Poster Session Abstracts

Poster sessions will be in the Regency Ballroom R on:
Tuesday, February 5 from 4:30-6:30pm
Wednesday, February 6 from 4:30-6:00pm.

Poster presenters with a last name starting with A-L (First Author Last Name) will be assigned to present their poster on Tuesday, February 5, 2019 from 4:30-6:30 PM. Posters must be mounted by 4:00 PM on Tuesday and will need to be removed after the Tuesday poster session ends.

Poster presenters with a last name starting with M-Z (First Author Last Name) will be assigned to present their poster on Wednesday, February 6, 2019 from 4:30-6:30 PM. Posters must be mounted by 4:00 PM on Wednesday and will need to be removed after the poster session ends.

All late-breaking abstracts will be assigned to the poster session on Wednesday, February 6, 2019 from 4:30-6:30 PM. Posters must be mounted by 4:00 PM on Wednesday and will need to be removed after the poster session ends.

*Attendees may take photos of posters **ONLY** if the poster author agrees. Authors who do not want their posters to be photographed will have to indicate as such on their posters. No other photography, or audio or video recording is allowed. Attendees who photograph or record poster information for which they have not obtained permission will be asked to leave the session immediately.*

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.

Tuesday, February 5 – Poster Presentations

1	Ibane Abasolo	Targeted nanoliposomes for the treatment of Fabry disease
2	Magy Abdelwahab	Characterization of epilepsy in a large Egyptian type 3 Gaucher disease (GD3) cohort: A 12-year prospective study
3	Alyssa Aburachis	Natural history of Sanfilippo syndrome
4	Alyssa Aburachis	Staging of Sanfilippo syndrome type A
5	Jacqueline Adam	Understanding Fabry in families: Preliminary findings from a global survey
6	Jacqueline Adam	Impact of two-year elosulfase alfa treatment on patient-reported outcomes in patients with Morquio syndrome type A: Results from an English managed access agreement
7	Jacqueline Adam	Patient reported outcomes in MPS IVA patients receiving enzyme replacement therapy
10	Patricio Aguiar	Plasma lyso-Gb3 in Fabry disease: Helpful distinguishing phenotypes, but not as predictor of organ involvement
11	Patricio Aguiar	MicroRNAs in Fabry disease: Distinguishing between phenotypes and correlations with organ involvement
13	Sujin Ahn	Development of a novel glucosylceramide synthase (GCS) inhibitor with increased blood-brain barrier penetration for treatment of Gaucher disease
15	Carlos Almeciga-Diaz	Chondrocytes and cardiomyocytes derived from Morquio syndrome type A induced pluripotent stem cells (iPCS)
16	Marcio Andrade-Campos	Twenty-five years diagnosing Gaucher disease in Spain: What we have learned?
17	Marcio Andrade-Campos	Prospective multi-center national study to standardize the follow-up of type 1 Gaucher disease patients treated with eliglustat under standard of care practice: TRAZELGA project
18	Kara Anstett	Bone density and treatment response in a large cohort of patients with type 1 Gaucher disease
19	Kara Anstett	Non-biliary gastrointestinal symptoms as the initial presenting symptom of type 1 Gaucher disease: A case series

20	Mathilda Antonini	The changing role of the clinical nurse specialist for lysosomal diseases: Suggestions for support in the role
22	Dustin Armstrong	A novel antibody-enzyme fusion (AEF) platform for treating glycogen storage disorders
23	Rhea Ashmead	Crossing biological membranes using PTD4: Implications for treatment of MPS IIIB through enzyme replacement therapy
24	Helen Ashton	One centres experience of sedation regimes for ICV and IT drug delivery in early phase pediatric clinical trials
25	Annalisa Astolfi	Hippo and necroptosis pathways are involved in cell growth defects in Gaucher disease
26	Nora Atanacio	Clinical, biochemical and molecular characteristics of five patients with late infantile neuronal ceroide lipofucsinosis type (CLN2 disease) phenotype clasical and atypical
27	Christiane Auray-Blais	Newborn mass urine screening for Morquio syndrome type A patients using an innovative UPLC-MS/MS approach
28	Stephanie Austin	Early-onset of symptoms and clinical course of Pompe disease associated with the c.-32-13T>G variant
29	Rachel Bailey	Development of scAAV9/SUMF1 gene therapy for multiple sulfatase deficiency
30	Manisha Balwani	Clinical manifestations of LAL-D: The international lysosomal acid lipase deficiency registry
31	Manisha Balwani	Outcomes of 19 unplanned pregnancies in women participating in phase 2 or 3 eliglustat clinical trials and 18 pregnancies in the partners of men who participated in these trials
32	Laura Barisoni	Migalastat reduces globotriaosylceramide (GL-3) inclusions in renal peritubular capillaries in patients with Fabry disease and migalastat-amenable mutations: Post hoc analyses from FACETS
33	Suelen Basgalupp	Is there any difference in GBA1 allele frequencies depending on the region of Brazil?
34	Luisa Bay	Head circumference in individuals with MPS I compared to CDC standard charts
35	Brendan Beaton	Soluble mannose receptor is a potential new biomarker for Gaucher disease
37	Michal Becker- Cohen	Retinal thinning in Gaucher patients as a predictive test of developing Parkinson disease
38	David Bedwell	Triamterene normalizes glycosaminoglycan accumulation in an IDUA-W402X mouse model of MPS I (Hurler syndrome) via nonsense suppression
39	Soumeya Bekri	Integrative metabolic profiling in Sanfilippo syndrome
40	Soumeya Bekri	Next generation sequencing sheds light on inherited metabolic diseases in nonimmune hydrops fetalis investigations
41	Maria Beltran-Quintero	Nerve conduction studies as a tool in early detection of metachromatic leukodystrophy
42	Daniel Bichet	Effect of long-term migalastat treatment on plasma globotriaosylsphingosine (lyso-Gb3) levels in patients with Fabry disease previously treated with enzyme replacement therapy: Results from ATTRACT and open-label extension studies
44	Ruben Boado	Platform technology for treatment of the brain in lysosomal disorders: Application to Tay-Sachs disease
45	Pedro Paulo Bozzo	Against all odds: enzyme replacement therapy in non-ambulatory and ambulatory Morquio syndrome type A patients
46	Colm Bradley	A new research initiative amongst hematologists to address current worldwide health disparities in the management and treatment of Gaucher disease
47	Elizabeth Braunlin	Cardiopulmonary findings with enzyme replacement therapy after hematopoietic cell transplantation for MPS VI
49	Elizabeth Braunlin	Consequences of newborn screening: Neuroimaging in infants with severe MPS I ≤6 months of age
50	Anders Bröijersén	Safety and tolerability of SOBI003 in pediatric MPS IIIA patients - Key study design features of the ongoing first-in-human study

