Authors who accepted a poster or ePoster presentation are assigned to present their abstract during a live in-person session or virtually during one of four sessions, based on the final abstract category for each abstract. All posters will be available to all attendees beginning at 3:00 PM PST on Monday, February 7, 2022, on the WORLD*Symposium* virtual platform and will remain open throughout WORLD*Symposium* 2022. Live Q&A will only occur during the assigned times as listed below.

Please note: "in-person" indicates the presenter will be available for questions at the live, in-person poster session during WORLD*Symposium* 2022 and "virtual" indicates the presenter will be available on the WORLD*Symposium* virtual site to answer questions via text-based chat.

All "in-person" posters will be in the Exhibit Hall in the Seaport Ballroom and "virtual" ePosters will be on the virtual platform:

Basic Science Abstracts will be presented on **Monday, February 7 from 3:00-5:00 PM PST**
Translational Research Abstracts will be presented on **Tuesday, February 8 from 3:00-5:00 PM PST**
Clinical Applications Abstracts will be presented on **Wednesday, February 9 from 3:00-5:00 PM PST**
Contemporary Forum Abstracts will be presented on **Thursday, February 10 from 3:00-5:00 PM PST**

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.

Monday, February 7 – Basic Science Poster Presentations

8	Hernan Amartino	How complete is our clinical assessment of patients with mucopolysaccharidosis type II in real life? A question from the Hunter Outcome Survey (HOS)	Virtual
12	Isidro Arevalo-Vargas	Evaluating the performance of 16 *in silico* predictors on 22 lysosomal diseases	Virtual
15	Frederick Ashby	Central nervous system distribution of stem cells in Sanfilippo syndrome type B mice by injection method	In-person
16	Christiane Auray-Blais	Mass spectrometry analysis of the vitreous fluid from a Gaucher disease type 3 patient	Virtual
19	Sukirhini Balendran	Rapid identification of IOPD and early-onset Pompe disease by biochemical enzymatic testing followed by genetic confirmation	In-person
20	Dishary Banerjee	Biomimetic 3D tissue printing to create an *in vitro* bone model for Gaucher disease	Virtual
31	Elizabeth Braunlin	Aortic dilation in murine mucopolysaccharidosis type I: A tale of two strains	Virtual
40	Martino Calamai	GM1 content evaluation in fibroblasts for pre-diagnosis and pharmacological management of patients affected by GM1 gangliosidosis	In-person
43	David Cassiman	Overlapping and divergent hepatic and lipoprotein phenotypes in untreated adults with acid sphingomyelinase deficiency versus untreated adults with Gaucher disease from two pivotal clinical trials	Virtual
45	Chase Chen	Investigation into the pathophysiology of *GBA1*-associated Parkinson disease using organelle-specific proteomics	In-person
53	Chloe Christensen	Generation of an induced pluripotent stem cell line from a patient with free sialic acid storage disorder	In-person
54	Heather Church	Graft rejection and spontaneous recovery in mucopolysaccharidosis type I post-HSCT	Virtual
59	Emily Curtin	A comparison of clinical features in two patients with mucopolysaccharidosis type VI treated with ERT versus HSCT	Virtual
60	Vânia D'Almeida	Influence of lysosomal diseases on reproductive parameters of animal models	In-person
68	Jordi Diaz-Manera	BNIP3 is involved in muscle fiber atrophy in late-onset Pompe disease patients	Virtual
70	Patricia Dickson	Co-expression of S1S3 phosphotransferase in production cell line improves mannose 6-phosphorylation and cellular uptake of alpha-N-acetylglucosaminidase (Sanfilippo syndrome type B)	In-person
74	Franklin Ducatez	Predictive biological patterns in Gaucher disease revealed by integrative omics-based machine learning analyses	Virtual
76	Mariola Edelmann	GALC-containing EVs as a tool to deliver ERT agents and treat neuronopathic processes in Krabbe disease	Virtual
77	Grigoris Effraimidis	X chromosome inactivation, α-galactosidase A activity and lyso-Gb_3 in Danish heterozygous females with Fabry disease	In-person

80	Takumi Era	Analysis of GM1 gangliosidosis iPS cells provides new phenotype of neural dysfunction and drug candidates	Virtual
83	Francois Eyskens	Serum bone alkaline phosphatase: A biomarker for Gaucher disease	Virtual
85	Joaquin Frabasil	Correlation analysis between lysosomal enzyme activities and the different types of leukocytes in dried blood spots	Virtual
86	Joaquin Frabasil	A new algorithm for Gaucher disease diagnosis in dried blood spots	Virtual
90	Yasuyuki Fukuhara	cDNA analysis disclosed presumable discordance of genotype-phenotype correlation in a patient with attenuated MPS II having 76 base deletions in the gene for iduronate-2-sulfatase	Virtual
95	Pilar Giraldo	A novel enzymatic diagnosis method of lysosomal diseases	In-person
100	Adenrele Gleason	Can the profile of extracellular vesicles (EVs) reflect the disease states of patients with Gaucher disease and *GBA1* carriers with or without Parkinson disease?	Virtual
115	Sangwoo Han	Storage of GM2 ganglioside and altered gene expression in infantile Tay-Sachs disease during fetal development has implications for therapeutic timing and efficacy	Virtual
116	Jerry Harb	CRISPR-mediated generation and characterization of the *GAA* homozygous c.1935c>a (p.d645e) Pompe disease knock-in mouse model	In-person
145	Tiffany Jong	Using genome-wide pooled CRISPRi screen to identify genetic modifiers of Gaucher disease and Parkinson disease	In-person
148	Asaka Katabuchi	Small molecule characterization as potential therapies for Krabbe disease	Virtual
155	Aditi Korlimarla	Seizures in infantile Pompe disease: Expanding our understanding of the clinical spectrum	In-person
157	Francyne Kubaski	Profile of disease-specific oligosaccharides in the cerebrospinal fluid of patients with mucopolysaccharidoses	In-person
162	Dawn Laney	Possible increased incidence of pathogenic *GALC* deletions in exons 11-17 causing infantile onset Krabbe disease in individuals from Ahmadabad in the Indian state of Gujarat	In-person
167	Steven Le	Spinal cord pathology in murine Sanfilippo syndrome type B	In-person
168	Andrés Leal	Efficient phenotype recovery of MPS IVA fibroblasts after CRISPR/nCas9-mediated genome editing	In-person
169	Andrés Leal	Genome editing on GM2 gangliosidoses fibroblasts using CRISPR/nCas9	In-person
173	Malte Lenders	Isolation and characterization of a polyclonal human anti-drug antibody as a reference in Fabry disease	In-person
176	Yi Lin	Modeling neuronopathic Gaucher disease with human patient-specific midbrain organoids	Virtual
179	Laura López de Frutos	Secondary dysfunction of lysosomal enzymes in Gaucher disease	In-person
181	Juan Losada	*In silico* identification and characterization of potential orthosteric and allosteric pharmacological chaperones of the NAGLU enzyme and evaluation of their chaperone effect *in vitro*	Virtual
188	Maissaa Mahir	Brain MRI findings of a glutaric aciduria type I: A case report and review of the literature	Virtual
192	Ryuichi Mashima	Generation of therapeutic iduronate-2-sulfatase enzyme using a novel RNA virus vector	Virtual
197	Iskren Menkovic	Plasma biomarker profile quantification by tandem mass spectrometry for early detection of Gaucher disease	Virtual
198	Emma Michl	Extensive diagnostic odyssey for a patient with Gaucher disease	In-person
199	Emma Michl	Early presentation with late-onset Pompe disease genotype due to a genetic modifier: Lessons from newborn screening	In-person
200	Emma Michl	Making the case for global carrier screening for Tay-Sachs disease	In-person
204	Travis Moore	iPSC derived neurons of mucopolysaccharidosis III patients show pronounced synaptic defects	In-person
208	Joseph Muenzer	Fifteen years of the Hunter Outcome Survey (HOS): Real-world insights into the patient population living with mucopolysaccharidosis type II (MPS II)	In-person
209	Sireesha Murala	Diffusion tensor imaging (DTI) findings in children with Pompe disease: Insights into white matter hyperintensities from a longitudinal study	In-person
211	Behzad Najafian	Globotriaosylceramide (GL3) accumulation in Fabry podocytes in female patients is progressive with age and associated with podocyte loss and proteinuria	In-person
225	Saida Ortolano	Enzyme replacement therapy tend to stabilize inflammatory and cardiovascular biomarkers in plasma samples from Fabry disease	In-person
226	Xuefang Pan	Neurodegenerative role of lysosomal cathepsin B in MPS IIIC	In-person
232	Gani Perez	Behavioral and whole transcriptome analyses of a gba-haploinsufficient Parkinson murine model	In-person
243	Gisele Pino	The synergy of multiplex testing to screen for lysosomal disorders (LD)	Virtual

