Poster Session Abstracts

Poster sessions will be in the Harbor Ballroom on:

Tuesday, February 6 from 4:30-6:30pm
Wednesday, February 7 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 6, 2018 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 7, 2018 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 7, 2018 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 6 – Poster Presentations

Poster #	First Author	Abstract Title
1	Magy Abdelwahab	Osteopenia in Egyptian Gaucher disease type 3 patients: an 11 year prospective study
2	Qais Abu Ali	Association between genotype, phenotype, and heat map assessments post enzyme replacement therapy in a highly heterogeneous MPS VII study population
3	Heather Adams	Using PEDI-CAT to assess functional capabilities in CLN3 (Batten) disease
4	Patricio Aguiar	Collagen type I synthesis biomarkers predict the progression of Fabry disease cardiomyopathy
5	Patricio Aguiar	Tubular dysfunction biomarkers in Fabry disease: better than albuminuria to identify patients at risk of nephropathy progression
6	Amelia Ahern-Rindell	A variant of GM1 gangliosidosis in a sheep model
7	Alia Ahmed	Association of hippocampus and amygdala volumes with neurocognitive and behavioral outcomes in patients with Hurler-Scheie syndrome heterozygous for the L238Q mutation
8	Rebecca Ahrens-Nicklas	Complex care of patients with multiple sulfatase deficiency: clinical cases and guideline consensus statement
10	Carlos Almeciga-Diaz	Characterization of two potential pharmacological chaperones for N-acetylgalactosamine-6-sulfate sulfates (GALNS) enzyme
13	Dominick Amato	Gaucher disease in Ontario, Canada: clinical manifestation, natural disease progression, and treatment response
14	Carolina Aranda	Can a mucopolysaccharidosis also be a primary immunodeficiency?
15	Graciela Arguello Florencio	Genistein activates TFEB and induces lysosomal clearance in Niemann-Pick disease type C models
16	David Arkadir	Glucosylsphingosine concentration is the most reliable response-parameter for low-dose ERT in homozygous N370S Gaucher patients
17	Christiane Auray-Blais	Mass or high-risk screening of mucopolysaccharidoses using urine samples collected on filter paper

18	Stephanie Austin	Insight into the phenotype of infants with Pompe disease identified by newborn screening with the common c.-32-13T>G “late-onset” GAA variant
20	Mitch Bailey	Mucopolysaccharidosis type VI (MPS VI) and molecular analysis: a review of published classified variants in the ARSB gene
21	Zahra Bakhtiar	Prevalence of autoimmune disorders and cancer in a Gaucher type 1 population at a large metropolitan referral center for lysosomal disorders in New York City
22	Amy Barczykowski	Death rates in the United States due to leukodystrophies in the global leukodystrophy initiative registry and related lysosomal disorders
23	Nicholas Bascou	Anesthesia safety in progressive leukodystrophies: a retrospective study of patients with Krabbe disease and metachromatic leukodystrophy undergoing general anesthesia
24	Nicholas Bascou	Natural history of late-infantile Krabbe disease
25	Suelen Basgalupp	Biomarkers of vitamin B12 status respond to therapy in Gaucher disease
27	Donna Bernstein	Lysosomal acid lipase deficiency is associated with premature death in children and adults
28	Abdelkrim Berrah	Comorbidities in Gaucher disease
30	Olaf Bodamer	Early initiation of enzyme replacement therapy in pediatric Fabry disease
31	Araceli Borja-Borja	Lysosomal diseases in ophthalmology
32	Elizabeth Braunlin	Natural history of cardiac findings in mucopolysaccharidosis type I: report from an international registry
33	Alexander Broomfield	10 years of galsulfase in a tertiary treatment center
35	Jillian Brown	Guanidinylated neomycin conjugation enhances intranasal enzyme replacement in the brain
36	Alberto Burlina	Neonatal screening for lysosomal disorders in Italy: the initial 17-month experience
37	Barbara Burton	Infusion-related reactions in patients with mucopolysaccharidosis type II on idursulfase enrolled in the Hunter Outcome Survey (HOS)
39	Enrique Calderón	Application of iTRAQ shotgun proteomics for revealing metabolic changes related to Gaucher disease
40	Fabrice Camou	Gaucher disease: French registry cohort of 58 patients treated with eliglustat
41	Randy Carter	A comparison of three proposed methods of newborn screening for early infantile Krabbe disease
42	David Cassiman	Disease severity scoring system for acid sphingomyelinase deficiency: severity score domains and components
43	Irene Chang	Proteolytic immuno-SRM-MSMS in dried blood spots to determine immunogenicity in patients with infantile Pompe disease
44	Erk Changsila	Effect of ERT on autophagy and mitochondrial functions with Fabry disease
45	Joel Charrow	Long-term stability in randomized and non-randomized patients in the phase 3 randomized, double-blind EDGE trial of once- versus twice-daily dosing of eliglustat in patients with Gaucher disease type 1
47	Brenden Chen	Precision medicine for Fabry disease: benign and like-benign missense mutations
49	Pin-Wen Chen	Multiplex UPLC-MS/MS assay of 8 lysosomal disorders in newborn screening
50	Yin-Hsiu Chien	Newborn screening for Gaucher disease and MPS I disease: a single center experience in Taiwan
51	Kyungsun Choi	Exosome-based delivery of glucocerebrosidase lysosomal enzyme for treatment of Gaucher disease
52	Samuel Chu	The impact of GC content on CRISPR/Cas9 gene editing: implications for mucopolysaccharidosis type IIIB
53	Heather Church	How many sulphatase deficiencies become multiple: an unusual presentation of multiple sulphatase deficiency

