Authors who accepted an ePoster presentation are assigned to present their abstract live during one of five ePoster sessions, based on the final abstract category for each abstract. The ePosters will open at 2:30pm EST on Monday, February 8, 2021, and will remain open throughout WORLD*Symposium* 2021. Live Q&A will only occur during the assigned times as listed below.

All ePosters will be located in the Virtual Exhibit & ePoster Hall:
Basic Science Abstracts will be presented on **Monday, February 8 from 2:30-3:30pm EST**
Translational Research Abstracts will be presented on **Tuesday, February 9 from 2:30-3:30pm EST**
COVID-19 and Clinical Trials Abstracts will be presented on **Wednesday, February 10 from 2:30-3:30pm EST**
Contemporary Forum Abstracts will be presented on **Thursday, February 11 from 2:30-3:30pm EST**
Late-Breaking Abstracts will be presented on **Friday, February 12 from 2:30-3:30pm 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.

Monday, February 8 – ePoster Presentations

13	Christiane Auray-Blais	The importance of mass spectrometry-based untargeted metabolomic approaches for biomarker discovery in lysosomal diseases
20	Claubia Bender	Effects of acid sphingomyelinase deficiency on oral health and craniofacial development
29	Daniela Castillo-Garcia	A novel nonsense LAMP2 variant associated with Danon disease in a pediatric male: A case report
30	Magdalena Cerón-Rodriguez	Female child with Fabry disease and two other genetic diseases: Spherocytosis, and congenital hypothyroidism - A case report
36	Chloe Christensen	Base editing of the N370S mutation in Gaucher disease skin fibroblasts
46	Simona D'Amore	Oral aspects of Gaucher disease: A case report
59	Tama Dinur	Impact of long-term enzyme replacement therapy on Lyso-Gb1 in patients with Gaucher disease: Comparison between 3 enzymatic formulations
66	Hassan Elsaid	Preliminary proteomic analysis reveals less abundant endosome-related proteins in gla- knockout zebrafish (Danio rerio)
76	Chaitanya Gadepalli	Assessment and management of tracheomalacia in adult mucopolysaccharidosis type II: A case report
79	Srinitya Gannavarapu	Increased sulfatide disrupt mitochondrial function in Schwann and mesenchymal stromal cells in metachromatic leukodystrophy
80	Eric Joshua Garcia	RNA-seq analysis in three Gaucher disease sib-pairs discordant for Parkinson disease
96	Coy Heldermon	The beta-glucuronidase intracisternal A particle insertion model results in similar overall MPS VII phenotype as the single base deletion model when on the same C57BL/6J mouse strain
105	Ayuko Iverson	Clinical course of patients with Gaucher-associated Parkinson disease
111	Shih-hsin Kan	iPSC-derived human neural stem cells engraft in the brains of immunocompromised MPS I mice
114	Gustavo Maegawa	Psychosine-reducing molecules as therapies for globoid-cell leukodystrophy
115	Marcel André Kelkel	Lysosphingolipid detection using a non-invasive urine multiplex mass spectrometry approach for various lysosomal diseases
117	Aram Kim	Case report: Neurologic outcome after stem cell transplant in a patient with neuronopathic Gaucher disease
124	Aditi Korlimarla	New insights into GI manifestations in late-onset Pompe disease: Lessons from the bench and bedside
125	Francyne Kubaski	Report of the first Brazilian patients with MPS IIID, with the observation of an unexpected increase of di-sulfated keratan sulfate