250	Felippe Previdi	Neuroradiological evaluation of Fabry disease patients in follow-up in a reference center before ERT	Virtual
253	Alexander Pushkov	Measurement of tripeptidyl peptidase 1 activity as a first level test and as a confirmatory test for the diagnosis of neuronal ceroid lipofuscinosis type 2	Virtual
260	Allisandra Rha	Prime editing corrects the c.1826dupA mutation in infantile-onset Pompe disease	In-person
261	Mercedes Roca-Espiau	Incorporation of machine learning technologies into the assessment of bone involvement in Gaucher disease	In-person
264	Christy Rohani-Montez	Fabry disease: Effectiveness of interactive case-based online education in improving knowledge and competence in diagnosis	Virtual
268	Marya Sabir	A novel experimental mouse model to investigate a free sialic acid storage disorder (Salla disease)	In-person
274	Markus Schwarz	Alpha-mannosidosis is underdiagnosed lysosomal disease	In-person
275	Markus Schwarz	High-risk population screening by differential diagnosis for mucopolysaccharidoses (MPSs)	In-person
278	JooHyun Seo	Natural history of cognitive development in neuronopathic mucopolysaccharidosis type II (Hunter syndrome): Contribution of genotype to cognitive developmental course	Virtual
279	Irene Serrano Gonzalo	Study of neutrophil extracellular traps (NETs) in the development of thrombotic complications in Fabry disease patients	In-person
282	Yuki Shiro	Novel insight into the compound heterozygosity-driven CLN6 disease pathomechanism	Virtual
283	Aishwarya Siddharth	Diagnostic odysseys for treatable lysosomal diseases and the role of whole exome sequencing: Lessons learned	In-person
284	Aishwarya Siddharth	Identifying barriers for access to care among immigrants living with lysosomal disorders	In-person
285	David Smerkous	Automated estimation of foot process width using deep learning in kidney biopsies from patients with Fabry disease	In-person
287	Miles Smith	Comparative effectiveness of intravenous and intrathecal AAV9.CB7.hIDS (RGX-121) in a murine model of mucopolysaccharidosis type II	In-person
298	Mahsa Taherzadeh	Expression of misfolded HGSNAT protein aggravates neurological phenotype in mucopolysaccharidosis type IIIC	Virtual
300	Marcia Terluk	Neurofilament light chain: A potential marker of neurological disease in Gaucher disease	In-person
306	Heidy Triana Rojas	*In vitro* evaluation of recombinant enzyme N-acetyl-alpha-glucosaminidase obtained from *Komagataella phaffii* GS115	In-person
310	Bethann Valentine	Evaluation of neurofilament light chain as a biomarker for mucopolysaccharidosis type IIIB	Virtual
313	Patricia Varela-Calais	Tenascin C down regulation in a neuron model of Fabry disease	Virtual
314	Viviana Vargas-López	Epigenetic changes in fibroblast from patients with mucopolysaccharidoses	In-person
316	Dana Velasquez Rivas	Increased levels of Lyso-Gb1 in dried blood spots in non-Gaucher patients	Virtual
317	Jesús Villarrubia	Adult acid sphingomyelinase deficiency (Niemann-Pick disease type B): A difficult pathway to a diagnosis in 4 novel cases	In-person
330	Dongshan Yang	A rabbit model of cystinosis has deposition of cystine crystals in the cornea	Virtual
LB-04	Sofia Annis	Transcriptomic profiling and characterization of treatment response in late-onset Pompe disease skeletal muscle	Virtual
LB-08	Kenneth Berger	Changes in forced vital capacity over ≤13 years among late-onset Pompe disease patients treated with alglucosidase alfa: New modeling of Pompe Registry data	Virtual
LB-12	Chloe Christensen	Base editing of *GAA* rescues Pompe disease phenotype in patient-derived human dermal fibroblasts	Virtual
LB-19	Chrissy Fortune	The impact of COVID-19 on Fabry disease patients receiving enzyme replacement therapy (ERT)	Virtual
LB-24	Emdadul Haque	Development of an assay to measure PPT1 activity in human cerebrospinal fluid (CSF) and serum for the evaluation of preliminary effectiveness of AAV9 gene therapy for CLN1 disease	Virtual
LB-26	Julia Hennermann	Mortality in patients with alpha-mannosidosis	Virtual
LB-33	Ayse Kilic	Pompe disease: Single center experience	Virtual
LB-34	Jin Young Kim	Preventive effect on the progressive renal disease by long-acting alpha-galactosidase A analog (HM15421) in Fabry disease mice model	Virtual
LB-40	Laura López de Frutos	Serum phospholipid profile changes in Iberic Gaucher disease and Parkinson disease patients	Virtual
LB-47	Carmine Mottolese	Spine fusion management and stability in patient with mucopolysaccharidoses	Virtual

LB-49	Behzad Najafian	Venglustat reduces globotriaosylceramide inclusions in skin arterial smooth muscle cells in treatment naive males with classic Fabry disease	Virtual
LB-55	Cho rong Park	Preventive effect on motor activity and bone deformity by long-acting arylsulfatase B analog (HM15450) in mucopolysaccharidosis type VI (MPS VI) mice model	Virtual
LB-57	Maria Picone	Unique patient insights captured from social media contribute to real-world evidence: Fabry disease	Virtual
LB-58	Tyler Pierson	Modeling CLN6 with IPSC-derived neurons and glia	Virtual
LB-64	Ida Schwartz	Lysosomal disease variants in cases of atypical parkinsonism	Virtual
LB-69	Matheus Wilke	Evolution of the frequency of pre motor symptoms of Parkinson disease in adult patients with Gaucher disease type 1: A cohort study	Virtual
LB-70	Jagdeep Walia	AZ-3102, a novel brain-penetrant small molecule, significantly improves survival of Sandhoff disease mice	Virtual
LB-74	Christine Yu	Timely diagnosis of mucopolysaccharidosis type III: Results from an innovative education program targeting US pediatricians	Virtual