54	Tricia Cimms	Qualitative research to characterize patients with galactosialidosis
55	Tanya Collin-Histed	Collaborative working to advance standards of care and the well-being of lysosomal disorder patients and families in the UK
56	Erin Conboy	Attenuated mucopolysaccharidosis type VI: the need for a high index of suspicion
57	Jonathan Cooper	Testing combinatorial therapies for juvenile Batten disease
59	Claudia Cozma	Lyso-SM-509 as highly sensitive biomarker for Niemann-Pick disease types A/B and C: three years experience
60	Claudia Cozma	Biochemical and genetic characteristics of the largest worldwide Fabry cohort reported to present
61	Claudia Cozma	Glucosylsphingosine concentration in the blood of Gaucher patients reflects the severity of GBA mutations - data from a large global cohort
62	Claudia Cozma	Quantification of glucosylshingosine (lyso-Gb1) for the diagnosis and monitoring of Gaucher disease
63	Andrea Crivaro	PPS beyond MPS: efficacy in Fabry and Gaucher *in vitro* studies
64	Christine Dali	Gender, mutations and residual enzymatic activity: investigation of predictive factors of alpha-mannosidosis phenotypic presentation and of response to velmanase alfa long term enzyme replacement therapy
65	Mark Dant	ConnectMPS registry project: connecting mucopolysaccharidosis and mucolipidosis patients
67	Adam Davis	AAV vector comparability across mammalian and insect cell production platforms for treatment of lysosomal diseases
68	James Davison	Dilated cardiomyopathy at presentation of mucopolysaccharidosis type IH (Hurler syndrome): clinical characteristics and management outcome
69	James Davison	Initiation of enzyme replacement therapy for pediatric Fabry disease: review of single centre practice and experience
70	Francisco del Castillo	Fast genetic diagnosis of lysosomal disorders by means of a novel NGS-based resequencing gene panel
71	Ankit Desai	An immune tolerance approach using methotrexate in the naïve setting of patients treated with a therapeutic protein: experience in infantile Pompe disease
72	Jordi Díaz-Manera	Anti-rh-GAA antibodies does not influence late onset Pompe disease progression
73	Patricia Dickson	Neuroimaging and neuropathology reveal progressively abnormal white matter and cerebrospinal fluid volume in MPS I dogs
74	Aimee Donald	Feasibility of wearable technologies as an outcome measure in neuronopathic Gaucher disease (nGD)
75	Aimee Donald	Neuronopathic Gaucher disease; patient association takes the lead to improve awareness and encourage collaborative working
76	Aimee Donald	Is it time to redefine what it means to have type 3 Gaucher disease?
77	Alícia Dornelles	Enzyme replacement therapy for late-onset Pompe disease: a systematic review
78	Karen Dublán-García	Experience with two different enzyme replacement therapies in siblings with Fabry disease: 2 years follow up analysis
79	Jonathan Dyke	Asymptotic neurodegeneration in CLN2 disease assessed by MRI cortical thickness histograms
80	Julie Eisengart	Neurocognitive outcomes of intrathecal enzyme replacement therapy and transplant in Hurler syndrome
81	Sean Ekins	Gaps and opportunities in MPS/ML for small rare disease companies to address
82	N. Matthew Ellinwood	Pharmacology of BMN 250 administered via intracerebroventricular infusion once every 2 weeks for twenty-six weeks or longer in a canine model of mucopolysaccharidosis type IIIB