126	Francyne Kubaski	Screening for Niemann-Pick disease type C in Latin American using Lyso-SM-509 measurement in dried blood spots
127	Francyne Kubaski	Quantification of lysosphingomyelin and lysosphingomyelin-509 for the screening of acid sphingomyelinase deficiency
136	Malte Lenders	Clinical impact of orally delivered alpha-galactosidase A on gastrointestinal symptoms in patients with Fabry disease
140	Renuka Limgala	Mast cell stabilizers in management of IgE independent mast cell activation in infusion related reactions in patients with Fabry disease
145	Laura López de Frutos	Testing new biomarkers for lysosomal diseases
156	Angela McCall	Glycogen accumulation in smooth muscle in the Pompe disease mouse
159	Iskren Menkovic	Metabolomic study for the identification and characterization of novel Gaucher disease biomarkers
163	Hideto Morimoto	Reduction of heparan sulfate in the brain by pabinafusp alfa results in prevention of neurodegeneration and neurocognitive impairment in a mouse model of mucopolysaccharidosis type II
167	Joseph Muenzer	Genotype-phenotype findings in patients with mucopolysaccharidosis type II (MPS II): Data from the Hunter Outcome Survey
169	Behzad Najafian	Direct intercellular cross-correction of α-galactosidase-A deficiency in Fabry disease podocytes through tunneling nanotubes in a mixed cell culture model
172	Stephanie Newman	AAV9-hARSA decreases sulfatide accumulation in the aged ARSA-/- mouse model for metachromatic leukodystrophy
178	Andrew Oldham	MPS VII - extending the classical phenotype
182	Christopher Orsborne	Mosaic Fabry disease in a male presenting as hypertrophic cardiomyopathy: When enzyme levels are not enough
194	Gani Perez	RNA-seq analysis of GBA1 haploinsufficiency in brain samples from a Parkinson mouse model
200	Tyler Pierson	Human induced pluripotent stem cell models for CLN6
204	Michael Przybilla	Examination of a blood-brain barrier targeting β-galactosidase-monoclonal antibody fusion protein in a murine model of GM1-gangliosidosis
205	Maria Alejandra Puentes-Tellez	A perspective on research, diagnosis, and management of lysosomal disorders in Colombia: An update
220	Richard Sam	Generating pluripotent stem-cell derived organoids to model Gaucher disease type 2
221	Gustavo Maegawa	CNS-targeting exosomes: A strategy to treat neurological lysosomal disorders
227	Ida Schwartz	Metab-Latam, four months of experience in the email discussion group
228	Ida Schwartz	A decade of molecular diagnosis of mucolipidosis II and III in Brazil: A pooled analysis of 32 patients
229	Ida Schwartz	Brazilian patients with Gaucher disease: Haplotype analysis
234	Allen Seylani	Novel regulatory function of GCN5L1 in lysosomal tubulation and biogenesis
240	Hatice Serap Sivri	Clinical characteristics and journey to diagnosis in patients with mucopolysaccharidosis type VII
243	Luis Soares	MPSBase: Comprehensive repository of differentially expressed genes for mucopolysaccharidoses studies
251	Xinze Tan	Genotype-phenotype correlation of MPS II: A meta-analysis
255	Heidy Triana	Production and characterization of a recombinant alpha-N-acetyl glucosaminidase enzyme in the Pichia pastoris yeast
262	Gerda Cristal Villalba Silva	Neuro-networks investigating the neurological impairment of mucopolysaccharidoses using a system biology approach
271	Matheus Wilke	Lyso-Gb1 levels in sisters with Gaucher disease type 1: A case report
273	Elizabeth Woo	Exploration of the role of whole exome sequencing variants in GBA1-associated Parkinson disease

Tuesday, February 9 – ePoster Presentations

2	Alia Ahmed	MPS-specific physical symptom score (PSS) and adaptive functions in MPS IVA: A cross sectional study
8	Pam Andrews	Gathering evidence for newborn screening for Niemann-Pick disease type C
14	Rachel Bailey	Preclinical studies to support the intrathecal delivery of scAAV9/SUMF1 as a gene replacement therapy for multiple sulfatase deficiency
17	Roberto Barriales-Villa	Red Fabry: First year results of a Spanish pedigree project on Fabry disease
24	Nicholas Buss	Subretinal injection of RGX-381 to cynomolgus monkeys leads to supraphysiological levels of TPP1 in the eye
31	Xin Chen	Preclinical results in rodents strongly support clinical evaluation of scAAV9/MFSD8 as a potential gene therapy for CLN7 patients
38	Jennifer Cohen	Early diagnosis and treatment of infantile-onset Pompe disease via newborn screen
42	Jonathan Cooper	Devising effective enzyme replacement therapy for infantile onset neuronal ceroid lipofuscinosis (CLN1 disease)
53	John Day	A phase I/II open-label gene replacement clinical study for late onset Pompe Disease
54	Emily Daykin	DeepGestalt as a potential novel method to help discriminate between the Gaucher disease subtypes
62	Halil Dündar	The chemical chaperone 4-phenylbutyrate enhances alpha-galactosidase activity subsequent to stopcodon readthrough therapy with triamterene in Fabry disease R227X fibroblasts
68	Kristina Elvidge	A new initiative to tackle childhood dementia
78	Jaya Ganesh	A phase I/II multicenter gene therapy clinical study for Fabry disease
86	Jason Glanzman	A tractography tool that detects abnormalities in function in early metachromatic leukodystrophy
87	Shelly Goomber	Functional analysis and clinical curation of human acid alpha glucosidase (GAA) variants of unknown significance (VUS) screened from infants diagnosed with Pompe disease via newborn screening (NBS)
104	Jackie Imrie	International Niemann-Pick Disease Registry: Establishing worldwide inclusivity
118	Brittany Kimball	Comparison of long-term outcomes for survivors among hematopoietic stem cell transplant subjects using the Living Independently Study Assessment (LISA 2.0)
128	Francyne Kubaski	Newborn screening for six lysosomal diseases in Brazil: Pilot study update
132	Ashley Lahr	Analysis of parent perception of newborn screening for lysosomal disorders
148	Troy Lund	Effect of supraphysiological alpha-L-iduronidase (IDUA) expression on skeletal manifestations in mucopolysaccharidosis type I (MPS I) mice following ex vivo lentiviral vector transduction of hematopoietic stem cells
153	Margaux Masten	Epidemiology and access to expert care for the neuronal ceroid lipofuscinoses (NCLs)
154	Margaux Masten	Genotype-phenotype associations in CLN3 disease
155	Margaux Masten	Diagnostic confidence for CLN3 disease
175	Loreanne Oh	An algorithm for early diagnosis of mucopolysaccharidosis types IIIA and B
177	Torayuki Okuyama	Prevention of cognitive decline in patients with neuronopathic mucopolysaccharidosis type II treated by intracerebroventricular enzyme replacement therapy: 100-week results of an open-label phase 1/2 study
201	Nishitha Pillai	Evaluation of the effectiveness of hematopoietic stem cell transplantation in multiple sulfatase deficiency
207	Bryan Pukenas	Intracisternal administration of AAV9 gene therapies to target the central nervous system
208	Allegra Quadri	Population-based newborn screening for mucopolysaccharidosis type II: A single center's experience
209	Julian Raiman	Update on safety and efficacy results for phase I/II trial of hydroxypropyl betacyclodextrin (HPâCD) administered intravenously in patients with Niemann-Pick disease type C1