52	Or Cabasso	The fruit fly Drosophila melanogaster as a model to study Gaucher disease
53	Umut Cagin	Functional, biochemical and transcriptional rescue of advanced Pompe disease in mice with liver expression of secretable GAA
54	Jacob Cain	Identifying a biomarker signature for Batten disease
55	Raíssa Caldeira	β-glucocerebrosidase activity is low in patients with multiple myeloma
56	Daniela Castillo-García	Mucopolysaccharidosis type II (Hunter syndrome) with multisystem Langerhans cell histiocytosis - A case report of a not described association
57	J Cebolla	Assessment of plasma 7-ketocholesterol concentration, chitotriosidase activity and CCL18/PARC concentration in Spanish patients treated with human recombinant lisosomal acid lipase
58	Magdalena Cerón-Rodríguez	Identification of a novel GLAmutation (V269L) in a Mexican 2 year old male with Fabry nephropathy: A case report
59	Anuj Chauhan	Potential role of stromal collagen in cystine crystallization in cystinosis patients
61	Huma Cheema	Clinical characteristics, genotype and outcome of Gaucher disease in Pakistani children
64	Tsui-Fen Chou	Enzyme replacement therapy for mucopolysaccharidosis type IIID
66	Wei-Lien Chuang	Development and validation of a novel multiplex LC-MS/MS assay of globotriaosylceramide and globotriaosylsphingosine in human plasma
67	Maureen Cleary	ICV-administered tralesinidase alfa (BMN 250; NAGLU-IGF2) is well-tolerated and reduces heparan sulfate accumulation in the CNS of subjects with Sanfilippo syndrome type B (MPS IIIB)
68	Paula Clemens	Safety and efficacy of AT-GAA (ATB200/AT2221) in ERT-switch non-ambulatory patients with Pompe disease: Preliminary results from the ATB200-02 trial
69	Pasqualina Colella	Tandem promoter design confers tolerogenic and persistent transgene expression to AAV gene therapy in neonate Pompe mice
71	Therese Conner	Healthcare resource use in severe mucopolysaccharidosis type I post-transplant children via parent survey
72	Therese Conner	Results of an online survey on family burden of illness in severe mucopolysaccharidosis type II
73	Therese Conner	Results of a Canadian survey on the family burden of illness in severe mucopolysaccharidosis type I
74	Maria Julia Costa	Clinical and biochemical study of Brazilian patients with metachromatic leukodystrophy
75	Timothy Cox	Effects of oral eliglustat on skeletal manifestations in patients with type 1 Gaucher disease: Results from four completed clinical trials after long-term treatment
76	Andrea Crivaro	Osteoblast and adipose differentiation of Gaucher mesenchymal stem cells
77	Vania D Almeida	Prevalence of mucopolysaccharidoses in samples sent to the laboratory of inborn errors of metabolism, Sao Paulo, Brazil
78	Vania D Almeida	Differential diagnosis for mucopolysaccharidoses: Evaluation of β-glucuronidase activity
79	Vania D Almeida	6-sulfatoxymelatonin daily profile in Fabry disease patients: Relationship to disease variants
80	Amanda Daniel	Nurse-led clinics for lysosomal storage unit disorders (LSDU) are we prepared? A scope of advanced nursing practice in the UK centres
81	Julia Dao	Evaluation of disease burden and therapy modifications using glucosylsphingosine (lyso-GL1) in Gaucher disease
82	Pronabesh DasMahapatra	Evaluation of daily activity patterns using a wearable device in Pompe disease
83	Pronabesh DasMahapatra	Agalsidase beta delays the progression to kidney disease in Fabry patients: Results from an individual patient data meta-analysis
84	Cristin Davidson	Improved disease amelioration with combination therapy for Niemann-Pick type C1 disease