Tuesday, February 8 – Translational Research Poster Presentations

3	Nicolas Abreu	Onset and evolution of symptoms in CLN8 disease	In-person
4	Nicolas Abreu	The 50-meter timed test as a simple, efficient and objective measure of gross motor function in CLN3 disease: A pilot study	In-person
7	Walla Al-Hertani	The Boston Children's Hospital (BCH) four year experience with the Massachusetts State Newborn Screening (NBS) pilot program for mucopolysaccharidosis type I (MPS I), Pompe disease, and X-linked adrenoleukodystrophy(X-ALD): Lessons Learned	In-person
9	Luise Ammer	Disease manifestations in mucopolysaccharidoses and their impact on anaesthesia-related complications: A retrospective analysis of 99 patients	Virtual
10	Luise Ammer	Neurocognitive development and adaptive behavior in mucolipidosis type II: A retrospective analysis of 11 patients	Virtual
13	Charlotte Aries	Combination of high-dose ambroxol and ERT in Gaucher disease type 2: A nearly age-appropriate neurocognitive and motor development after three years of treatment	Virtual
25	Lisa Berry	Newborn screening for lysosomal disorders: The Ohio experience	Virtual
27	Tierra Bobo	Facilitate by-stander effects by EV-mRNA cargo in AAV gene replacement therapy for treating MPS IIIC	In-person
29	Natalie Boychuk	It's a matter of opinion: An exploratory study of parent attitudes towards newborn screening for later-onset and untreatable disorders	Virtual
30	Natalie Boychuk	Parental depression and stress associated with newborn screening for complex disorders	Virtual
34	Liesl Broadbridge	A review of common data elements in RUSP submission packages	Virtual
56	Jonathan Cooper	Amelioration of enteric nervous system defects via gene therapy in CLN1 disease mice	In-person
61	Julia Dao	Wnt signaling pathway inhibitor, sclerostin, correlates with bone pain and bone marrow infiltration in patients with Gaucher disease	Virtual
64	Francisco del Castillo	NGS-based panel screening of suspected lysosomal disease cases identifies novel pathogenic variants underlying acid sphingomyelinase deficiency (ASMD), Krabbe disease, and lymphatic dysplasia with non-immune hydrops fetalis	In-person
65	Francisco del Castillo	Genetic screening of lysosomal disorders: An account of five years' experience with NGS-based resequencing panels	In-person
75	Halil Dündar	Synergistic action of the chemical chaperone 4-phenylbutyrate and the pharmacological chaperone migalastat on restoration of α-galactosidase activity of Fabry G258R mutation	Virtual
78	Stuart Ellison	Enhanced transduction and immunophenotyping demonstrates preclinical safety and efficacy of haematopoietic stem cell gene therapy for mucopolysaccharidosis II using an IDS.ApoEII brain targeted therapy	In-person
88	Haiyan Fu	Transient depletion of pre-existing antibodies for efficient AAV gene delivery	In-person
91	Jillian Gallagher	Sialidosis: From gene editing to gene therapy	In-person
92	Dominique Germain	Interpreting the pathogenicity of genetic variants in rare diseases: Lessons from Fabry disease	Virtual
104	Kimberly Goodspeed	A cross-sectional natural history study of aspartylglucosaminuria	Virtual

107	Christina Grant	Newborn screening for Pompe disease: The Washington, DC experience	In-person
110	Melissa Greco	Tractography and psychosine as biomarkers of neurodegeneration in babies with Krabbe disease	In-person
125	Claire Horgan	Ex-vivo autologous stem cell gene therapy for MPS II (Hunter syndrome)	Virtual
126	Erin Huggins	Early clinical phenotype of late-onset Pompe disease: Lessons learned from newborn screening	In-person
129	Derralynn Hughes	Estimation of health state utility values in Fabry disease using vignette construction and valuation	Virtual
132	Marjan Huizing	A concerted action to explore therapies for free sialic acid storage disease (FSASD)	In-person
133	Sarah Hurt	An adenoviral mediated gene therapy for mucopolysaccaridoses type I	In-person
137	Margarita Ivanova	Identification of circulated biomarkers in Fabry disease patients associated with hypertrophic cardiomyopathy	In-person
147	Neil Kasaci	The expression and secretion profile of TRAP5 isomers correlate with bone involvement in Gaucher disease	Virtual
150	Nicole Kelly	Screenplus: A model for collective funding of pilot newborn screening	Virtual
159	Francyne Kubaski	Pilot study update: Newborn screening for lysosomal disorders in Brazil	In-person
164	Dawn Laney	Initial symptom presentation in young pediatric patients with classic pathogenic variants in the *GLA* gene: Data from the Fabry MOPPet study	In-person
191	Craig Martin	Improving patient identification, inclusion and engagement in research for LDs and other rare CNS conditions (PIE4CNS)	In-person
201	John Mitchell	Farber disease clinical impact: Patient reported outcomes as a measure of disease burden	In-person
202	John Mitchell	Subcutaneous nodules as a clinical biomarker of Farber disease	In-person
203	Marc Moltó-Abad	Efficacy of targeted nanoliposomes in reducing globotriaosylceramide (Gb3) accumulation in mouse models of Fabry disease	Virtual
206	Tahseen Mozaffar	AT845 gene replacement therapy for late onset Pompe disease: Overview of clinical data from FORTIS, a phase 1/2 open-label clinical study	In-person
210	Ikuma Musha	Newborn screening for Fabry disease is useful for early diagnosis of the family members who are affected but are not yet diagnosed	Virtual
212	Hemanth Nelvagal	Efficacy of recombinant human PPT1 enzyme replacement therapy in mouse and sheep models of CLN1 disease	In-person
213	Igor Nestrasil	Quantitative brain MRI morphology in severe and attenuated forms of mucopolysaccharidosis type I	In-person
214	Miriam Nickel	Hamburg iNCL scale: A new tool for the quantitative description of disease progression in infantile CLN1 patients	In-person
219	Torayuki Okuyama	Current status of newborn screening for lysosomal diseases in Japan: Importance of novel therapies for central nervous system manifestation in MPS II, and importance of family screening of Fabry disease after newborn screening	Virtual
242	Nishitha Pillai	Newborn screening experience and outcome from a Minnesota Pompe disease consortium	In-person
249	Maximiliano Presa	Efficacy of a scAAV9/SUMF1 viral vector for the treatment of multiple sulfatase deficiency	In-person
251	Michael Przybilla	Prevention of murine GM1-gangliosidosis following heterotopic insertion of Glb1 using gene editing	In-person
255	Kayla Quirin	A study to identify individuals at risk to be affected by Pompe disease who had previously been given a non-specific or tentative diagnosis for their muscle weakness (Pompe PURSUE)	In-person
272	Angela Schulz	Natural history of CLN7 disease: Quantitative prospective assessment of disease characteristics and rate of progression	In-person
277	Varshaa Senthilkumar	Variation in quantitative glycosaminoglycan analysis results from different clinical laboratories	Virtual
295	Diego Suárez	Genome editing in mucopolysaccharidosis type IVA fibroblasts using CRISPR/Cas9	In-person
296	Dean Suhr	A proposal to efficiently improve diagnostic clarity, therapeutic and clinical referrals, disease and therapeutic understanding, and quality of life in the newborn screening ecosystem while reducing cost and overhead	In-person
297	Angela Sun	A rapid and non-invasive proteomic analysis using DBS and buccal swab for multiplexed second-tier screening of Pompe disease and Hurler syndrome	In-person
312	Todd Vanyo	Comparison of therapeutic potential of ERT to chaperone therapy in I270T related Fabry disease	In-person
319	Chelsey Walsh	Newborn screening for Krabbe disease in Illinois: A single center's experience	In-person