226	Ida Schwartz	Phlebotomy in the treatment of hemosiderosis associated with Gaucher disease
235	Patty Sheehan	PR001 gene therapy increased GCase activity and improved neuronopathic Gaucher disease phenotypes
236	Patty Sheehan	PR001 gene therapy increased GCase activity and improved Gaucher disease type 1 phenotypes in mouse models
241	Miles Smith	Ex vivo lentiviral transduction of hematopoietic stem cells in mucopolysaccharidosis type II (MPS II) mice achieves high levels of systemic iduronate-2-sulfatase (IDS) enzyme activity and normalization of glycosaminoglycans (GAGs)
244	Chanan Stauffer	Dual therapy with migalastat and agalsidase-beta in a patient with Fabry disease with progressing hypertrophic cardiomyopathy
249	Dean Suhr	RANSIP newborn screening program: Working to bring early diagnostics, inclusion, and therapeutic access to metachromatic leukodystrophy patients
256	Michael Ueberall	Development of a Fabry disease screening tool for chronic pain patients - step 1: Categorization based on phenotypic risk profiles
260	Sarah Viall	Oregon's experience with newborn screening for Fabry, Gaucher, Pompe and mucopolysaccharidosis type I
264	Raymond Wang	First in-human intracisternal dosing of RGX-111 (adeno-associated virus 9/human α-L-iduronidase) for a 20-month-old child with mucopolysaccharidosis type I (MPS I): 1 year follow-up
278	Brianna Yund	A longitudinal report of neurocognitive abnormalities and their impact on quality of life in non-neuronopathic MPS II
281	Zion Zibly	Safety of intracisternal administration of AAV9 based gene therapy: Case series of PR001 in infants with Gaucher disease type 2

Wednesday, February 10 – ePoster Presentations

1	Carlos Acosta-Rodriguez-Bueno	Impact of SARS-CoV-2 pandemic on the care for patients with lysosomal disorders: The experience of a Mexican pediatric center
7	Marcio Andrade-Campos	Social distancing, home-infusion and oral therapy: Evolving paradigms on Gaucher disease management in Spain during COVID-19 pandemic
11	Jane Ashworth	Assessment and outcomes of MPS patients having corneal transplantation
18	Michael Beck	Fabry Outcome Survey (FOS): Highlights from a 20-year patient registry of Fabry disease
23	Lindsey Bulleid	Appraisal of the use of FASTEX in clinical practice in the All Wales Inherited Metabolic Disease Service
25	Valeria Calbi	Lentiviral haematopoietic stem cell gene therapy for metachromatic leukodystrophy: Results in nine patients treated with a cryopreserved formulation of OTL-200
26	M. Camprodon	Characterization of surgical procedures in patients with mucopolysaccharidosis
27	Maria Domenica Cappellini	Impact of SARS-CoV-2 infection on Gaucher disease patients in Italy
28	Francesca Carubbi	Liver steatosis is highly prevalent and is associated with metabolic risk factors and liver fibrosis in adult patients with type 1 Gaucher disease
37	Tzu-Hung Chu	First MPS case in Kingdom of Eswatini
39	Jennifer Coker	Phenotypic target organ and biomarker variation within a family with late-onset Fabry disease
40	Duncan Cole	Clinical outcomes in an adult patient with alpha-mannosidosis treated with velmanase alfa for 5 years
45	Kirsten Cowley	RGX-121 gene therapy for severe mucopolysaccharidosis type II (MPS II): A clinical program to address central nervous system manifestations
47	Amanda Daniel	Development and evaluation of a nurse-led Anderson-Fabry clinic in Wales
48	Amanda Daniel	The impact of the COVID-19 pandemic on patients with inherited metabolic conditions: A survey in Wales
52	James Davison	Mini-COMET study: Effects of repeat avalglucosidase alfa dosing on ptosis in participants with infantile-onset Pompe disease (IOPD) who were previously treated with alglucosidase alfa
55	Emily de los Reyes	Single-dose AAV9-CLN6 gene transfer slows the decline in motor and language function in variant late infantile neuronal ceroid lipofuscinosis 6: Interim results from phase 1/2 trial
58	Mazen Dimachkie	NEO1/NEO-EXT studies: Safety and exploratory efficacy of repeat avalglucosidase alfa dosing after up to 6 years in participants with late-onset Pompe disease (LOPD)
61	Christina Dumke	Clinical characterization and therapy discussion of the p.Asp313Tyr variant in GLA
63	Marina Dutra-Clarke	Variable clinical features of patients with Fabry disease and outcome of enzyme replacement therapy
70	Ulla Feldt-Rasmussen	Long-term treatment with migalastat 150 mg every other day is associated with sustained cardiac efficacy and is well tolerated
71	Luca Fierro	Gaucher disease and SARS-CoV-2 infection: Experience from 181 patients in New York
74	Aline Frey	Measures to mitigate disruption due to the COVID-19 pandemic of the MODIFY phase 3 pivotal trial in patients with Fabry disease
75	Francesca Fumagalli	Lentiviral hematopoietic stem and progenitor cell gene therapy provides durable clinical benefit in early-symptomatic early-juvenile metachromatic leukodystrophy
77	Chaitanya Gadepalli	Salford Mucopolysaccharidosis Airway Score (SMAS): A novel marker of the severity of the airway disease in adult mucopolysaccharidoses
82	Blake Gimbel	Does extending enzyme replacement therapy after transplant provide neurocognitive benefit in Hurler syndrome?
84	Roberto Giugliani	Exploration of the efficacy of pabinafusp-alfa (JR-141) on neurocognitive development in Hunter syndrome (MPS II): 52-week data from clinical trials in Japan and Brazil