85	James Davison	Carpal tunnel syndrome in mucopolysaccharidosis type I Hurler-Scheie/ Scheie and effect of enzyme replacement therapy
86	James Davison	Hypogammaglobulinemia, impaired vaccine response and recurrent infections in mucolipidosis type II
87	Patrick Deegan	A composite fracture risk score for assessing adult fracture risk in imiglucerase-treated type 1 Gaucher disease patients using data from the International Collaborative Gaucher Group (ICGG) Gaucher Registry
88	Francisco del Castillo	NGS-based, 107-gene resequencing panel as first-line screening test for lysosomal diseases
89	Mireia del Toro	Severe cardiac involvement: Management in a homozygous D409H Gaucher patient under enzyme replacement therapy
91	Jordi Díaz-Manera	Quantitative muscle MRI in Pompe disease: A 4 years follow-up study
93	Jenny Do	A 3'-UTR variant in SCARB2 modulates LIMP2 in patients with Gaucher disease and myoclonic epilepsy
94	Aimee Donald	From birth to the sixth decade - A natural history study of 42 patients with neuronopathic Gaucher disease
95	Theodore Drivas	Identification of lysosomal diseases by expanded carrier screening
96	Katie Duke	RVT-801, a developmental enzyme replacement therapy for Farber disease, ameliorates characteristic features of the disease phenotype in a Farber mouse model
97	Consuelo Durand	Mucopolysaccharidosis type VII: Clinical and biochemical data of 8 patients from Argentina
98	Hatim Ebrahim	Renal involvement in classical and late onset patients with Fabry disease and the role of co-existing pathologies
99	Areian Eghbali	How do we explain very discordant phenotypes among three siblings with neuronopathic Gaucher disease? Whole exome sequencing and transcriptome analyses
100	Farah El Turk	Lipidomics in translational research and clinical relevance for the identification of biological fluids sphingolipids biomarkers for muccopolysaccharidoses
101	Deborah Elstein	Gaucher disease (GD)-specific patient-reported outcome (PRO) measures for clinical monitoring and for clinical trials
102	Kaoru Eto	The correlation between brain MRI imaging and biochemical and molecular findings in Japanese female patients with Fabry disease
103	Francois Eyskens	Multiple sclerosis as a misdiagnosis of Fabry disease
104	Ulla Feldt-Rasmussen	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 30-month results from the randomized phase 3 ATTRACT study
105	Sergio Figueroa Sauceda	Prevalence of Fabry disease in the hemodialysis unit of the Instituto Mexicano del Seguro Social, in Ciudad Obregon, Sonora, Mexico
107	Lauren Flueckinger	Evolving challenges in the era of newborn screening for Pompe disease
108	Lauren Flueckinger	Corticobasal syndrome in a man with type 1 Gaucher disease: Expansion of the understanding of the neurological spectrum
109	Stuart Forshaw-hulme	Self-management using wearable technology to promote patients' knowledge and skills in managing their own care
110	Stuart Forshaw-Hulme	The use of Fitbit data in monitoring the improved functioning and quality of life in a case of Fabry disease
111	Stuart Forshaw-hulme	The effectiveness of vein mapping in reducing the number of missed infusions among Fabry disease patients: One centre experience
112	Joaquin Frabasil	Mucopolysaccharidosis type VII: Selective retrospective screening detects 4 new cases
113	Omar Francone	Single intravenous dose of AAVHSC15 vector with human phenylalanine hydroxylase transgene results in sustained correction of phenylketonuria in the PAHenu2 mouse model

114	Mahoko Furujo	Safety evaluation of pentosan polysulfate for treatment of two Japanese siblings with mucopolysaccharidosis type VI in a phase 2 study
115	Eric Joshua Garcia	Methylomic and whole transcriptome analyses reveal several potential modifier genes in GBA1-associated Parkinson disease
116	Jose Garcia	Biochemical and molecular analysis of MPS III in 6 Mexican patients
117	Jose Garcia Fernandez	Tailoring the inhibitory versus chaperoning behavior of amphiphilic sp2-iminosugar glycomimetics targeting β-glucocerebrosidase: From micromolar to picomolar chaperones for Gaucher disease
118	Jose Garcia Fernandez	Screening sp2-iminosugar *N*-glycosides as pharmacological chaperone candidates for α mannosidosis: The effect of aglycone nature and valency
119	Michael Gelb	Tandem mass spectrometry of 15 lysosomal diseases, biotinidase deficiency and galactosemia type 1 at 2.4 minutes per assay
120	Kelly George	Biomarker and pathway analysis in Pompe disease
121	Arunabha Ghosh	Membranous nephropathy in a patient with infantile onset lysosomal acid lipase deficiency and anti-sebelipase antibodies
122	Arunabha Ghosh	High dose genistein aglycone in Sanfilippo syndrome: Results of a randomized, double-blinded, placebo controlled clinical trial
123	Pilar Giraldo	Strain-elastography in musculoskeletal evaluation in Gaucher disease
124	Pilar Giraldo	Localized lymphedema in a male with classic Fabry disease
125	Roberto Giugliani	The MPS I Registry - 15 years of service to the community
127	Stella Godinho	Hurler syndrome: severe sleep apnea as initial presentation in a 10-month-old child
128	Jorge Francisco Gomez Cerezo	Incidence of Fabry disease in patients with angiokeratoma
129	Kevin Goncalves	MGTA-456, a first-in-class cell therapy that enables a reduced intensity conditioning regimen and enhances speed and level of human microglia engraftment in the brains of NSG mice
130	Sofia Goncalves	Pitfalls and potential clues in diagnosis in attenuated form of Hunter syndrome
131	Antonio González-Meneses	Spine instability in patients with mucopolysaccharidosis (MPS) type VII
132	Janet Gorton	Adherence to a pharmacological chaperone therapy among patients with Fabry disease: One centre experience
134	Diane Green	A review of gastrointestinal symptoms among patients affected with Fabry disease-One centre experience
135	Giuseppina Grillo	Pharmacological chaperone therapy using migalastat: 1 year experience of starting a new therapy as reported by Fabry patients at a single UK centre
137	Nathalie Guffon	The first study investigating safety and efficacy of velmanase alfa (human recombinant alpha mannosidase) in alpha-mannosidosis patients below six years of age
138	Ersin Gumus	Long-term visceral and hematologic outcomes of enzyme replacement therapy in a pediatric cohort of type 1 and type 3 Gaucher disease: A single center experience
139	Ersin Gumus	Type 3 Gaucher disease presented with cardiac manifestations
141	Punita Gupta	Detecting a variant in the GLA gene in multiple family members as an incidental finding
142	Stephanie Gurnon	Understanding Sanfilippo syndrome signs, symptoms and physician testing patterns: Insights from the Simply Test for MPS™ enzyme-panel program (ST4MPS)
143	Alaa Hamed	Measurement properties of the Fabry Disease Patient Reported Outcome (FD-PRO), a new instrument to measure symptoms in Fabry disease
144	Sang-oh Han	Salmeterol with liver depot gene enhances the skeletal muscle response in murine Pompe disease
145	Rosenbaum Hanna	Fatigue in Gaucher disease: A key quality-of-life concern
146	Paul Harmatz	Enzyme replacement therapy in patients with mucopolysaccharidosis type VI: Updated findings from the MPS VI clinical surveillance program