324	Amy White	Comparison of psychosine analysis in dried blood spots and red blood cells from children with Krabbe disease	In-person
325	Amy White	Outcomes of newborn screening for Krabbe disease and their impact on selecting an effective screening approach	In-person
332	Brianna Yund	Complex neurocognitive function and quality of life in Morquio and Maroteaux-Lamy syndromes: A longitudinal investigation	In-person
333	Brianna Yund	Newborn screening, new metrics: Methods for detecting developmental change in very young children	In-person
334	Tong Zhang	Development of multiplexed proteomic quantification of GAA and IDUA signature peptides in dried blood spots and buccal swabs by immuno-SRM-MS/MS for second-tier screening of Pompe disease and Hurler syndrome	In-person
LB-10	Nicholas Buss	Safety, pharmacodynamics and efficacy of AAV9.hCLN2 in preclinical studies	Virtual
LB-18	Taylor Fields	N-acetyl-l-leucine improves symptoms and functioning in Niemann-Pick disease type C (NPC) and GM2 gangliosidosis (Tay-Sachs disease & Sandhoff disease): Results from two parallel, multi-national, rater-blinded clinical trials	Virtual
LB-20	Jaya Ganesh	Preliminary results of the STAAR study, a phase I/II study of isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease	Virtual
LB-21	Michael Gelb	Newborn screening for metachromatic leukodystrophy: Biochemical and molecular analyses	Virtual
LB-29	Jianyong Huang	Development of a high sensitivity biomarker assay to measure β-hexosaminidase A to assess preliminary effectiveness of AAV gene therapy for GM2 gangliosidoses	Virtual
LB-30	Derralynn Hughes	Safety and efficacy of FLT190 for the treatment of patients with Fabry disease: Results from the MARVEL-1 phase 1/2 clinical trial	Virtual
LB-37	Priya Kishnani	Avalglucosidase alfa improves health-related quality of life (HRQoL) in patients with late-onset Pompe disease (LOPD) vs. alglucosidase alfa: Patient-reported outcome measures (PROMs) from the phase 3 COMET trial	Virtual
LB-46	Georgina Morton	The importance of early diagnosis and views on newborn screening in metachromatic leukodystrophy: Results of a caregiver survey in the UK and Republic of Ireland	Virtual
LB-50	Juana Navarrete Martinez	Outcomes of the first institutional screening program in Mexico to screen 6 lysosomal diseases	Virtual
LB-56	Natalia Perez	Development of structurally targeted allosteric regulators for the treatment of neuronopathic Gaucher disease	Virtual
LB-59	Ana Puhl	Repurposing drugs and natural products for CLN1 Batten disease using machine learning	Virtual
LB-61	Aviva Rosenberg	Gaucher patients and family members perceptions of gene therapy as a treatment option	Virtual
LB-62	Sofia Saenz Ayala	Insights on genotype-phenotype correlations for Pompe disease in the newborn screening era	Virtual
LB-65	Anupam Seghal	A bicistrionic AAV9-based gene therapy, TSHA-101, for the treatment of GM2 gangliosidosis: Preliminary results from a phase 1/2 clinical study	Virtual
LB-67	Kate Simmons	Global three-year sponsored MPS testing program: Parallel biochemical and genetic testing informs a timely and accurate diagnosis of MPS VII	Virtual
LB-72	Knut Wittkowski	A novel formulation of alpha-cyclodextrin for the safe (not ototoxic) and convenient (oral) prevention and treatment of lysosomal diseases (LDs)	Virtual

Wednesday, February 9 – Clinical Applications Poster Presentations

2	Magy Abdelwahab	Long term follow up of lymphadenopathy in Egyptian Gaucher disease children and adolescents	Virtual
5	Heather Adams	Version control and crosswalk in cognitive assessment: Transitioning from the fourth to fifth edition of the Wechsler Intelligence Scale for Children (WISC) in children with NCL disorders / Batten disease	In-person
22	Michal Becker-Cohen	Prodromal Parkinsonian features in *GBA* variant carriers	In-person
23	Michal Becker- Cohen	An 18-month report on the safety and efficacy of rapid intravenous velaglucerase alfa infusions in naïve patients with Gaucher disease	In-person
35	Alexander Broomfield	Baby-COMET methodology: A clinical study of the efficacy, safety, pharmacokinetics, and pharmacodynamics of avalglucosidase alfa in treatment-naïve participants with IOPD	In-person
36	Alexander Broomfield	Neurocognitive outcome in mucopolysaccharidosis type 1 (Hurler phenotype) post HSCT.	In-person