85	Roberto Giugliani	Natural history of Sanfilippo syndrome type B in young patients: Ongoing results from two large, prospective studies
93	Paul Harmatz	Design and preliminary results of a first-in-human, 24-week study of intravenous DNL310 (brain-penetrant IDS) in MPS II
95	Caroline Hastings	Update on phase I study to evaluate the single- and multipledose pharmacokinetics of intravenous hydroxypropyl betacyclodextrin (HPβCD) in patients with Niemann-Pick disease type C (NPC-1) and the effects of dosing upon biomarkers of NPC disease
97	Nadene Henderson	Phenocopy of acroparesthesias complicating a Fabry disease diagnosis
98	Benedicte Heron-Longe	The RETRIEVE Study: A natural history study of Gaucher disease type 2, and GM1 and GM2 gangliosidoses with early onset, in preparation of a clinical trial
101	Dafne Horovitz	Need leads to change: Transition to home infusion in Pompe disease in Brazil in the COVID-19 pandemic
102	Derralynn Hughes	Switching between ERT and SRT in patients with Gaucher disease: Data from the Gaucher Outcome Survey (GOS)
109	JoAnn Johnson	Long-term outcomes of patients with mucopolysaccharidosis type VI treated with galsulfase enzyme replacement therapy since infancy
112	Sanjana Kapoor	Hydroxychloroquine use and toxicity in patients with Fabry disease: A case series
116	Aram Kim	Mullen Scales of Early Learning (MSEL) and Bayley Scales of Infant and Toddler Development (BSID): Utility in assessing cognitive endpoints in MPS clinical trials
119	Virginia Kimonis	Safety and effectiveness of resistance training in patients with late-onset Pompe disease: A pilot study
122	Priya Kishnani	Mini-COMET study: Individual participant-level responses to treatment in patients with infantile-onset Pompe disease receiving repeated dose regimens of avalglucosidase alfa or alglucosidase alfa who were previously treated with alglucosidase alfa
133	Dawn Laney	Fabry disease and COVID-19: International expert recommendations for management based on real-world experience
135	Heather Lau	Disease characteristics, early effectiveness, and safety of vestronidase alfa for the treatment of mucopolysaccharidosis type VII (MPS VII) assessed in a novel, longitudinal, multicenter Disease Monitoring Program (DMP)
139	Daniel Lewi	Unravelling the differences between infantile Tay-Sachs and Sandhoff disease using the GM2 Disease Registry (GM2DR)
141	Ales Linhart	Switching from agalsidase alfa to pegunigalsidase alfa to treat patients with Fabry disease: 1 year of treatment data from BRIDGE, a phase 3 open-label study
142	Bianca Link	Orthopedic manifestations in patients with mucopolysaccharidosis type II enrolled in the Hunter Outcome Survey
144	Mabel Lopez	Meaningful endpoints in clinical trials for Infantile Krabbe disease
146	Allan Lund	Evaluation of 2 patients with alpha-mannosidosis and history of conductive hearing impairment participating in a placebo-controlled, phase 3 program receiving velmanase alfa (human recombinant alpha-mannosidase)
147	Allan Lund	Effect of velmanase alfa (human recombinant alpha-mannosidase) enzyme-replacement therapy on quality of life and disease burden of patients with alpha-mannosidosis: Results from caregiver feedback
157	Atul Mehta	Long-term treatment of Gaucher disease with velaglucerase alfa in a cohort of patients from Paraguay
160	Habitha Mohammed Sulaiman	Ten years of Fabry disease in the Northern Irish population
161	Adriana Montano	Growth patterns in subjects with mucopolysaccharidosis type VII
162	Marta Morado	Persistent thrombocytopenia in Gaucher disease (GD): A case report
164	Joseph Muenzer	Comparison of cognitive function in siblings with neuronopathic mucopolysaccharidosis type II: Evaluation of early treatment with intravenous idursulfase and intrathecal idursulfase-IT