148	Katie Harvey	The evolving role of enzymology and metabolomics in the diagnosis of lysosomal disorders in the post genomic era
149	Kimberly Hawkins	Effects of ketogenic diet on lysosomal storage and CNS metabolism in MPS IIIB mice
150	Simon Heales	High α-galactosidase A over expression and/or mitochondrial dysfunction may inhibit efficacy of gene therapy for Fabry disease
151	Simon Heales	Urinary glucose tetrasaccharide, a useful prognostic biomarker for Pompe disease?
152	Garrett Heffner	Continued analysis of GAA -/- mice treated with novel hybrid promoter rAAV vectors expressing acid alpha-glucosidase
153	Nadene Henderson	Increased frequency of enzyme replacement therapy in a Fabry disease cohort
154	Christian Hendriksz	Methodology to develop guidelines for the management of patients with neuronal ceroid lipofuscinosis type 2 disease
155	Christian Hendriksz	Evidence-based, expert-agreed recommendations for the management of patients with MPS IVA/VI: Recommendations to replace the specific missing enzyme
156	Julia Hennermann	The SPARKLE study: Shedding light on alpha mannosidosis
157	Julia Hennermann	Retina and optic nerve degeneration in alpha-mannosidosis
158	Anastasia Henry	Improved brain uptake and efficacy of iduronate 2-sulfatase with the enzyme transport vehicle
159	Aki Hietaharju	Screening for Fabry disease and hereditary ATTR amyloidosis in idiopathic small fiber and mixed neuropathy
160	Myrl Holida	Once every 4 weeks - 2 mg/kg of pegunigalsidase alfa for treating Fabry disease; Preliminary results of a phase 3 study
161	Robert Hopkin	Renal and cardiac outcomes of young male patients with Fabry disease initiated on agalsidase beta treatment before age 30: A Fabry registry analysis
162	Robert Hopkin	Significant abdominal and acute pain improvements in young patients with Fabry disease initiated on agalsidase beta treatment before age 30: A Fabry registry analysis
163	Dafne Horovitz	Bone/joint abnormalities in children/adolescents: A screening protocol for mucopolysaccharidosis
164	Dafne Horovitz	Enzyme replacement therapy in mucopolysaccharidosis type II with alternative dosing 1mg/kg idursulfase in every other week infusions
165	Dafne Horovitz	Mucopolysaccharidoses and laryngeal, tracheal and bronchial disease: Type-specific abnormalities and long-term implications
166	Mohammad Hossain	Evaluation of long-term effects by ERT for Fabry disease biochemical and EM pictures
167	Jeffrey Huang	CRISPR-Cas9 generated Pompe knock-in murine model exhibits early-onset cardiac hypertrophy and motor impairment
168	Derralynn Hughes	A global consensus on early indicators of organ damage in Fabry disease and implications for treatment initiation
169	Derralynn Hughes	Clinical features of Fabry disease in patients with mutations amenable and non-amenable to migalastat
171	Marshall Huston	Liver-targeted AAV gene therapy vectors produced by a clinical scale manufacturing process result in high, continuous therapeutic levels of enzyme activity and effective substrate reduction in mouse model of Fabry disease
172	Jackie Imrie	Challenges of regulatory requirements for patient registries in different countries
173	Rina Itagaki	Neuronal ceroid lipofuscinosis (NCL) types 1 and 2: Enzyme characteristics of PPT1 and TPP1, and their high risk and newborn screenings
175	Margarita Ivanova	Effects of small molecule therapies on lysosomal function in Gaucher disease
176	Siamak Jabbarzadeh-Tabrizi	Effects of genetic background on disease phenotypes in a mouse model of Fabry disease
177	Juliette Janson	Distribution of chemically modified rhSulfamidase to CNS monitored by brain microdialysis and repeated CSF sampling after intravenous administration in rat