37	Alexander Broomfield	Paediatric experience of Fabry patients since the advent of ERT	In-person
39	Barry Byrne	Cipaglucosidase alfa/miglustat versus alglucosidase alfa/placebo in late-onset Pompe disease (LOPD): PROPEL study subgroup analyses	In-person
42	Maria Camprodon Gómez	New severity scale on Fabry disease: Fabry stabilization score (FASTEX) score	Virtual
44	Magdalena Cerón-Rodriguez	Genotype/phenotype correlation from mucopolysaccharidosis type I: Hurler, Hurler-Scheie, and Scheie syndromes and the response to enzymatic replacement therapy	In-person
47	Yin-Hsiu Chien	Immunogenicity of cipaglucosidase alfa/miglustat versus alglucosidase alfa/placebo in late-onset Pompe disease (LOPD): A phase III, randomized study (PROPEL)	In-person
62	James Davison	Mucolipidosis type II growth trajectories and requirement for enteral tube feeding: A single centre review	Virtual
63	Joaquín de Juan-Ribera	Safety reduction of agalsidase beta infusion time in Fabry disease patients	In-person
67	George Diaz	Continued improvement in pulmonary, visceral, biomarker and growth outcomes in children with chronic acid sphingomyelinase deficiency treated with olipudase alfa enzyme replacement therapy: 2-year results of ASCEND-Peds	In-person
69	Jordi Díaz-Manera	Home-infusion experience in patients with Pompe disease receiving avalglucosidase alfa during three clinical trials (COMET, NEO-EXT, and Mini-COMET)	Virtual
71	Tama Dinur	Gaucher disease diagnosis using lyso-Gb1 on dry blood spot samples: Seven years of experience	In-person
72	Imke Ditters	Effect of alglucosidase alfa dosage on survival and walking ability in patients with classic infantile Pompe disease: A multicentre observational follow-up study of the European Pompe Consortium	Virtual
73	Imke Ditters	Safety of home-based infusion of alglucosidase alfa in late onset Pompe disease: 13 years of experience from the Erasmus MC University Medical Center	Virtual
82	Jessica Espolaor	Natural history of Gaucher disease: Description of patients followed at a reference center in São Paulo	Virtual
94	Pilar Giraldo	Recommendations on the follow-up of patients with Gaucher disease in Spain: Results from a Delphi survey	In-person
96	Roberto Giugliani	A double-blind placebo-controlled phase 2 study to evaluate the safety and tolerability of pentosan polysulfate sodium in subjects with mucopolysaccharidosis type VI (MPS VI)	In-person
97	Roberto Giugliani	Vestronidase alfa for the treatment of mucopolysaccharidosis type VII (MPS VII): Updated results from a novel, longitudinal, multicenter disease monitoring program (DMP)	In-person
98	Roberto Giugliani	Prospective longitudinal study of neurological disease trajectory in children living with late-infantile or juvenile onset of GM1 or GM2 gangliosidosis (PRONTO study)	In-person
99	Roberto Giugliani	Long term efficacy and safety of pabinafusp-alfa (JR-141) in Hunter syndrome (MPS-II): 104-week data from the clinical trials in Japan and Brazil	In-person
101	Ozlem Göker-Alpan	Real-world safety and effectiveness of velaglucerase alfa in pediatric patients with Gaucher disease younger than 4 years of age: A combined retrospective and prospective cohort study	Virtual
103	Domingo González-Lamuño	Spanish Fabry and Gaucher disease patients show striking differences in Beliefs about Medicines (BMQ) and Brief Illness Perception (BIPQ) questionnaires	In-person
108	Nathan Grant	Take him home and love him: The experiences of families with Hunter syndrome at home	In-person
109	Nathan Grant	Timing is everything in Hunter syndrome: Differential clinical courses associated with age at initiation of therapy in a sibling pair	In-person
111	Samuel Groeschel	The effect of intrathecal recombinant arylsulfatase A therapy on demyelination load in children with metachromatic leukodystrophy	Virtual
112	Leanne Hagen	Novel neurological findings in an adult patient with Gaucher disease	In-person
113	Takashi Hamazaki	A phase I/II clinical study of intravenous administration of JR-171, a blood-brain barrier-crossing enzyme, in mucopolysaccharidosis type I : An update	Virtual
117	Paul Harmatz	RGX-121 gene therapy for the treatment of severe mucopolysaccharidosis type II (MPS II): Interim analysis of data from the first in-human study	In-person
121	Nadene Henderson	Alternative dosing strategies among a variety of patients with lysosomal diseases	In-person
123	Robert Hopkin	Long-term multisystemic efficacy with migalastat in ERT-naive and ERT-experienced patients with amenable *GLA* variants	In-person
124	Robert Hopkin	A study to evaluate the effect of venglustat on neuropathic and abdominal pain in symptomatic adult patients with Fabry disease	In-person

127	Derralynn Hughes	Design of GALILEO-1, a phase 1/2 safety and efficacy study of FLT201 in adult patients with Gaucher disease type 1	Virtual
128	Derralynn Hughes	Long-term safety and efficacy of pegunigalsidase alfa: A multicenter extension study in adult patients with Fabry disease	Virtual
130	Derralynn Hughes	Living with Pompe disease in the UK: Characterizing the patient journey and burden on physical, emotional and social quality of life	Virtual
131	May Hui	Characterization of pain dimensions in Fabry disease	In-person
136	Majdolen Istaiti	Eliglustat in patients with Gaucher disease previously treated with enzyme replacement therapy: Real-life experience from Israel	In-person
141	Jeanine Jarnes	Phase 1/2 open-label, multi-center study to assess the safety, tolerability and efficacy of a single dose of PBGM01 delivered into the cisterna magna of subjects with type 1 (early onset) and type 2a (late onset) infantile GM1 gangliosidosis	Virtual
144	Simon Jones	Clinical trial update: Ex-vivo autologous haematopoietic stem cell gene therapy in MPS IIIA	In-person
149	Saima Kayani	Preliminary safety data of a phase 1 first in-human clinical trial support the use of high dose intrathecal AAV9/CLN7 for the treatment of patients with CLN7 disease	In-person
151	Aneal Khan	The getting global rare disease insights through technology (GRIT) study: Patient activation and pain management through a digital app for patients with metabolic genetic disease	Virtual
153	Priya Kishnani	The avalglucosidase alfa phase 3 COMET trial in late-onset Pompe disease patients: Efficacy and safety results after 97 weeks	Virtual
154	Priya Kishnani	Avalglucosidase alfa immunogenicity in alglucosidase alfa-experienced participants with Pompe disease: Pooled analysis of clinical trial data	Virtual
156	David Kronn	Mini-COMET study: Safety, biomarker, and efficacy data after avalglucosidase alfa dosing for ≥97 weeks in participants with infantile-onset pompe disease (IOPD) previously treated with alglucosidase alfa who had demonstrated clinical decline	In-person
160	Robin Lachmann	Sustained and continued improvements in pulmonary function, hepatosplenomegaly, dyslipidemia, and disease biomarkers in 5 adults with chronic acid sphingomyelinase deficiency after 6.5 years of olipudase alfa enzyme replacement therapy	In-person
163	Dawn Laney	Development and implementation of an automated severity scoring system to identify patients at possible increased risk for ten lysosomal disorders	In-person
171	Grace Lee	Assessing bone mineral density in Fabry disease	Virtual
174	Olivier Lidove	Insights into patients' expectations and treatment preferences based on the patient needs questionnaire: Interim results from the SATIS-Fab study in Fabry disease	Virtual
175	Aglina Lika	Association between changes over time in pulmonary function and in patient reported outcomes of adult Pompe disease patients	Virtual
189	Eric Mallack	A phase 1/2 open-label, multicenter, dose ranging and confirmatory study to assess the safety, tolerability and efficacy of PBKR03 administered to pediatric subjects with early infantile Krabbe disease (globoid cell leukodystrophy; GALax-C)	In-person
196	Eugen Mengel	Characterization of a late-infantile subtype in GM2-gangliosidosis: First result of the German "Eight At One Stroke: Attention Gangliosidoses" registry	In-person
207	Tahseen Mozaffar	Analysis of pooled data from clinical trials in treatment-naïve patients with late-onset Pompe disease (LOPD) to inform on the efficacy of avalglucosidase alfa	In-person
215	Kinza Noman	Long-term follow-up in an adult patient with Schindler disease	Virtual
216	Peter Nordbeck	Baseline demographics and clinical characteristics of patients enrolled in the followME Fabry Pathfinders Registry	In-person
217	Ian O'Connor	Incidental diagnosis of lysosomal diseases by expanded carrier screening and direct-to-consumer genetic testing	In-person
223	Maria Carolina Oliveira	The impact of COVID-19 on Brazilian children with MPS: Advocate group perspective	In-person
229	Marc Patterson	Persistent effect of arimoclomol in patients with Niemann-Pick disease type C: 24-month results from an open-label extension of a pivotal phase 2/3 study	In-person
234	Gisele Perillo	Clinical and laboratory profile of a pediatric Fabry disease cohort followed at a Brazilian reference center	Virtual
240	Paivi Pietila Effati	Fabry cardiomyopathy in Finland: A Fabry registry study	In-person
241	Nishitha Pillai	Bone marrow transplantation in multiple sulfatase deficiency: 1 year follow up	In-person