165	Joseph Muenzer	Long-term safety and efficacy of intrathecal idursulfase-IT in patients with neuronopathic mucopolysaccharidosis type II: 2-year results from a phase 2/3 extension study
166	Joseph Muenzer	Single-arm, open-label, phase 2/3 substudy and extension evaluating safety and efficacy of intrathecal idursulfase-IT in patients younger than 3 years old with neuronopathic mucopolysaccharidosis type II
168	Nicole Muschol	Tralesinidase alfa (AX 250) enzyme replacement therapy for Sanfilippo syndrome type B
170	Behzad Najafian	Podocyte globotriaosylceramide (GL-3) content declines sharply within 5 months of agalsidase-β enzyme replacement treatment followed by a more gradual decline thereafter
176	Torayuki Okuyama	Drug delivery across the blood-brain barrier and resultant reduction of heparan sulfate in the cerebrospinal fluid in the patients with Hunter syndrome (MPS II): An integrated analysis of 25-week Japanese and Brazilian data on pabinafusp alfa (JR-141)
179	Andrew Oldham	Joint contractures and clonus as main indications for botulinum injections in adult patients with mucopolysaccharidoses: One centre experience
181	Cristol O'Loughlin	Caring for the caregivers: Sensory solutions to CTSD (Chronic Traumatic Stress Disorder)
183	Damara Ortiz	Transition to eliglustat in an individual with Gaucher disease type 1 on antipsychotic medication
191	Marc Patterson	Persistent effect of arimoclomol in patients with Nuemann-Pick disease type C: 12-month results from an open-label extension of a pivotal phase 2/3 study
203	Lynda Polgreen	Open-label clinical trial of anakinra in mucopolysaccharidosis type III: Interim analysis
210	Uma Ramaswami	Migalastat 150 mg every other day achieves bioequivalent exposures in adolescent and adult patients with Fabry disease
212	Shoshana Revel-Vilk	Study design: Development of an advanced machine learning algorithm for the early diagnosis of Gaucher disease using real-world data
214	Barry Rosenbloom	Hematologic malignancies and monoclonal gammopathy of undetermined significance in Gaucher disease type 1 patients in the International Collaborative Gaucher Group Gaucher Registry
216	Anne Rugari	KrabbeConnect patient journey map
217	Emory Ryan	COVID-19 patient impact: A survey of the Gaucher community involving patients, caregivers and family members based in the US to determine impact of the pandemic
219	Siddhee Sahasrabudhe	Modeling potential interactions between oral Gaucher disease treatment and investigational COVID-19 therapies
222	Jessica Scherr	Comparing developmental outcomes of children with CLN2 disease receiving cerliponase alfa to a natural history cohort
223	Raphael Schiffmann	Venglustat combined with imiglucerase positively affects neurological features and brain connectivity in adults with Gaucher disease type 3
224	Angela Schulz	Cerliponase alfa for the treatment of CLN2 disease in a patient cohort including children younger than three years: Interim results from an ongoing clinical study
225	Marisa Schwab	In utero enzyme replacement therapy in fetuses with lysosomal diseases: A phase I clinical trial
230	Ida Schwartz	COVID-19 pandemic impact on Brazilian patients with lysosomal diseases: A patient's perspective
231	Ida Schwartz	Impact of COVID-19 on treatment and follow-up in patients with selected lysosomal diseases in a Brazilian center
246	Karolina Stepien	Long-term outcomes in adult patients affected with fucosidosis: Psychosis as a new complication
247	Karolina Stepien	Cardiac surgical interventions in MPS I and VI patients in adulthood
248	Karolina Stepien	Prevalence of hearing problems in adult mucopolysaccharidosis
261	Amy Vierhile	Neuronal ceroid lipofuscinosis assessment utilizing virtual visits during a pandemic

263	Jesus Villarrubia	PREDIGA project: Preliminary results of the Spanish multicenter epidemiological and medical education project in acid sphingomyelinase deficiency disease (ASMD) and Gaucher disease (GD)
265	Melissa Wasserstein	Adults with chronic acid sphingomyelinase deficiency show significant visceral, pulmonary, and hematologic improvements after enzyme replacement therapy with olipudase-alfa: 1-year results of the ASCEND placebo-controlled trial
266	Michael West	Canadian Fabry disease registry study group: Report on the A143P Nova Scotia genotype
267	Michael West	Lyso Gb3 and Gb3 analogues in Fabry disease patients with A143P genotype: A cross-sectional analysis by the CFDR study group
270	Matheus Wilke	Informing patients with rare diseases about COVID-19: Creation of the “Beto and the Coronavirus” booklet
274	Puriya Daniel Yazdanfard	Hearing loss in Fabry disease: A 16 year follow-up study of the Danish nationwide cohort