178	Jeanine Jarnes-Utz	Adding enzyme replacement therapy after hematopoietic stem cell transplantation results in increased metabolic correction in MPS VI
179	Susheela Jayaraman	Fabry disease A143T genotype-phenotype investigation
180	Jey Jeyakumar	Liver-directed gene therapy corrects Fabry disease in mice
181	Franklin Johnson	Migalastat pharmacokinetic (PK) exposure comparisons between race/ethnic groups and between males and females are similar
182	Tyler Johnson	Characterization of a novel porcine model of CLN3-Batten disease
183	Kofler Julia	Clinical and neuropathologic findings in two long-term survivors of Krabbe disease with and without umbilical cord blood transplantation
184	Ilkka Kantola	Enzyme replacement therapy together with renin-angiotensin system inhibition seems to prevent kidney function decrease in most Finnish Fabry patients treated either for 5 or 10 years
186	Scott Kerns	An improved, novel, systemically administered AAV gene therapy for treatment of CLN3 juvenile neuronal ceroid lipofuscinosis
187	Brian Kevany	AAV gene therapy for the treatment of Fabry disease: A novel capsid with improved tropism to heart, kidney and CNS and improved GLA expression
188	Brian Kevany	A novel AAV capsid with improved CNS tropism for treating Pompe disease by intravenous administration
189	Sachiho Kida	Non-clinical evaluation of a blood-brain barrier-penetrating enzyme for the treatment of mucopolysaccharidosis type I
190	Gee-Hee Kim	A case of a 39-year-old man with novel mutation and classic Fabry disease who showed different changes of several biomarkers and speckle tracking after enzyme replacement therapy
191	Virginia Kimonis	Effects of enzyme replacement therapy on bone density in late onset Pompe disease
192	Virginia Kimonis	Safety and effectiveness of resistance exercise training in a pilot study of patients with late onset Pompe disease
194	Kelly King	Feasibility of quantifying behavior in early progressive MPS II
195	Priya Kishnani	Safety and efficacy of VAL-1221, a novel fusion protein targeting cytoplasmic glycogen, in patients with late-onset Pompe disease
196	Priya Kishnani	First-in-human study of AT-GAA (ATB200/AT2221) in patients with Pompe disease: Preliminary functional assessment results from the ATB200-02 trial
197	Jennifer Klein	Data mining and machine learning for lysosomal disease drug discovery and beyond
198	Aditi Korlimarla	Quantitative evaluation of white matter hyperintensities in the central nervous system in infantile Pompe disease
199	Nerissa Kreher	Evaluating the content validity of the Diary of Irritable Bowel Syndrome Symptoms - Mixed (DIBSS-M) to assess gastrointestinal symptoms associated with Fabry disease
200	Francyne Kubaski	Identification of MPS clusters in Latin America: An opportunity for targeted health care programs
201	Francyne Kubaski	Can MPS patients be identified by facial features
202	Francyne Kubaski	MPS Brazil Network: A summary of all mucopolysaccharidosis type IIIB patients
203	Gé-Ann Kuiper	Thoracolumbar kyphosis in MPS I: A natural history study and an international consensus procedure for the development of a clinical practice guideline
204	Anatalia Labilloy	Deep vein thrombosis is a common life-threatening complication in mucopolysaccharidosis type II
205	Jean Lacey	Mucopolysaccharide quantitation in urine by LC-MS/MS
206	Karima Lafhal	Development of a technical colorimetric for the determination of galactosis in blood
207	Christina Lampe	Transition from paediatric to adult care in patients with mucopolysaccharidosis (MPS)
208	Dawn Laney	Interfamily variability in patients with classical Fabry disease

209	Heather Lau	Long-term analysis of velaglucerase alfa-treated patients with Gaucher disease who entered the Gaucher Outcomes Survey (GOS) real-life registry
210	Heather Lau	Clinical characteristics of patients with neuronopathic and non-neuronopathic mucopolysaccharidosis type II: Data from the Hunter Outcome Survey
211	Ralph Laufer	AAV gene therapy LYS-SAF302 demonstrates widespread sulfamidase distribution in primate brain and correction of disease pathology in MPS IIIA mice
212	Christiane Auray-Blais	High-risk screening for Fabry disease in chronic kidney disease patients
213	Chris Lee	Pharmacological chaperone therapeutics for Krabbe disease
214	Malte Lenders	Dose-dependent impact of ERT on neutralizing anti-drug antibodies and long-term outcomes in Fabry disease
215	Malte Lenders	Patient-specific Fabry disease cell models as a tool to evaluate the amenability to chaperone therapy
216	Malte Lenders	Generation of patient-specific human induced pluripotent stem cells to analyze mutation- and cell-specific pathomechanisms in Fabry disease
217	Renuka Limgala	Selective large scale screening for lysosomal diseases in minority groups shows higher incidence rates
218	Ales Linhart	Pegunigalsidase alfa for the treatment of Fabry disease: Preliminary results from a phase III open label, switch over study from agalsidase alfa
219	Valynne Long	Like mother, like daughter: A case report of multiple family members affected by Pompe disease
220	Laura López de Frutos	Cyp2d6 allelic characterization on type 1 Gaucher disease patients
221	Georgia Loucopoulos	Determining the disease specific knowledge gaps in patients, family members, and caregivers living with lysosomal diseases
222	Eric Hui	Platform technology for treatment of the brain in lysosomal disorders: Application to Niemann Pick type A disease
223	Zoltan Lukacs	Twelve-year experience with a rapid and simple fluorometric tripeptidyl peptidase 1 (TPP1) assay using dried blood specimens to diagnose CLN2 disease
225	Cathleen Lutz	Preclinical gene therapy in a mouse model of Charcot-Marie-Tooth disease type 4J
LB-55	Alan Finglas	Multiple Sulfatase Deficiency -**The Diagnosis Needs the Patients**