85	Jeffrey Esko	Impaired mitophagy causes brown adipose tissue activation and cachexia in Sanfilippo syndrome type A mice
87	Francois Eyskens	Newborn screening for lysosomal diseases: the need for sex and gestational age dependent reference ranges
88	Ulla Feldt-Rasmussen	The D313Y variant in the GLA gene - no evidence of a pathogenic role in Fabry disease in 2 Danish families
89	Nicolas Fernandez Escobar	Use of trabecular bone score (TBS) as a complementary approach to bone mineral density (BMD) in Gaucher disease patients treated with imiglucerase
90	Henrique Ferrer	Identification of a novel GLA mutation in a family with classical phenotype of Fabry disease
91	Sergio Figueroa-Sauceda	Neurological manifestations in Fabry disease, series of 11 cases, from a high specialty medical unit, IMSS, Ciudad Obregon, Sonora, Mexico
92	Niamh Finnigan	Significant variability in phenotype in an unusual family with attenuated MPS II (Hunter syndrome)
93	Jessica Fischetti	Clinical and neurophysiological assessments of an infant with acute neurovisceral acid sphingomyelinase deficiency
95	Stefanie Flunkert	Pathological hallmarks of the Gaucher disease mouse model 4L/PS-NA
96	Stefanie Flunkert	Early onset of the pathological phenotype in line 61 α-synuclein transgenic mice
97	Stefanie Flunkert	Hepatic and neuronal phenotype of the murine Niemann-Pick disease type C mouse model NPC1-/-
98	Allison Foley	Planning, implementation, and initial results of newborn screening for Pompe disease and MPS I in Georgia
99	Joaquin Frabasil	Screening of Fabry disease in male hemodialysis patients: prevalence and mutations found in Argentina
100	Cecile Freihuber	Effects of miglustat therapy on neurological disorder and survival in early-infantile Niemann-Pick disease type C: a national French retrospective study
101	Vyacheslav Furtak	Identification of a novel mutation in the α-galactosidase A gene in a large family using NGS platform
102	Mahoko Furujo	Enzyme replacement therapy attenuates disease progression in two Japanese siblings with mucopolysaccharidosis type VI: 10-year follow up (case report)
103	Eric Joshua Garcia	Exploring the role of epigenetics in Gaucher-associated alpha-synucleinopathies
104	John Gargus	Sudden-death of infant with lysosomal acid lipase deficiency successfully completing sebelipase alfa clinical trial
105	Sam Gary	Mouse macrophage model of Gaucher disease for evaluating non-inhibitory chaperone candidates
106	Eric Gaukel	Pharmacokinetics and tissue distribution of RVT-801, a recombinant human acid ceramidase, at efficacious doses in a murine model of Farber disease
107	Eric Gaukel	Prediction of the human equivalent dose (HED) of RVT-801, a recombinant human acid ceramidase, for the treatment of Farber disease
108	Amy Gaviglio	State of national implementation for lysosomal diseases
109	Michael Gelb	High accuracy enzymatic assay for enhanced second-tier follow up of lysosomal diseases
110	Kelly George	Comprehensive exploratory study to identify novel biomarkers of Pompe disease
111	Arunabha Ghosh	Experience of the nutritional management of infantile onset lysosomal acid lipase deficiency (LAL-D)
113	Jane Gillis	North American experience with laronidase enzyme replacement therapy for mucopolysaccharidosis type I in a home infusion setting
114	Pilar Giraldo	Pharmacogenetic study in type 1 Gaucher disease patients
115	Pilar Giraldo	Correlation between bone disease and genotype in type 1 Gaucher disease: data from Spanish Gaucher disease registry
116	Pilar Giraldo	Defective function of KCa3.1 channels in lysosomal disorders

117	Luciana Giugliani	Somatic effects of AGT-181 in patients with mucopolysaccharidosis I enrolled in a phase I/II clinical trial in Brazil
118	Roberto Giugliani	Intrathecal delivery of recombinant human arylsulfatase A in children with late-infantile metachromatic leukodystrophy: a post hoc analysis of responders and non-responders
121	Kirill Gorshkov	High-throughput assay development for Niemann-Pick disease type A small molecule therapeutics
122	Nathan Grant	Safety of enzyme replacement therapy to treat Hunter syndrome in children aged 5 years and under
123	John Gray	Establishing the optimal tissue target for alpha-glucosidase gene delivery in Pompe disease
124	Abhinav Grover	Optimization of CRISPR mediated genome correction of Pompe disease-specific GAA mutations in C2C12 mouse myoblasts
126	Punita Gupta	Positive newborn screening results for late onset conditions: the clinician's dilemma
127	Susanne Gustavsson	Therapeutic correction of central nervous system pathology by intravenously administered chemically modified sulfamidase in a mouse model of mucopolysaccharidosis type IIIA
128	Alaa Hamed	Fabry disease symptoms and impacts on daily life − a conceptual model
129	Natalja Haninger-Vacariu	Pregnancy outcome after exposure to migalastat: a case study
130	Paul Harmatz	Global treatment responder analysis demonstrates clinically relevant effect of velmanase alfa long term enzyme replacement therapy for alpha mannosidosis, in a phase III randomized placebo controlled trial
131	Paul Harmatz	Global treatment response analysis of velmanase alfa long term enzyme replacement therapy for alpha-mannosidosis shows treatment benefit across ages
132	Paul Harmatz	Mucopolysaccharidosis VI enzyme replacement therapy initiated in adulthood: findings from the MPS VI clinical surveillance program
133	Paul Harmatz	Mucopolysaccharidosis type VI enzyme replacement therapy outcomes across the disease spectrum: findings from the MPS VI clinical surveillance program
134	Paul Harmatz	Update on phase 1/2 clinical trials for MPS I and MPS II using ZFN-mediated in vivo genome editing
135	Magdalena Harrington	Metachromatic leukodystrophy and caregiver perspectives: understanding the natural history of the disease from interviews with caregivers
136	Caroline Hart	Improving the management of enzyme replacement therapy infusion reactions: increasing preparedness through multi disciplinary clinical simulation training
138	Christian Hendriksz	Assessing the effectiveness of miglustat in Niemann-Pick disease type C
139	Christian Hendriksz	Safety, efficacy, and immunogenicity of elosulfase alfa in patients with Morquio A syndrome participating in 2 sequential open-label studies (MOR-002/MOR-100), representing 5 years of treatment
140	Julia Hennermann	Pharmacokinetics, pharmacodynamics, and safety of moss agalactosidase A in patients with Fabry disease
141	Julia Hennermann	Clinical variability of GM1 gangliosidosis
142	Mrudu Herbert	Cognition and brain involvement in infantile Pompe disease
143	Stacy Hewson	A multidisciplinary approach to the transition of adolescents with a lysosomal disorder from a pediatric to an adult health care centre
144	Wendy Heywood	The effect of treatment on urinary glycosylated lysine a marker of extracellular matrix integrity and lysosome function in mucopolysaccharidosis types I, II, IV and VI patients
145	Max Hilz	Do we understand the pathophysiology of gastrointestinal symptoms in patients with Fabry disease?
146	Max Hilz	Proposal of a rating scale to recognize Fabry disease in patients with nonspecific