Thursday, February 11 – ePoster Presentations

5	Thomas Anderson	Pharmacokinetics properties of arimoclomol in Niemann-Pick disease type C: Modest and not clinically relevant effect of bodyweight or age
6	Allyson Anding	Miglustat does not enhance alglucosidase alfa or avalglucosidase alfa efficacy in Pompe mice
15	Eileen Baranowski	The qualitative development of the Pompe disease symptom scale and Pompe disease impact scale
21	Akhil Bhalla	Evaluation of fluid biomarkers reveals lysosome dysfunction and neurodegeneration in neuronopathic MPS II patients
22	Rachel Botham	Engineering α-glucosidase to improve protein stability and cellular uptake for the potential treatment of Pompe disease
34	Yoonjin Cho	Clinical and numerical presentation of neurocognitive assessments for MPS II patients using the Bayley Scales of Infant Development- version 3 (BSID-III)
41	Fabrizio Comper	Generation of β-Glucocerebrosidase variants with increased half-life in human plasma for liver directed AAV gene therapy aimed at the treatment of Gaucher disease type 1
43	Fernanda Copeland	Uncovering the burden of Gaucher disease type 1: Patient perspectives on unaddressed symptoms, impact of disease, and the future of treatment
44	Romuald Corbau	FLT201: An AAV-mediated gene therapy for type 1 Gaucher disease designed to target difficult to reach tissues
49	Magdalena Daurer	Evaluation of CD45 positive cells in the brain and liver of NPC1-/- mice
50	Elin Haf Davies	Decentralised convenience: Digitised clinical assessment for impaired ambulation
51	Elin Haf Davies	Assessing paediatric feeding in lysosomal diseases using remote smartphone video technology
60	Marissa Donovan	SIG-005: Novel encapsulated non-viral cell-based therapy for MPS I
64	Florian Eichler	Initial signs and symptoms of metachromatic leukodystrophy: A caregiver perspective
67	Deborah Elstein	Psychometric validation of the Gaucher Disease Questionnaire (GDQ) to assess quality of life in patients with Gaucher disease
73	Brian Fluharty	SIG-007: Novel encapsulated non-viral cell-based therapy for Fabry disease
83	Jacinthe Gingras	HMI-202: A gene therapy development candidate for metachromatic leukodystrophy (MLD)
90	Lavesh Gwalani	Invariant natural killer T cell-mediated cytokine secretion is a potential biomarker to monitor the efficacy of treatment for Fabry disease
94	Hidehiko Hashimoto	Usefulness of hexose tetrasaccharide as a biomarker for monitoring glycogen accumulation in peripheral tissues and brain in Pompe disease
99	Ryo Higurashi	Phase I/II clinical trial design for a novel therapy for mucopolysaccharidosis type I with an intravenously administered blood-brain barrier-crossing enzyme (JR-171)
100	Max Hilz	Burden of illness of Fabry disease: A retrospective claims analysis of a German sickness fund database
103	Atsushi Imakiire	Non-clinical evaluation of a blood-brain barrier-penetrable a-N-acetylglucosaminidase in a mouse model of mucopolysaccharidosis type IIIB
106	Leslie Jacobsen	The GuardOne clinical trial: A first-in-human, open-label, multinational phase 1/2 study of AVR-RD-02 ex vivo lentiviral vector, autologous gene therapy for Gaucher disease
108	Franklin Johnson	Migalastat clinical dose is highly extracted by hemodialysis and hemodiafiltration
110	Simon Jones	Demographic and clinical characteristics of patients with metachromatic leukodystrophy in the United Kingdom: Interim results from an observational real-world study
123	Jennifer Klein	The Mucolipidosis Collaborative Research Network (MCRN)
129	Ian Kurashige	Enzyme replacement therapy treatment patterns and patient outcomes in late-onset Pompe disease