245	Lynda Polgreen	A Hunter syndrome sibling pair: Differential effects of age at initiation of enzyme replacement therapy on growth, orthopedic disease, and daily living skills	In-person
247	Lynda Polgreen	Phase I/II clinical trial of anakinra in Sanfilippo syndrome: Outcomes from 8 weeks of a palliative treatment	In-person
254	Allegra Quadri	Outcome in infants treated with very early ERT supports newborn screening for mucopolysaccharidosis type II	In-person
257	Uma Ramaswami	Lyso-Gb3 as a biomarker for renal and cardiac involvement in Fabry disease: An analysis from the Fabry Outcome Survey (FOS)	In-person
258	Uma Ramaswami	Migalastat HCl 150 mg every other day is well-tolerated and efficacious in adolescent patients with Fabry disease	In-person
259	Shoshana Revel-Vilk	Markers of inflammation and alpha degranulation defect of platelets in patients with Gaucher disease	In-person
266	Paula Rozenfeld	Effect of COVID19 pandemic on Argentinian Fabry and Gaucher patients	Virtual
271	Linda Scheffers	Effects of enzyme replacement therapy on cardiac function and structure in classic infantile Pompe disease: Up to 22 years of follow-up	In-person
273	Angela Schulz	Real-world clinical outcomes of intraventricular cerliponase alfa in CLN2 disease: 4.5-year update from an independent ongoing observational study	In-person
280	Elsa Shapiro	Improving metrics to measure change: Developmental growth scores	In-person
289	Carolina Souza	Behavioral improvement in a 9-year-old patient with MPS II undergoing enzyme replacement therapy with pabinafusp alfa: A case report	In-person
292	Rodrigo Starosta	Treatment dilemmas in an individual diagnosed with infantile-onset Pompe disease and sickle-cell anemia	In-person
293	Karolina Stepien	Implications for neuropsychology assessments in adult mucopolysaccharidosis: A systematic review to inform service development in a large tertiary lysosomal disorders centre	Virtual
294	Theresa Stokes	The role of the inherited metabolic disorders coordinator	Virtual
302	Cynthia Tifft	Phase 1/2 trial of AXO-AAV-GM1 (AAV9-GLB1) gene therapy for infantile- and juvenile-onset GM1 gangliosidosis	In-person
303	Mateus Torres	Niemann-Pick disease type C: A description of patients followed at a reference center in São Paulo - a retrospective study	Virtual
304	Antonio Toscano	Multicenter, non-interventional, double cohort study to assess the safety of alglucosidase alfa and laronidase in real-world home infusion setting	Virtual
307	Ecenur Tuc Bengur	Psychosine predicts age of onset in babies with Krabbe disease	In-person
315	Nato Vashakmadze	Outcomes of idursulfase treatment in non-neuropathic mucopolysaccharidosis type II: A family case	Virtual
318	Jerry Vockley	An open-label, phase 1/2 trial of gene therapy 4D-310 in adult males with Fabry disease	In-person
320	Raymond 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	In-person
322	Melissa Wasserstein	Two-year results of the ASCEND trial of olipudase alfa adults with chronic acid sphingomyelinase deficiency show parallel improvements in former placebo patients and further improvement in continuing olipudase alfa patients	In-person
326	Kaylee Williams	Dual genetic diagnoses in a lysosomal disorders patient population	In-person
328	Hiroyuki Yamakawa	The need for home enzyme replacement therapy for patients with lysosomal disease in Japan	Virtual
337	Natalia Zhurkova	Clinical, genetic characteristics in Russian patients with Hurler syndrome	Virtual
LB-01	Mario Aguiar	Successful dose escalation of olipudase alfa enzyme replacement therapy in patients with chronic acid sphingomyelinase deficiency: Rationale and clinical trial experiences in children and adults	Virtual
LB-02	Patricio Aguiar	Migalastat in real world setting: A single center experience	Virtual
LB-03	Mariam Ahmed	An integrated approach evaluating the relationship between intrathecal idursulfase-IT treatment, cerebrospinal fluid glycosaminoglycans and cognitive function in mucopolysaccharidosis type II	Virtual
LB-05	Dustin Armstrong	Emptying the "basket": VAL-1221 glycogen clearance	Virtual
LB-07	Julie Batista	Overall slowing of decline in kidney function and reduction in severe clinical events in Fabry disease patients treated with agalsidase beta: A matched analysis	Virtual
LB-09	Elizabeth Berry-Kravis	Clinical benefit of treatment with adrabetadex in subgroups of patients with Niemann-Pick disease type C1	Virtual

LB-13	Marie-Anne Colle	FOXO3a over-expression in Pompe disease alleviates muscle impairments autophagic buildup	Virtual
LB-14	Carmen Silvia Curiati Mendes	Alpha-mannosidosis: Case report and follow-up proposal	Virtual
LB-15	Javier de las Heras	Importance of timely treatment initiation in infantile-onset Pompe disease: A single-centre experience	Virtual
LB-16	Simon Dulz	Retinal degeneration in MPS I quantified by optical coherence tomography (OCT) imaging	Virtual
LB-17	Yoshikatsu Eto	Intrafamilial differences of *in vitro* and *in vivo* amenability to migalastat in Fabry disease	Virtual
LB-22	Arunabha Ghosh	Mortality and cause of death in individuals with MPS I: Data from the MPS I Registry	Virtual
LB-23	Sabire Gökalp	An alternative for early detection of cardiac involvement in Gaucher disease type 1: Speckle tracking echocardiograpy	Virtual
LB-25	Robert Henderson	Intravitreal cerliponase alfa for the treatment of neuronal ceroid lipofuscinosis type 2 (CLN2) related retinal dystrophy: A first in-man report of ocular enzyme replacement	Virtual
LB-27	Clara Hildebrandt	Assessing barriers to enzyme replacement therapy for lysosomal disorders at a single pediatric center	Virtual
LB-28	Myrl Holida	Safety and efficacy of pegunigalsidase alfa administered every 4 weeks in patients with Fabry disease: Results from the phase 3, open-label, BRIGHT study	Virtual
LB-31	Derralynn Hughes	A collaborative approach to developing international clinical patient centric guidelines for Gaucher disease	Virtual
LB-32	Victoria Jensen	Long-term correction of mucopolysaccharidosis type IIIB disease phenotype following central nervous system administration of AAV-NAGLU	Virtual
LB-36	Sarah Kim	Cerebrospinal fluid chitotriosidase as a surrogate endpoint of the efficacy of the PS gene editing system in neurodegenerative lysosomal diseases	Virtual
LB-38	Lise Kjems	TransportNPC: A phase 3 global trial of trappsol®cyclo™ administered intravenously to patients with Niemann-Pick disease type C1 (NPC1)	Virtual
LB-39	Olivier Lidove	Regards Croisés: A national survey on clinical pathways of patients with Fabry disease	Virtual
LB-41	Monica A. Lopez-Rodriguez	Screening for late onset Pompe disease by dried blood spot in departments of internal medicine in Spain	Virtual
LB-42	Deborah Marsden	Management and progression of arginase 1 deficiency over 2 decades of follow-up	Virtual
LB-43	Naresh Kumar Meena	Liver-directed and systemic AAV gene transfer approaches for Pompe disease therapy	Virtual
LB-45	Alexandra Morrison	Age at diagnosis and previous diagnoses of Fabry disease patients in the UK	Virtual
LB-48	Joseph Muenzer	Clinically meaningful benefit of intrathecal idursulfase-IT in patients younger than 6 years old with mucopolysaccharidosis type II and missense iduronate-2-sulfatase gene variants: A *post hoc* analysis	Virtual
LB-51	Dmitriy Niyazov	Mucopolysaccharidosis type I: Timely diagnosis and treatment avoid multiple surgeries	Virtual
LB-52	Albina Nowak	Covid-19 infection in Fabry disease: A systematic cohort study	Virtual
LB-53	Albina Nowak	Health-related quality of life in Fabry disease: A cross-sectional international multi-center study	Virtual
LB-54	Cecile Paquet Luzy	A first-in-human, randomized, double-blind, placebo-controlled, ascending single- and multiple-dose study to evaluate the safety, tolerability, pharmacokinetics and pharmacodynamics of the L-ido azasugar AZ-3102 in healthy volunteers	Virtual
LB-60	Harleigh Quick	The impact of Fabry disease on growth in males	Virtual
LB-63	Angela Schulz	Long-term treatment with intracerebroventricular cerliponase alfa for children with CLN2 disease: Safety and efficacy after >5 years	Virtual
LB-66	Elis Silva	Gaucher disease type 2 presenting onset with congenital cytomegalovirus and fatal outcome with hemophagocytic syndrome	Virtual
LB-68	Sophie Thomas	The burden of disease in metachromatic leukodystrophy: Results of a caregiver survey in the UK and Republic of Ireland	Virtual
LB-73	Karen Yee	Categorizing associations between cognitive phenotype and genotype in patients with mucopolysaccharidosis type II	Virtual