		gastrointestinal symptoms
147	Christian Hinderer	Preclinical development of a platform for enzyme therapy in the CNS of MPS I and MPS II patients based on intrathecal AAV delivery
148	Yuan-Yuan Ho	Molecular diagnostic findings of lysosomal storage diseases in children and adults suspected to have inborn errors of metabolism
149	Carla Hollak	Agalsidase alfa versus agalsidase beta for the treatment of Fabry disease: an international cohort study
150	Elise Holmes	Minnesota Department of Health long-term follow-up of newborn screening conditions: new applications for Pompe disease and MPS I
151	Robert Hopkin	A survivor analysis for major clinical events in heterozygous female patients with Fabry disease using group consensus phenotype classifications from hemizygous male patients
153	Mohammad Hossain	Methylation study of CpG islands in GLA gene: best clinical phenotype predictor for heterozygous Fabry female
154	Jeffrey Huang	Characterization of CRISPR-Cas9 generated Pompe disease models
155	Derralynn Hughes	Menarche, menopause, and pregnancy data in untreated females and females treated with agalsidase alfa in the Fabry Outcome Survey
157	Derralynn Hughes	Pegunigalsidase alfa, a novel PEGylated ERT for Fabry disease - two years safety and efficacy follow up
158	Marshall Huston	Liver-based expression of the human alpha-galactosidase A gene in a murine Fabry model results in continuous therapeutic levels of enzyme activity and effective substrate reduction
159	Jackie Imrie	The International Niemann-Pick Disease Registry (INPDR) - a new model of patient-empowered data ownership and management
160	Kohji Itoh	A novel glycotechnology to produce human lysosomal enzymes carrying synthetic N-glycans with terminal mannose 6-phosphate residues and application to enzyme placement therapy for lysosomal diseases
161	Margarita Ivanova	Individualized screening for chaperone activity in Gaucher disease using multiple patient derived primary cell lines
162	Emanuela Izzo	Use of epilepsy gene panels for early diagnosis of epilepsy in children 2-4 years of age: expert considerations on current and future practices in Europe
163	Kabir Jalal	Laboratory reliability and validity for measuring psychosine concentration and GALC enzyme activity for use in newborn screening for Krabbe disease
164	Jinlong Jian	Chitinase-3-like protein 1: a novel biomarker for Gaucher disease
165	Jinlong Jian	Progranulin stabilizes hexosaminidase A and is therapeutic in Tay-Sachs disease
166	Lei Jiang	Efficacy of systemic messenger RNA therapy to treat and prevent porphyria attacks in animal models of acute intermittent porphyria
168	Franklin Johnson	First-in-human preliminary pharmacokinetic data on a novel recombinant acid α-glucosidase, ATB200, co-administered with the pharmacological chaperone, AT2221, in patients with late-onset Pompe disease
169	Harrison Jones	Lingual pathophysiology in late-onset Pompe disease
170	Harrison Jones	Respiratory muscle training in Pompe disease
171	Simon Jones	Management guidelines for infantile onset lysosomal acid lipase deficiency (LALD)
172	Simon Jones	Profile of natural history in 104 patients with mucopolysaccharidosis type II: insights from the Hunter Outcome Survey (HOS)
173	Simon Jones	Effect of sebelipase alfa on survival to 3 years of age and liver function in infants with rapidly progressive lysosomal acid lipase deficiency: results from two studies
174	Chanchala Kaddi	Integrated quantitative systems pharmacology (QSP) model of lysosomal diseases provides an innovative computational platform to support research and therapeutic development for the sphingolipidoses