130	Kristin LaBounty Phillips	Co-creating a gene therapy clinical trial with GM2 gangliosidosis caregivers: A virtual approach to patient engagement
137	Daniel Lewi	Aparito's six year journey in lysosomal disorders
138	Daniel Lewi	The new normal: Smartphone technology and its impact on the logistics of healthcare
143	Tina Loeffler	Characterization of 4L/PS-NA mice for cytokine activity and neurodegeneration
151	Joe Marion	A disease progression model for trials in mucopolysaccharidosis type IIIA
152	Deborah Marsden	Significant unmet need in infants with mucopolysaccharidosis type VII and non-immune hydrops fetalis: A summary of cases
158	Eugen Mengel	Impacts and burden of Niemann-Pick disease type C: A patient and caregiver perspective
171	Marie-Laure Nevoret	RGX-121 gene therapy for severe mucopolysaccharidosis type II (MPS II): Interim results of an ongoing first in human trial
186	Francis Pang	Quality of life of patients with metachromatic leukodystrophy and their caregivers in the US, UK, Germany and France
187	Francis Pang	Health-related quality of life in metachromatic leukodystrophy based on a societal utility study in the UK
188	Tiffany Pang	Clinical utility of a sponsored gene panel testing program for pediatric epilepsy and CLN2 disease diagnosis: Results from 4246 tests
189	Kruti Patel	HMI-203: Investigational gene therapy for mucopolysaccharidosis type II (MPS II), or Hunter syndrome
190	Marc Patterson	Validation of a short-form 5-domain Niemann-Pick type C clinical severity scale (5-domain NPCCSS)
192	Erika Pearson	Development of a novel encapsulated non-viral cell-based therapy for MPS VI
196	M. Judith Peterschmitt	Oral venglustat in Parkinson disease patients with a GBA mutation: Study design of part 2 of the MOVES-PD trial and patient characteristics
197	Nikolaj Petersen	Rescue of NPC1 protein by the heat shock response amplifier arimoclomol across multiple genotypes
198	Dawn Phillips	Characterizing expressive language skills in children with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2): The caregiver perspective
199	Dawn Phillips	Characterizing visual function in children with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2): The caregiver perspective
211	Sujata Ravi	Development of a GLA nAb assay with a fully-human, neutralizing IgG4 positive control to characterize antibody response in Fabry disease patients
213	Camille Rochmann	A qualitative study of the experience of venglustat for patients with Gaucher disease type 3 (GD3) in LEAP: A phase II open-label, multicenter, multinational study
232	Guillermo Seratti	Clinical utility of a sponsored, no-cost skeletal dysplasia gene panel testing program: Results from 850 tests
233	Irene Serrano Gonzalo	New insights in the TRAZELGA project for the adult type 1 Gaucher disease patients treated with eliglustat follow-up
238	Akashdeep Singh	Diagnostic yield and clinical utility of genetic testing in children with seizure onset after two years of age: Update over 2 1/2-year program in Europe and the Middle East
239	Akashdeep Singh	Molecular basis of mucopolysaccharidosis type IVA (Morquio syndrome type A): A review and classification of GALNS gene variants and reporting of new variants
242	Sairei So	Therapy for mucopolysaccharidosis type II with an intravenous blood-brain barrier-crossing enzyme (JR-141): Phase III global clinical trial design
252	Satowa Tanaka	Non-clinical evaluation of a blood-brain barrier-penetrable N-sulfoglucosamine sulfohydrolase in a mouse model of mucopolysaccharidosis type IIIA
254	Drew Tietz	SIG-018: Novel encapsulated non-viral cell-based therapy for MPS II
272	Tobias Willer	Development of a novel gene therapy for Fabry disease: Engineered alpha-galactosidase A transgene for improved stability

276	Karen Yee	Assessment of cognitive development in patients with neuronopathic mucopolysaccharidosis type II treated with intrathecal idursulfase-IT using Projected Retained Ability Score (PRAS): A post hoc analysis
277	Karen Yee	Caregiver experiences of intrathecal idursulfase-IT treatment in pediatric patients with neuronopathic mucopolysaccharidosis type II

Friday, February 12 – ePoster Presentations

121	Priya Kishnani	Efficacy and safety results of the avalglucosidase alfa phase 3 COMET trial in late-onset Pompe disease patients
LB-02	Elizabeth Berry-Kravis	Evidence for long-term efficacy of intrathecal adrabetadex for the treatment of neurological decline in patients with Niemann-Pick disease, type C1
LB-03	Shabnam Bhuiyan	Impact of COVID-19 on lysosomal disease patients
LB-05	Fabrice Camou	Gaucher disease: Final results of ELIglustat Patient Reported Outcomes (ELIPRO), a prospective and multicentric study
LB-06	Pierre Carlier	NEO1/NEO-EXT studies: Muscle MRI results in patients with Pompe disease after long-term avalglucosidase alfa treatment
LB-07	Jennifer Clarke Matthews	Murine models of lysosomal diseases exhibit differences in brain protein aggregation and neuroinflammation
LB-08	Tanya Collin-Histed	Development of quality of life measures in neuronopathic Gaucher disease: Qualitative research findings
LB-09	Maria de Castro	Updated results of Transpher B, a multicenter, single-dose, phase 1/2 clinical trial of ABO-101 gene therapy for Sanfilippo syndrome type B (MPS IIIB)
LB-10	Emily de los Reyes	An open-label, phase 1/2a, AAV9-CLN3 gene transfer clinical trial for juvenile neuronal ceroid lipofuscinosis
LB-11	Claudia Cozma	Lyso-sphingolipid biomarker profiles of PSAP-associated disorders
LB-12	Vivian Fernandez	New research examines the evolution of data sharing practices in natural history studies and patient registries among patient advocate leaders, industry and academia
LB-13	Veronica Ferrari	VCP mutants induce lysosomal damage and autophagy activation in amyotrophic lateral sclerosis (ALS)
LB-14	Claudia Cozma	Building a human metabolome reference with applications in discovering new biomarkers and affected pathways in lysosomal diseases
LB-16	Takahiro Fujii	Discovery & characterization of a novel brain-penetrant glucosylceramide synthase inhibitor as a potential therapeutic for Gaucher disease
LB-17	Ana García-Collazo	Preclinical development of brain-penetrant structurally targeted allosteric regulators for the treatment of neuronopathic Gaucher disease
LB-18	Robert Gotschall	Mucolipidosis type II AAV9 gene therapy pilot study: In vivo safety of over-expressing modified GlcNAc-1-phosphotransferase (S1S3) in wild-type mice
LB-19	Jerry Harb	Weekly enzyme replace therapy reverses hypertrophic cardiomyopathy in two Pompe knock-in murine models
LB-21	Justin Hopkin	Understanding health insurance literacy and challenges in accessing health services in Niemann-Pick disease
LB-22	Claudia Cozma	Methods of monitoring of neuropathic Gaucher disease in CSF
LB-23	Margarita Ivanova	TRAP5b and RANKL/OPG predict bone pathology in patients with Gaucher disease
LB-24	Alyssa Johnson	SVIP is a molecular determinant of lysosomal dynamic stability, neurodegeneration and lifespan
LB-25	Adam Kanack	Prothrombotic phenotypes in a rat model of Fabry disease
LB-26	Jin Young Kim	Efficacy of a novel long-acting arylsulfatase B analog (HM15450) with subcutaneous administration in mucopolysaccharidosis type VI (MPS VI) mouse model
LB-27	Dawn Laney	Pregnancy outcomes in agalsidase beta-treated and untreated females with Fabry disease and their offspring: A Fabry pregnancy sub-registry and pharmacovigilance database 15-year retrospective study
LB-28	Heather Lau	Long-term renal efficacy and incidence of Fabry-associated clinical events in treatment-naive and enzyme replacement therapy-experienced female patients receiving migalastat for Fabry disease up to 8.5 years
LB-29	Lucia Lavalle	Chloroquine differential impact on lysosomal hydrolases