Wednesday, February 6– Poster Presentations

226	Farrah Mahan	Pain and fatigue associated with generalized joint hypermobility in Gaucher disease
227	Samantha Marcellus	The impact of newborn screening for lysosomal disorders in a non-screening adjacent state
228	Ignacio Marin-Leon	Spanish multidisciplinary clinical practice guideline on Anderson-Fabry disease in adults: A live guideline
229	Deborah Marsden	The MPS VII disease monitoring program (DMP) is a novel, longitudinal, cohort program with rigor beyond a traditional registry
230	Ryuichi Mashima	Quantification of 11-plex LSD enzyme activity using liquid chromatography-tandem mass spectrometry
231	Lauren Mason	Long term biomarker analysis to assess cardiac involvement in Fabry disease
233	Atul Mehta	Development of an algorithm to facilitate diagnosis of Gaucher disease
235	Carlos Miranda	Liver directed AAV gene therapy to treat Gaucher disease
236	Pramod Mistry	Two years of efficacy of oral eliglustat in treatment-naïve and switch patients enrolled in the International Collaborative Gaucher Group Gaucher registry
237	Takashi Miyajima	Generation of iPS cells derived from skin fibroblasts of patients with Fabry disease using RNA-reprogramming

238	Francisco del Castillo	Unexpected genetic findings in a Gaucher disease patient analysed by NGS-based panel sequencing
239	Luciana Moreira	CRISPR/Cas in iPSCs from sphingolipidoses patients
240	Maria Moreira	Are we missing complex rearrangements by next generation diagnostic approaches: A case report of a complex rearrangement in MPS II
241	Patricia Moreno	Accreditation: A challenge for a research laboratory
242	Branden Moriarity	CRISPR/Cas9 mediated insertion of α-L-iduronidase (IDUA) and anti-PE receptor in B-lymphocytes for selective activation into long-lived plasma cells for sustainable IDUA expression
243	Juan Mucci	Evaluation of PPS treatment in osteoclast-osteoblast imbalance using in vitro models of Gaucher disease
244	Joseph Muenzer	Neurodevelopmental status and adaptive behavior of pediatric patients with Hunter syndrome: A longitudinal observational study
245	Joseph Muenzer	Evaluation of the long-term treatment effects of idursulfase using statistical modelling: Data from the Hunter outcome survey (HOS)
246	Joseph Muenzer	Characteristics of patients with mucopolysaccharidosis type II who have received a bone marrow transplant: Data from the Hunter Outcome Survey
249	Behzad Najafian	A novel method for quantification of globotriaocylceramide (GL-3) inclusions in affected podocytes in females with Fabry disease shows progressive accumulation of GL-3 in podocytes with age and no cross-correction between affected and non-affected podocytes
250	Luba Nalysnyk	Saccadic eye movements and its use as clinical endpoints in lysosomal disorders: A literature review
251	Igor Nestrasil	Quantitative brain MRI in patients with alpha-mannosidosis: Study from 3 centers
252	Igor Nestrasil	Discovery of brain MRI signatures in infants with severe form of MPS I in the pre-HSCT and post-HSCT stages
255	Yann Nguyen	Monoclonal gammopathies and hypergammaglobulemia in Gaucher disease: An analysis from the French Gaucher disease registry
256	Kathleen Nicholls	The effects of long-term migalastat treatment in Fabry disease patients previously treated with enzyme replacement therapy who have migalastat-amenable variants with low alpha-galactosidase A response in the in vitro migalastat amenability assay
257	Kim Nickander	Multiplex assay for the tandem detection of ceramide trihexosides and sulfatides: Efficient first tier screening for Fabry, MLD, MSD, and Saposin B in urine
258	Vera Niederkofler	CBE treatment of alpha-synuclein over-expressing and wildtype mice models Gaucher disease pathology
259	Vera Niederkofler	Characterization of 4L/PS-NA mice for enzyme activity, substrate concentrations as well as inflammation to model Gaucher disease
260	Vera Niederkofler	Behavioral and histological hallmarks of a mucopolysaccharidosis type IIIA mouse model
261	Graeme Nimmo	Reduction of paraprotein levels in type 1 Gaucher disease with enzyme therapy
262	Esther Noël	Fabry patients' needs and expectations regarding their treatment in France: Development of a Patients' Need Questionnaire (PNQ Fabry)
263	Albina Nowak	Fabry disease: Incidence of pathogenic GLA mutations estimated by newborn screening studies
264	Albina Nowak	Fabry disease genotype, phenotype and amenability: A full country overview
265	Andrew Oldham	Potential benefits of Fitbit device in managing a patient with mucopolysaccharidosis
266	Claire O'Leary	Improving brain delivery of adeno-associated viral gene therapy vectors for the treatment of MPS IIIC
268	Cara O'Neill	The natural history of facial features observed in Sanfilippo syndrome (MPS IIIB) using a next generation phenotyping tool