175	Ilkka Kantola	Deterioration in ECG parameters in Fabry females and males treated by enzyme replacement therapy either for 5 or 10 years
176	Amel Karaa	Factors influencing patient preferences for oral versus intravenous (IV) enzyme replacement medication
177	Reena Kartha	Blood and brain biomarkers of oxidative stress and inflammation in type 1 Gaucher disease: effect of antioxidant therapy
178	David Kasper	Five year experience of diagnostic testing for lysosomal diseases in Germany, Austria and Switzerland
179	David Kasper	Importance of lyso-GL-3 (lyso-Gb3) for primary diagnostics of Fabry disease: two-year experience in a daily routine laboratory
180	Asaka Katabuchi	Effects of psychosine-reducing agents in the twitcher murine model for Krabbe disease
181	Asaka Katabuchi	Generation of neurologically relevant disease-cell models for lysosomal diseases
182	Zoheb Kazi	A prediction model to identify infantile Pompe disease (IPD) patients at high-risk of developing significant anti-drug antibodies (ADA) utilizing acid α-glucosidase (GAA) variants and HLA-type
183	Shauna Kearney	A comparison of stem cell transplant versus enzyme replacement therapy in infants with MPS II
185	Scott Kerns	Intravenous administration of CLN3 gene therapy for juvenile neuronal ceroid lipofuscinosis
186	Gee-Hee Kim	A case of a 50-year-old woman with classic Fabry disease who showed serial electrocardiographic and echocardiographic changes for 17 years
187	Jin Sug Kim	Diagnosis of Fabry disease in a patient with end stage renal disease on hemodialysis: a case report
188	Sarah Kim	Optimizing immune tolerizing regimens for in-vivo gene therapy
189	Virginia Kimonis	Variable clinical features and progression in 18 patients with Pompe disease
190	Virginia Kimonis	Variable clinical features in Fabry disease in patients with novel mutations
191	Thomas Kirkegaard	Arimoclomol, a clinical candidate for neuronopathic Gaucher disease, increases glucocerebrosidase activity through amplification of heat shock proteins
192	Minako Kobayashi	Efficacy and safety of JR-051, a biosimilar of agalsidase beta, in patients with Fabry disease; results of a multicenter, open-label phase 3 study in Japan
193	Dwight Koeberl	Correction of biochemical abnormalities and gene expression associated with improved muscle function in a phase I/II clinical trial of clenbuterol in Pompe disease patients stably treated with ERT
194	Anja Koehn	A Gaucher type III patient with a homozygous p.D409H mutation and chronic inflammatory bowel disease
195	Simon Körver	Subjective cognitive complaints and symptoms of depression are highly prevalent in Fabry disease and are not related to objective cognitive impairment
196	Konstantinos Koulousios	The D313Y mutation of the GLA gene could be pathogenic for Fabry disease
197	Konstantinos Koulousios	Novel pathogenic GLA mutations revealed in a Greek population study for Fabry disease
198	David Kronn	Response to omalizumab in a patient with Pompe disease
199	David Kuter	Open-label expanded access study of taliglucerase alfa in patients with Gaucher disease requiring enzyme replacement therapy
200	Dawn Laney	ThinkGenetic: a pilot project to create an educational website/application providing increased access to information on accurate natural history, diagnosis, and treatment information with treatable genetic disorders to healthcare providers
201	Dawn Laney	A prospective, multicenter pilot study of Fabry disease clinical and biochemical findings in young pediatric patients: the MOPPet baseline data
202	Thomas Langan	Survey of quality of life, phenotypic expression and response to treatment in patients with Krabbe leukodystrophy

203	Heather Lau	Long-term treatment response based on severity of Gaucher disease type 1 at baseline after 8 years of treatment with oral eliglustat: final efficacy and safety results from a phase 2 clinical trial in treatment-naïve adult patients
206	Malte Lenders	New insights in efficacy of different dosages of enzyme replacement therapy due to treatment switch in Fabry disease
207	Malte Lenders	Characterization of drug-neutralizing antibodies in patients with Fabry disease during infusion with enzyme replacement therapy
208	Sarah Lewis	A thorough autonomy assessment of individuals with Hurler syndrome who survive into adulthood: a feasibility study using the Independent Living Study(ILS)
209	Jing Li	Impact of hepatic and renal impairment on the pharmacokinetics and tolerability of eliglustat
212	HsuanChieh Liao	Functional and biological studies of alpha galactosidase A variants with uncertain significance from newborn screening in Taiwan
213	Jeong-A Lim	The pros and cons of manipulating different pathways to address defective autophagy in Pompe disease
214	Renuka Limgala	Effect of two different therapeutic interventions: SRT in comparison to ERT on immune aspects and bone involvement in Gaucher disease
215	Renuka Limgala	Selective large scale screening for lysosomal storage disorders in minority groups shows higher incidence rates
216	Na Lin	The influence of zinc ion in measurement of leukocyte acid sphingomyelinase activity
217	Tatiana Lobry	New interaction between galectin-3 and cystinosin reveals a role of inflammation in kidney pathogenesis in cystinosis
218	Valynne Long	A case report of successful treatment of patient experiencing infusion associated reactions with sebelipase alfa
219	Laura López de Frutos	Discovery study for new genetic variants related to Niemann Pick disease type C
220	Laura López de Frutos	Erythrocyte osmotic resistance test as a screening tool for lysosomal diseases
221	Zoltan Lukacs	Multiplexed testing for Gaucher, Niemann Pick types A/B and acid lipase deficiency
222	Zoltan Lukacs	Evaluation of an investigational product for the measurement of enzyme activity related to lysosomal disorders
223	Jan Lukas	Investigation of ERAD components in GAL A processing and therapeutic targeting of ERAD/proteostasis in Fabry disease
226	Troy Lund	Post-transplant iduronidase attenuates skeletal disease in MPS IH