Thursday, February 10 – Contemporary Forum Poster Presentations

1	Nina Aaron	Liver-specific AAV gene therapy corrects lipid storage in LAL-D model mice but does not prevent lipid accumulation in acquired fatty liver model mice	In-person
11	Lene Andersen	Arimoclomol reduces levels of biomarkers of lipid burden in patients with Niemann-Pick disease type C	In-person
14	Ramesh Arjunji	Systematic literature review of the clinical effectiveness, safety, quality of life, epidemiology and economic burden associated with cystinosis	In-person
17	Olulade Ayodele	Novel insights into mucopolysaccharidosis type II based on an analysis of genetic variants in 763 patients	In-person
21	Isabela Batsu	ELISAFE: Baseline characteristics from an observational study to evaluate real-world safety of eliglustat in patients with Gaucher disease	Virtual
33	Livia Breznik	Behavioral characterization of homozygous 6^{neo} mice as model of Pompe disease	Virtual
48	Alexandra Chiorean	Clustered analysis of Fabry disease progression in a large US electronic health records database: A retrospective observational cohort study	In-person
49	Yoonjin Cho	Beyond the normative data: Understanding the Bayley Scales of Infant Development version 3 (BSID-III)	In-person
50	Yoonjin Cho	Beyond the normative data: Understanding the Mullen Scales of Early Learning (MSEL)	In-person
57	Ruda Cui	Identification of cathepsin D as a potential biomarker of CLN5 function in an early stage potency assay	Virtual
58	Filipa Curado	Defining the role of Lyso-Gb1 as a biomarker over 12 months after first initiation of enzyme replacement therapy in patients with Gaucher disease in LYSO-PROVE study	Virtual
66	Kenneth Der	Translational pharmacokinetic-pharmacodynamic (PKPD) model of ST-920 from mouse to human in Fabry disease	In-person
79	Patti Engel	Genetic testing and awareness campaign for rare movement disorder	In-person
81	Maria Escolar	FBX-101, an intravenous AAV gene replacement therapy given after infusion of hematopoietic stem cells, extends efficacious dose ranging and corrects disease manifestations in Krabbe disease	In-person
87	Francois-Xavier Frapaise	A study of intracisternal administration of adeno-associated viral vector serotype rh.10 carrying the human β-galactosidase cDNA for the treatment of GM1 gangliosidosis: Preliminary results of the safety cohort	Virtual
89	Tomoki Fukatsu	Suppression of anti-alpha-GalA antibody production by blockade of T-cell costimulation in mice	Virtual
102	Jessica Gómez	Application of artificial intelligence to predict protein biomarker candidates for the assessment of prognosis in patients with metachromatic leukodystrophy	Virtual
105	Russell Gotschall	M011: A novel highly phosphorylated β-glucocerebrosidase enzyme with broader tissue biodistribution for the treatment of Gaucher disease	Virtual
106	Russell Gotschall	M021: A uniquely glycosylated, highly phosphorylated acid-alpha glucosidase enzyme replacement therapy for the treatment of Pompe disease	In-person
114	Christiane Hampe	Iduronidase-transposed human B lymphocytes correct enzyme deficiency and glycosaminoglycan storage disease in immunodeficient mucopolysaccharidosis type I mice	In-person
118	Jeffrey Haroldson	Patient and physician perspectives inform clinical trial design for a single intravenous dose of HMI-203, a gene therapy candidate for adults with mucopolysaccharidosis type II (MPS II, Hunter syndrome)	In-person
119	Jeffrey Harris	A blinded randomized phase 2/3 study of the efficacy and safety of intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II	In-person
120	Andrew Hedman	A novel S1S3 phosphotransferase co-expression gene therapy platform for lysosomal disorders	In-person
122	Julian Homburger	Genetic reduction of muscle glycogen is well tolerated in UK Biobank participants	In-person
134	Elizabeth Hwang-Wong	Defining phenotype reversibility in lysosomal disease: Leveraging a COIN model in mucopolysaccharidosis type VI (MPS VI)	In-person
135	Asuka Inoue	Efficacy of an anti-human transferrin receptor antibody-fused N-sulfoglucosamine sulfohydrolase in mucopolysaccharidosis type IIIA mice	Virtual
138	Skyler Jackson	Patient voice in access studies	In-person