269	Cara O'Neill	Meaningful treatment outcomes for Sanfilippo syndrome: A study of caregiver preferences and prioritization
271	Chris Orsborne	Real-world baseline data in patients established on migalastat for Fabry disease
272	Saida Ortolano	Functional evaluation of an AAV9 based vector expressing alpha-galactosidase A for potential gene therapy of Fabry disease
273	Anureet Pabla	Qualitative interviews with patients with mucopolysaccharidosis type I (MPS I) and caregivers to evaluate the Mucopolysaccharidosis Health Assessment Questionnaire (MPS-HAQ)
274	Ron Padilla	Dysfunctional autophagy impairs muscle regeneration in lysosomal diseases
275	William Pardridge	Platform technology for treatment of the brain in lysosomal diseases: Application to NCL1 Batten disease
276	Samantha Parker	Capturing the MPS IIIA patient and family voice in orphan drug development to appreciate what is important in managing the disease and improving quality of life
277	Livia Paskulin	Rare GBA1 genotype in two siblings with a severe bone phenotype of type 1 Gaucher disease
278	Loren Pena	NEO1 and NEO-EXT studies: Long-term safety of repeat avalglucosidase alfa dosing for 4.5 years in late-onset Pompe disease patients
280	Merlene Peter	Case control study to identify the prevalence of menstrual and pregnancy complications in women with mucopolysaccharidosis
281	M. Judith Peterschmitt	Correlations between glucosylsphingosine (lyso-GL-1) and baseline disease severity as well as response to treatment in two clinical trials of eliglustat in treatment-naïve adults with type 1 Gaucher disease
282	M. Judith Peterschmitt	Safety, tolerability and pharmacokinetics of oral venglustat in Parkinson's disease patients with a GBA mutation
284	Päivi Pietilä-Effati	The natural course of the Fabry disease in a Finnish R227X cohort
285	Luisa Pimentel	Intranasal delivery of the CRISPR-Cas9 system for gene editing in MPS II mice
287	Laura Pollard	Multiplex DBS enzyme assay for MPS II, IIIB, IVA, VI, VII and CLN2 via LC-MS/MS expands clinical utility of DBS enzyme testing
288	Carlos Prada	Utility of multiple myeloma screening in Gaucher disease
289	Alejandra Puentes-Tellez	Evaluation of lentiviral vectors in Morquio syndrome type A patients' fibroblasts
290	Ana Puhl	Industrializing enzyme replacement therapy development
292	Alexander Pushkov	Molecular description of the first case of Hunter syndrome burdened with Turner syndrome in Russian girl
294	Uma Ramaswami	Fabry disease mobile phone application - A new service improvement tool
295	Lewis Raynor	Real-world outcomes in pregnant imiglucerase-treated patients with Gaucher disease: Data from the global safety database and International Collaborative Gaucher Group (ICGG) Gaucher registry pregnancy sub-registry maintained by Sanofi Genzyme
296	Catherine Rehder	Improving Pompe diagnostics through modification of the ACMG-AMP variant classification criteria by the Clinical Genome Resources (ClinGen) Lysosomal Storage Diseases Variant Curation Expert Panel
297	Shoshana Revel-Vilk	Longitudinal follow-up (4 years) of lyso-Gb1 in children with Gaucher disease in a single center cohort
298	Shoshana Revel-Vilk	Screening for serum free light chains in patients with type 1 Gaucher disease
299	Salvador Rico	Clinical program to evaluate safety, preliminary efficacy, and dose selection of AAV8 gene therapy in patients with infantile and late onset Pompe disease (IOPD and LOPD)
300	Nilton Rosa Neto	Assessment of bone mineral density and FRAX tool analysis in a cohort of male Fabry disease patients and GLA gene GVUS subjects
301	Nilton Rosa Neto	Inflammation in Fabry disease: Correlation with Mainz Severity Score Index (MSSI)

302	Nilton Rosa Neto	Depression, sleep disturbances, disability and quality of life in a cohort of Brazilian Fabry disease patients and GLA gene GVUS subjects
303	Nilton Rosa Neto	Dual-energy X-ray absorptiometry body composition assessment in male Fabry disease patients and GLA gene GVUS subjects
304	Nilton Rosa Neto	Rheumatic and autoimmune/autoinflammatory manifestations in a cohort of Brazilian Fabry disease patients
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311	Tom Rouwette	Real-world/health economic publication patterns before and after enzyme replacement therapy approval across lysosomal diseases
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313	Brante Sampey	The pro-inflammatory immunophenotype of a Farber disease mouse model is ameliorated by repeated dosing with RVT-801, a developmental enzyme replacement therapy for Farber disease
314	Angela Sanchez	Gaucher disease and associated plasma cell neoplasia: A diagnostic dilemma
315	Maria Dolores Sanchez-Niño	Lyso-Gb3 modulation of gut microbiota biofilms: Potential contribution to Fabry disease gastrointestinal symptoms and systemic complications
316	Markku Savolainen	Study to determine predictive potential of an algorithm for earlier diagnosis of Gaucher disease: Retrospective biobank study utilizing real-world data available in Finland
317	Kirill Savostyanov	Lyso-Gb3 is as a primary biomarker for Fabry disease screening among high-risk contingents
318	Kirill Savostyanov	Glucosylfingosine (Lyso-GL1) may be the primary biomarker for screening Gaucher disease in Russian patients
319	Maurizio Scarpa	The European Reference Network Program for Hereditary Metabolic Diseases (MetabERN)
320	Raphael Schiffmann	Venglustat in adult Gaucher disease type 3: Preliminary safety, pharmacology, and exploratory efficacy from a phase 2 trial in combination with imiglucerase (LEAP)
321	Lars Schlotawa	Improved description of clinical features of multiple sulfatase deficiency: A meta-analysis of published cases
322	Lars Schlotawa	Protein disulfide isomerase is a possible target for disease modification in multiple sulfatase deficiency
323	Ashley Schneider	Emotional, social, behavioral, and pain self-report measures and outcomes in Morquio syndrome
324	Benedikt Schoser	Preliminary patient-reported outcomes and safety of advanced and targeted acid α-glucosidase (AT-GAA) in patients with Pompe disease from the ATB200-02 trial
325	Roselena Schuh	Newborn genome editing improves phenotype, cardiovascular, respiratory, and bone disease in mucopolysaccharidosis type I mice
327	Ida Schwartz	Genetic approaches for diagnosis of two Brazilian patients with mucolipidosis II/III alpha/beta
328	Gurpreet Seehra	The utility of the swallow study in the diagnosis and management of type 2 Gaucher disease
329	Jin-Song Shen	Dysregulated DNA methylation in the pathogenesis of Fabry disease
330	Pavel Shiyanov	Phase I/II gene transfer clinical trial of scAAV9.U1a.hSGSH for MPS IIIA vector shedding results over 6 months post-gene transfer