Wednesday, February 7 – Poster Presentations

Poster #	First Author	Abstract Title
227	Samantha Marcellus	Late development of oculomotor apraxia in a male adolescent with Gaucher disease
228	M. Valerie Marrero-Stein	Diagnostic challenges for Pompe disease newborn screening in a pre-term infant
229	John Marshall	Efficacy of Genz-682452-mediated inhibition of glucosylceramide synthase in a mouse model of Sandhoff disease
231	Ryuichi Mashima	Quantification of enzyme activities of lysosomal disorders in a neonatal population using mass spectrometry
232	Dena Matalon	The diagnosis and natural history of mucopolysaccharidosis type IVA in one family
233	Dietrich Matern	Efficient and effective newborn screening (NBS) for early infantile Krabbe diseases (KD)
234	Dietrich Matern	Precision newborn screening for three lysosomal disorders in Kentucky
236	Blanca Medrano Engay	Multimorbidity in type 1 Gaucher disease patients under miglustat therapy

237	Atul Mehta	Management goals and normalization concept for type 1 Gaucher disease: results from a survey of expert physicians
238	Langis Michaud	5-years longitudinal assessment of ocular manifestations in Fabry patients
239	Langis Michaud	Optic nerve capillaries blood oxygenation investigation in Fabry disease patients
241	Jonathan Mink	The Unified Batten Disease Rating Scale (UBDRS): validation and reliability in an independent sample
242	Jonathan Mink	A proposed staging system for CLN3 disease (juvenile Batten disease)
244	John Mitchell	Presenting signs and symptoms of MPS: results of an international physician survey
245	John Mitchell	Presenting signs and symptoms of MPS: results of a systematic literature analysis
246	Sara Mole	Molecular basis of CLN2 disease: a review and classification of TPP1 gene variants reported worldwide
248	Ken Momosaki	A pilot study of high-risk screening for neuronopathic Gaucher disease in Japan
249	Patricia Moreno	Implementation of internal quality control for the determination of the activity of lysosomal enzymes in dried blood samples
250	Alex Morrison	Patient organizations working in partnership to research the patient experience of rare diseases: the MPS III survey
251	Nina Movsesyan	Three-year evaluation of intravenous and intrathecal treatment with HP-β-CD in a patient with Niemann-Pick disease type C1
252	Juan Mucci	In vitro osteoclast differentiation correlates with bone mineral density in Gaucher disease patients
254	Neslihan Mungan	Cukurova University experience of lysosomal diseases in adulthood: report of 57 patients
255	Neslihan Mungan	A case with Pallister-Killian syndrome misdiagnosed as mucopolysaccharidosis
256	Neslihan Mungan	Early onset alpha-mannosidosis: a Turkish case
257	Neslihan Mungan	Mucopolysaccharidosis type VI, 9 sibling pairs and 1 set of three siblings: single center experience from Turkey
258	Maria Veronica Munoz-Rojas	Awareness of MPS I among cardiologists
259	Laura Murillo	Genetic testing for lysosomal disorders in a commercial laboratory: use of pathognomonic criteria in variant interpretation
260	Simona Murko	Validation of a novel MS/MS-based dried blood assay for the assessment of arylsulfatase B and N-acetylgalactosamin-6-sulfatase activities
262	Murtaza Nagree	In vivo enrichment of transduced cells to enhance gene therapy for Fabry disease
263	Behzad Najafian	Parietal epithelial cells (PEC) in male patients with Fabry neuropathy
264	Behzad Najafian	Podocyte structural parameters predict glomerular filtration rate (GFR) loss in male patients with classic Fabry disease
265	Luba Nalysnyk	Disease epidemiology and prevalence of neurological manifestations in neuronopathic Gaucher disease: comprehensive review of the literature
267	Igor Nestrasil	Longitudinal study of brain volume changes in infantile and juvenile gangliosidoses
268	Igor Nestrasil	Neuroradiological brain phenotype in mucopolysaccharidosis type II patients from 5 European countries
269	Brian Netzel	Expanded analysis of Lyso-GB3 analogues and correlation with total Lyso-GB3 and Fabry status in 59 clinical patients
270	Kathleen Nicholls	Renal outcomes with up to 9 years of migalastat in patients with Fabry disease: results from an open-label extension study
271	Dau-Ming Niu	Identification of lysosomal and extralysosomal globotriaosylceramide (GB3) accumulations in the endomyocardial biopsies before the occurrence of typical pathological changes of the patients with Fabry disease

