance for Rare Disorders Access (GARDaccess) strategic initiative for persons living with rare disease in low and middle income countries	12-B
301	Shyam Ramachandran	AAV-ARSA-mediated gene replacement for the treatment of metachromatic leukodystrophy	11-B
302	Charu Reddy	An engineered β-galactosidase with improved stability and cross-correction for the potential treatment of GM1 Gangliosidosis via AAV gene therapy	10-B
303	Laith Refaei	Gaucher disease type 3 patient journey: Results from interviews with neuronopathic Gaucher disease patients highlight the challenges of diagnosis and care	9-B
304	Pascal Reisewitz	Real-world impact of enzyme replacement therapy on endurance in patients with MPS IVA	8-B
307	Susan Richards	Immune response to agalsidase-beta has limited impact on therapeutic effect in Fabry disease patients: Lessons from 20+ years of clinical research	7-B
309	Camille Rochmann	A summary of neurological and neurocognitive manifestations for Gaucher disease type 3 from infancy to adulthood	13-A
315	Rosa Romano	Development of a B cell protein factory to produce sustained therapeutic levels of alpha-galactosidase A for the treatment of Fabry disease	14-A
321	Terrence Satterfield	Quantification of peripheral blood mononuclear cell (PBMC) glycogen as a novel biomarker for therapeutic intervention in Pompe disease	15-A
322	Adam Scheller	DNL310-treated study participants with MPS II show improvements in toileting abilities	16-A
323	Raphael Schiffmann	Cardiac effects of 4D-310 in adults with Fabry disease in a phase 1/2 clinical trial: Functional, quality of life, and imaging endpoints in patients with 12 months of follow up	17-A
324	Irene Schilcher	Myopathic and neuronal pathologies of the 6^{neo} mouse model of Pompe disease	18-A
326	Christopher Schmitt	*In vitro* modeling of GM1 gangliosidosis using iPSC-derived cellular and organoid CNS models	18-B

335	Seung-Yub Shin	Discovery of YH35995A, a novel highly-potent BBB-penetrating GCS inhibitor for the treatment of Gaucher disease	17-B
336	Charles Shyng	Investigational liver gene transfer of alpha galactosidase for the treatment of Fabry disease	16-B
348	Theodore Sullivan	Transduction and AGA expression by 4D-310, an AAV genetic medicine, in the hearts and kidneys of nonhuman primates and *Gla* knockout mice: Efficient delivery to cardiomyocytes and glomeruli, including podocytes	15-B
351	Mengdi Tao	Pediatric and adult acid sphingomyelinase deficiency (ASMD) patients exhibit mechanistic similarity in their disease and response to olipudase alfa	14-B
354	Michael A. Tortorici	Centyrin-targeted glycogen synthase-1 siRNA conjugates: A novel therapeutic modality for the treatment of Pompe disease	13-B
355	Julie C. Ullman	Small molecule inhibition of glycogen synthase 1 restores autophagolysosomal and metabolic pathway dysfunction in a mouse model of Pompe disease	19-A
363	Biliana Veleva-Rotse	High prevalence of cardiac findings in individuals with Fabry disease identified via no-charge sponsored testing programs	20-A
367	Eileen Wall	Usability study of a new bottle for cystaran eye drops in cystinosis subjects currently on eyedrop therapy for corneal cystine crystal accumulation	21-A
380	Tianhong Yang	Intracellular stability and efficacy of β-galactosidase fusions as a GM1 gangliosidosis ERT are significantly influenced by mouse versus human multi-enzyme complex interactions	22-A
381	Eiji Yoden	A fusion protein of anti-human transferrin receptor antibody and alfa-L-fucosidase 1 is a prospective candidate for the treatment of the symptoms in CNS and visceral tissues of fucosidosis	23-A
382	Bailin Zhang	CD63 as the potential universal biomarker for lysosomal diseases	24-A
LB-04	Ruslan Al-Ali	Transcriptomics on dried blood spots facilitate better understanding of Gaucher disease	24-B
LB-11	Kenneth Berger	Defining clinically meaningful thresholds (CMT) for forced vital capacity (FVC) and six-minute walk test (6MWT) in patients with late-onset Pompe disease (LOPD)	23-B
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