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335	Martha Solano Villarreal	Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)
336	Alexander Solyom	Farber disease (acid ceramidase deficiency): Data from an ongoing natural history study
337	Katherine Spurlock	Evidence of attention problems in Morquio syndrome
338	Rodrigo Starosta	Liver biopsy findings in patients with Gaucher disease: Experience of the reference center of Rio Grande do Sul, Brazil
339	Mihaela Stefanescu	Parent report of school functioning and behavior in children with Pompe disease
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342	Alta Steward	The natural history of cognition in type 3 Gaucher disease
343	Ashlee Stiles	Quantification of glucosylsphingosine in plasma/serum by UPLC-MS/MS
344	David Stockton	Impact of time from diagnosis to treatment on lung function among patients with late-onset Pompe disease: Data from the Pompe registry
345	Katharina Stumpfe	Reconstructive aortic valve surgery in a type 3 Gaucher patient homozygous for the p.D409H mutation
346	Gere Sunder-Plassmann	Design of a prospective, multicenter, multinational, observational safety and outcomes registry in Fabry disease patients treated with migalastat and untreated patients
347	Gere Sunder-Plassmann	Clinical outcomes after switching to migalastat from agalsidase alfa or agalsidase beta in patients with Fabry disease: Post hoc analysis from ATTRACT
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350	Lorraine Thompson	Mortality in Fabry disease cohort: One centre experience
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352	Lina Titievsky	Baseline characteristics of patients with Gaucher disease enrolled in the taliglucerase alfa surveillance (TALIAS) registry
353	Shunji Tomatsu	Newborn screening of mucopolysaccharidoses: Past, present, and future
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362	Alfredo Uribe	High-risk population screening for Sly syndrome: Application of a micro-method in collected dried blood spots
363	Vassili Valayannopoulos	Alglucosidase alfa inhibitory antibodies and clinical correlates in treatment-naive late-onset Pompe disease patients in the late-onset treatment study (LOTS) over 78 weeks: A new post hoc analysis
364	Ans Van der Ploeg	Lysosomal subnetwork of MetabERN: Objectives and organizational structure
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367	Ravi Vijapurapu	The natural progression of cardiac involvement in Fabry disease
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369	Susanne Walls	The Fabry PRO online tool for secure and continuous patient follow-up and communication
370	Feng Wang	Evaluation of a potential of chaperone therapy for mucopolysaccharidosis type IIID
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372	Christoph Wanner	Renal and cardiac outcomes in female patients with Fabry disease treated with agalsidase beta: A Fabry registry analysis of pre- versus post-treatment comparison
373	David Warnock	Analysis of the baseline characteristics of Fabry disease patients screened for the pegunigalsidase alfa phase III BALANCE study
374	Jill Weimer	Promise of AAV9 gene therapy in the treatment of Batten disease: Systematic approach in therapy design reduces pathological and behavioral deficits and prolongs survival in mouse models of CLN3-, CLN6-, and CLN8-Batten disease
375	Neal Weinreb	Type 1 Gaucher disease severity and risk for liver fibrosis in untreated patients and in patients treated primarily with enzyme replacement therapy for a median of 20 years
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380	Frits Wijburg	Design, baseline characteristics, and 18-24 months follow-up from the MPS IIIA natural history study
381	Matheus Wilke	Evaluation of the frequency of pre-motor symptoms of Parkinson disease in adult patients with type 1 Gaucher disease
382	Kara Woolgar	Immune modulation for a female Hunter syndrome patient prior to starting idursulfase
383	Kara Woolgar	Superior vena cava syndrome in an infantile onset Pompe patient
384	Kara Woolgar	Intravenous 2-hydroxypropyl-beta-cyclodextrin for a Niemann-Pick disease type C1 infant with liver cirrhosis
385	Hiroyuki Yamakawa	Fabry disease has been found by using of the tumor mutational burden analysis of 3000 Japanese cancer genomes using whole exome and targeted gene panel sequencing: Project Hightech Omics-based Patient Evaluation (Project HOPE)
387	Karen Yee	Analysis of cognitive ability and adaptive behavior assessment tools used in an observational study of patients with Hunter syndrome
388	Mildrid Yeo	Airway stenting in MPS IVA (Morquio syndrome type A)
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390	Natalia Zhurkova	Deletion of chromosomal region Xq28 cause of Hunter syndrome in a patient with mild clinical phenotype
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