272	Esther Noël	Interest of the pharmacological chaperone migalastat in the treatment of Fabry Disease
273	Sabrina Nordin	Proposed stages of phenotype development in cardiac Fabry disease: a prospective 182-patient study by cardiovascular magnetic resonance
274	Sabrina Nordin	The subclinical phenotype of cardiac Fabry disease
275	Scott Norton	Development of automated enzymatic assays for NAGLU, GALNS, ARSB and TPP1 for high throughput newborn screening using digital microfluidics
276	Albina Nowak	α-galactosidase activity increases after short-term treatment with migalastat in patients with Fabry disease
277	Albina Nowak	Long-term outcomes of kidney transplantation in Fabry disease
278	Albina Nowak	Pulmonary involvement in Fabry disease: effect of plasma globotriaosylsphingosine (Lyso-Gb3) and time to initiation of enzyme replacement therapy, an observational study
279	Ilyas Okur	Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)
280	Torayuki Okuyama	Investigator-initiated clinical trial of intra-cerebroventricular enzyme replacement therapy for neuronopathic mucopolysaccharidosis type II
282	Sergio Olarte-Avellaneda	In-silico assessment of a potential pharmacological chaperone for human GALNS: evaluation in a mutated protein model
283	Petra Oliva	Integrated approaches for Fabry disease biomarker discovery and qualification
285	Cara O'Neill	Patient advocacy groups and industry collaborate to establish a distinct Sanfilippo syndrome (MPS III) facial phenotype for use in visual diagnostic tool
286	Damara Ortiz	A case of Schindler disease in the setting of familial cardiomyopathy
289	Li Ou	CRISPR-mediated genome editing to treat MPS I mice with AAV vectors
290	Li Ou	Establishing genotype-phenotype correlation of lysosomal diseases with in silico tools
292	Li Ou	Proteomics and interactomics analysis of brain samples from MPS mice and patients
294	William Pardridge	Plasma pharmacokinetics of a human insulin receptor antibody-iduronidase fusion protein in patients with mucopolysaccharidosis type I
295	Samantha Parker	Design, baseline characteristics, and early findings of the MPS IIIA (mucopolysaccharidosis type IIIA) clinical observational study
296	Livia Paskulin	Breastfeeding in Gaucher disease: is taliglicerase-alpha safe?
298	Nita Patel	The patient and clinician point of view: living with late-onset Pompe disease
300	Dawn Peck	Laboratory follow up after abnormal newborn screening for lysosomal disorders
301	Yanyan Peng	Evaluation of a novel, non-invasive iPSC based cell therapy for neuronopathic Gaucher disease
302	Hira Peracha	Epidemiology of mucopolysaccharidoses
303	M. Judith Peterschmitt	Long-term adverse event profile from four completed trials of oral eliglustat in adults with Gaucher disease type 1
304	M. Judith Peterschmitt	Evaluation of glucosyl ceramide synthase (GCS) inhibition for GBA-associated Parkinson's disease
305	Samia Pichard	Limited benefits of presymptomatic cord blood transplantation in neurovisceral acid sphingomyelinase deficiency (ASMD) intermediate type
306	Ana Pinto	Mucopolysaccharidosis type VI and aberrant right subclavian artery: a rare association in a challenging airway
308	Edina Poletto	Geographic distribution and possible origins of most frequent mutations observed in mucopolysaccharidosis type I patients
310	Lynda Polgreen	Defining clinical measures of skeletal disease severity in MPS I
311	Juan Politei	Clinical, serum lyso-Gl3 and kidney histological findings in 14 pediatric patients with classic phenotype of Fabry disease: is it possible a correlation?