LB-30	Lin Liu	Phosphorylated acid beta-glucosidase (M011, GCaseM6P) enzyme replacement therapy, leads to better tissue distribution, cellular uptake and efficacy in the GaucherD409A mouse model compared to conventional alpha-mannosyl terminated acid beta-glucosidase
LB-31	Anna Luzzi	Immunophenotyping of patients with Sanfilippo syndrome reveals a deficiency in natural regulatory t cells that is reversed with anakinra
LB-33	Benedicta Marshall-Andrew	Maintaining access to clinical trials during the COVID-19 pandemic
LB-34	Toni Mathieson	Patient and family perspective of lysosomal disease clinical care and services in the UK
LB-35	Alexandra Morrison	Diagnosis of mucopolysaccharidoses in the UK
LB-36	Juana Navarrete	Use of biomarkers to follow up positive lysosomal diseases in newborn screening
LB-37	Alberto Ortiz	Stabilization of kidney function decline and cardiomyopathy in male patients with classic Fabry disease: A pre- vs. post-agalsidase beta treatment Fabry Registry analysis
LB-38	Saida Ortolano	Systemic treatment of Fabry disease using a novel AAV9 vector expressing α-Galactosidase A
LB-39	Cho Rong Park	Efficacy of a novel long-acting alpha-galactosidase A analog (HM15421) with subcutaneous administration in Fabry disease mouse model
LB-40	Natalia Pérez-Carmona	Insights into the mechanism of action of structurally targeted allosteric regulators for the treatment of Gaucher disease
LB-41	Eva Raebel	Understanding challenges for ultra-rare lysosomal diseases: Patient and caregiver experience of care and support through the disease journey
LB-42	Tiziana Rosa	Insights into the mechanism of action of structurally targeted allosteric regulators for the treatment of GLB1-related disorders
LB-43	Yuki Shiro	Differential impairment of CLN6's anti-aggregate activity as a pathogenic mechanism of CLN6 disease
LB-44	Dylan Simon	Bringing together all newborn screening stakeholders
LB-45	Dylan Simon	Working to ensure that every state screens for all RUSP conditions
LB-46	Volha Skrahina	Towards defining the dynamics of Lyso-Gb1 accumulation in Gaucher patients
LB-47	Miles Smith	Ex vivo lentiviral transduction of hematopoietic stem cells in mucopolysaccharidosis type II (MPS II) mice achieves high levels of systemic iduronate-2-sulfatase (IDS) enzyme activity and normalization of glycosaminoglycans (GAGs)
LB-48	Marie Szymanowski	First case of neutralizing anti-ERT (enzyme replacement therapy) antibodies in Gaucher disease (GD) in a 9-year-old child
LB-49	Daisy Tapia	Prevalence of small vessel disease in a Fabry disease cohort
LB-50	Kazuyoshi Tomita	A comparison of developmental trajectories in sibling cases with neuropathic MPS II receiving conventional and novel enzyme replacement therapies
LB-51	Christoph Wanner	Reduced delays in diagnosis of patients with Fabry disease over time: A Fabry Registry analysis of data (1985-2020) stratified by gender and phenotype