139	Leslie Jacobsen	Efficacy of gene therapy in a CLN5 sheep model using a dual route of administration supports a first-in-human clinical trial	In-person
140	Leslie Jacobsen	A natural history and outcome measure discovery study of variant late infantile neuronal ceroid lipofuscinosis type 5 and variant late infantile neuronal ceroid lipofuscinosis type 7	In-person
142	Karl Johe	A one-time treatment to continuously and permanently deliver lysosomal enzymes to the CNS	In-person
143	Franklin Johnson	Plasma total GAA protein PK profiles differ between cipaglucosidase alfa/miglustat and alglucosidase alfa	Virtual
146	Venediktos Kapetanakis	Analysis of overall survival in patients with acid sphingomyelinase deficiency type B using the standardized mortality ratio method	Virtual
161	Kyle Landskroner	Characterization of AZ-3102, a novel brain-penetrant small molecule, in the Niemann-Pick disease type C mouse model	In-person
165	Heather Lau	A natural history study of Sanfilippo syndrome type D (MPS type IIID)	In-person
170	Fernanda Leal-Pardinas	Connect: Designing a first-in-human gene replacement therapy clinical trial for CLN1	In-person
172	Ashley Leek	Distance to expert care for patients with lysosomal disorders enrolled in a real-world data research platform	Virtual
177	Tina Loeffler	Characterization of in vivo and in vitro drug screening models for Gaucher disease based on GBA-D409V-KI mice	Virtual
180	Mariah Lopshire	GM2-gangliosidosis patient journey: Results from interviews with late-onset GM2-gangliosidosis patients and frontline treaters show that the lack of disease awareness significantly delays diagnosis	In-person
184	Nicole Lyn	Neuropathic and abdominal pain items of the Fabry Disease Patient-Reported Outcome (FD-PRO) show robust measurement properties in treatment naïve Fabry patients	In-person
185	Nicole Lyn	Establishing the content validity of the Fabry Disease-Patient Reported Outcome (FD-PRO) for adolescent patients with Fabry disease	In-person
186	Alasdair MacCulloch	Fabry disease, symptom burden, health-related quality of life burden and treatment satisfaction	Virtual
187	Alasdair MacCulloch	Treatment preferences in Fabry disease, a discrete choice experiment in the UK and Denmark	Virtual
193	Maggie McCue	GOAL-GD: A smartphone application to enhance patient engagement in a real-world study of treatment switching in patients with Gaucher disease type 1	Virtual
194	Heather McLaughlin	Detect Lysosomal Storage Diseases: A no-charge, sponsored, testing program that enables access to genetic testing, treatment, and clinical trials for individuals with suspected lysosomal disorders	In-person
195	Thomas Mechtler	Prospective of newborn screening and rare disease diagnostic initiatives in Europe	In-person
205	Hideto Morimoto	Enzyme replacement with a blood-brain barrier-penetrating antibody-fused alfa-L-iduronidase prevents neurobehavioral performance of mucopolysaccharidosis type I mice	Virtual
218	Christina Ohnsman	Pilot study of novel optokinetic nystagmus-based visual acuity test in children with CLN2 disease	In-person
220	Petra Oliva	Diagnostic strategy for suspected cases of Fabry disease	In-person
221	Petra Oliva	Differential diagnosis of Niemann-Pick disease type A/B in cases of suspected Gaucher disease	In-person
222	Petra Oliva	Newborn screening for metachromatic leukodystrophy in northern Germany	In-person
224	Ryan Oliver	Peptide-conjugated phosphorodiamidate morpholino oligomers for the treatment of late-onset Pompe disease	In-person
227	Francis Pang	The cost-effectiveness of atidarsagene autotemcel for the treatment of metachromatic leukodystrophy in France	Virtual
228	Francis Pang	Quality of life and caregiver burden in metachromatic leukodystrophy: Results from a cross-national study of 6 countries	Virtual
230	Erika Pearson	Development of a novel encapsulated non-viral cell-based, BBB-penetrant therapy for MPS I	In-person
231	Erika Pearson	Development of a novel encapsulated non-viral cell-based therapy for MPS VI	In-person
233	Jordi Pérez-López	The prevalence of carriers for lysosomal disorders in a large Spanish cohort	In-person
235	M. Judith Peterschmitt	Pharmacokinetics and biomarker responses in patients with Gaucher disease type 3 or *GBA*-associated Parkinson disease treated with venglustat	Virtual
236	Nikolaj Petersen	Arimoclomol increases the transcription of lysosomal genes, including *NPC1* and *NPC2*, to facilitate lysosomal function	In-person

237	Miloš Petrović	Venglustat, a novel brain-penetrant glucosylceramide synthase inhibitor, for GM2 gangliosidosis and related diseases: Phase 3 AMETHIST trial design	Virtual
238	Dawn Phillips	Natural history of neurodevelopment in neuronopathic mucopolysaccharidosis type II (MPS II): Mullen Scales of Early Learning (MSEL) cognitive, motor and language developmental trajectories	In-person
239	Dawn Phillips	The expanded neuronal ceroid lipofuscinosis 2 (CLN2) clinical rating scale for motor and language function: Development and inter-rater reliability	In-person
244	Robin Pokrzywinski	A qualitative study to understand caregivers' burden of acid sphingomyelinase deficiency (ASMD)	In-person
248	Maria Praggastis	BBB-targeted GAA delivered as gene therapy treats CNS and muscle in Pompe disease model mice	In-person
252	Ruth Pullikotil-Jacob	Analysis of survival in patients with acid sphingomyelinase deficiency type B using a large, deidentified US electronic health record database	In-person
256	Marianna Raia	All for one, not one for all: Developing and implementing a multifaceted approach to NBS education equitable access to newborn screening education	In-person
262	Camille Rochmann	Identifying patients with Gaucher disease type 3 (GD3) in the Optum's de-identified Market Clarity Database: A clustering analysis	In-person
263	Camille Rochmann	Identification of late-onset GM2 gangliosidoses (LOGG) patients using Optum's de-identified Market Clarity Database	In-person
265	Tom Rouwette	Medical information consumption and sharing practices in lysosomal diseases: A clinician perspective	Virtual
267	Es-Said Sabir	Determination of urinary mannose by gas liquid chromatography mass spectrometry using a dried urine spot	In-person
269	Rosario Sánchez	First results from The Spanish Fabry Women Study: A retrospective observational study describing the phenotype of female carrying genetic variants associated to Fabry disease	Virtual
270	Maurizio Scarpa	Continued improvement in pulmonary outcomes in 3 clinical trials of olipudase alfa in children and adults with chronic acid sphingomyelinase deficiency treated for 2 to 6.5 years	In-Person
276	Tania Seabrook	AAVHSCs and nervous system-targeted gene therapy for lysosomal disorders	Virtual
281	Patty Sheehan	PR001 gene therapy increased GCase activity and improved Gaucher disease type 1 phenotypes in mouse models	Virtual
286	Laura Smith	Summary of nonclinical data for gene therapy developmental candidate HMI-203 for mucopolysaccharidosis type II (MPS II, or Hunter syndrome)	In-person
288	Lisa Sniderman King	*Project Searchlight* Gaucher study design: Real-world evaluation and validation of a rare disease algorithm to identify persons at risk of Gaucher disease using data from electronic health records in the United States	In-person
290	Gillian Spitzley	The supportive care needs of parents following diagnosis of late onset Pompe disease through newborn screening	Virtual
291	Lisa Stanek	Sulfatide accumulation begins as early as four months of age in ARSA knockout mice	Virtual
305	Alayna Tress	Industry working with rare disease patient advocacy organizations to further the awareness of lentiviral gene therapy clinical studies for Fabry disease and Gaucher disease type 1	In-person
321	Ibrahim Warsi	Symptoms of Fabry disease in adolescents	In-person
327	Yannan Xi	Pharmacology of small molecule inhibitors of GYS1 in a mouse model of Pompe disease	In-person
329	Narutoshi Yamazaki	Enhanced osteoblastic differentiation of parietal bone in a novel murine model of mucopolysaccharidosis type II	Virtual
331	Karen Yee	Clinical investigator perspectives on the effects of intrathecal idursulfase-IT treatment in children with neuronopathic mucopolysaccharidosis type II	Virtual
335	Xiaoli Zhang	Functional modeling of human lysosomal acid alpha-glucosidase variants	Virtual
336	Xiangli Zhao	Progranulin deficiency markedly exacerbates Gaucher disease phenotypes in Gba1 mutant mice	Virtual