312	Juan Politei	Fabry disease: multi-disciplinary evaluation after 15 years of treatment with agalsidase beta
313	Juan Politei	Lessons learned after 5 years of treatment in adult patient with MPS VI. ERT, spinal decompression and home infusion therapy: a triangle for success?
314	Juan Politei	Long term renal function in patients with Fabry disease
315	Juan Politei	Recommendations for evaluation and management of pain in patients with mucopolysaccharidosis in Latin America
316	Laura Pollard	Increased utility of dried blood spots in clinical diagnostic testing
317	Lena Provoost	Cognitive abilities of dogs with mucopolysaccharidosis type I: learning & memory
318	Michael Przybilla	Fetal accumulation of glycosaminoglycan in a mouse model of Hurler syndrome
319	Michael Przybilla	Utilizing CRISPR/Cas9 genome editing to generate a novel murine model of GM1 gangliosidosis
321	Ana Puga	Functional performance in late-onset GM2 gangliosidosis (Tay-Sachs and Sandhoff diseases), longitudinal data over 3 consecutive years
323	Deborah Ramsdell	Treatment of Pompe disease with VAL-1221
324	Prerna Rastogi	Fabry disease presentation in a heterozygote female patient
325	Kimiyo Raymond	Mucopolysaccharides quantitation in serum by liquid chromatography-tandem mass spectrometry
326	Alexander Rodríguez-López	Recombinant n-acetylgalactosamine-6-sulfate sulfatase (GALNS) enzyme produced in pichia pastors as an alternative for Morquio syndrome type A ERT
328	Juan Romero Trejo	Aortic stenosis in Fabry disease patients
329	Alejandra Rozenberg	Combination dosing of CLN1 gene therapy extends lifespan in a mouse model of infantile neuronal ceroid lipofuscinosis
330	Roberto Sandobal Pacheco	Description of a patient with infantile onset Pompe disease after 45 months of enzyme replacement therapy
331	Chelsee Sauni	Pilot enzyme replacement therapy with recombinant human glucosamine (N-acetyl)-6-sulfatase in mucopolysaccharidosis type IIID mouse model
332	Amy Schadewald	Coping strategies, stress, and support needs in caregivers of children with mucopolysaccharidosis
333	Raphael Schiffmann	A 7-year quantitative neurological natural history in mucolipidosis type IV
334	Mathias Schmidt	Anti-drug antibody response in mucopolysaccharidosis type I patients treated with AGT-181, a brain penetrating human insulin receptor antibody-iduronidase fusion protein
338	Scott Selleck	A Drosophila model of multiple sulfatase deficiency
339	Arthavan Selvanathan	Effectiveness of early hematopoietic stem cell transplantation in preventing neurocognitive decline in mucopolysaccharidosis type II: a case series
340	Guillermo Seratti	CLN2 Disease (neuronal ceroid lipofuscinosis type 2): experience in the real world with cerliponase alfa intracerebroventricular enzyme replacement therapy in a public hospital in Cordoba, Argentina
342	Christine Serratrice	Untreated patients with type 1 Gaucher disease: who are they? Preliminary results from the Gaucher non- treated study
344	Volkan Seyrantepe	Abnormal GM2 accumulation alters the function of the autophagic pathway in early-onset Tay-Sachs disease mouse model
345	Elsa Shapiro	A consensus conference for cognitive endpoints for clinical trials and natural history studies in MPS diseases
346	Jin-Song Shen	Priapism in a Fabry disease mouse model is associated with upregulated penile nNOS and eNOS expression
347	Jin-Song Shen	α-Galactosidase A activity modulates DNA methylation of androgen receptor promoter in Fabry disease endothelial cells
349	Daesung Shin	Temporal Galc deletion reveals a critical vulnerable period in the pathogenesis of Krabbe leukodystrophy

350	Morgan Simmons	Identification of a HEXB variant of unknown clinical significance in a family with Sandhoff disease
352	Raj Singh	Demonstration of a digital microfluidic platform for the high throughput analysis of 12 discrete fluorimetric enzyme assays using a single newborn dried blood spot punch
353	Barbara Smith	Compensatory responses to unexpected inspiratory loads are distinct from clinical tests of respiratory capacity
354	Elizabeth Smith	Reclassification of common variants of unknown significance in the hexosaminidase A gene: implications for Tay-Sachs carrier screening
355	Jodi Smith	Novel lysosomal disease in a juvenile golden retriever
356	Barbara Soberon	Combined therapy for type 3 Gaucher disease
357	Barbara Soberon	Bone lesions in type 1 Gaucher disease after improving dose and compliance with imiglucerase
358	Alexander Solyom	Clinical study design for enzyme replacement therapy in acid ceramidase deficiency presenting as Farber disease, an ultra-rare condition with broad phenotypic heterogeneity
360	Rodrigo Starosta	Iron quantification in liver biopsies of patients with Gaucher disease: new method and results
361	Rodrigo Starosta	Gaucheroma mimicking hepatocellular carcinoma in a cirrhotic type I Gaucher disease patient
362	Rodrigo Starosta	Liver disease in a Gaucher disease cohort from southern Brazil: a cross-sectional study
363	Alta Steward	Phenotype-based latent class clustering of GBA1 mutation carriers with and without Parkinson disease
364	David Stockton	Long-term study of growth and development outcomes in patients with infantile-onset Pompe disease receiving alglucosidase alfa: safety data update
365	Katharina Stumpfe	Intrathecal laronidase administration and hematopoietic stem cell transplantation in an 11 months old infant with Hurler syndrome
366	Fernando Suarez-Obando	Spectrum of GBA mutations in a group of Colombian families with Gaucher disease
367	Fernando Suarez-Obando	Genetic and enzymatic studies in a cluster of mucopolysaccharidosis type I patients in northwest Colombia
370	Andrew Talbot	The N215S Fabry disease mutation is not just a “cardiac” variant: strong evidence of renal involvement
371	Ravi Thadhani	Kidney information network for disease research and education (KINDRED) abstract: screening for Fabry disease among United States hemodialysis patients
372	Cynthia Tifft	Clinical outcomes and brain metabolites in patients with late onset Tay-Sachs and Sandhoff disease
373	Shunji Tomatsu	Natural history of Morquio syndrome type A patient with tracheal obstruction from birth to death
374	Shunji Tomatsu	Newborn screening for mucopolysaccharidoses by GAG assay with tandem mass spectrometry
375	Shunji Tomatsu	Novel surgical reconstruction rescues life-threatening severe tracheal obstruction in mucopolysaccharidosis type IVA
376	Shunji Tomatsu	Gene therapy for mucopolysaccharidoses
377	Shunji Tomatsu	Substrate degradation enzyme therapy (SDET) for MPS IVA
378	Shunji Tomatsu	Diagnosis of mucopolysaccharidoses
379	Camilo Toro	Phenotypic characterization of sialidosis type I: a prelude to therapeutic interventions
380	Steven Troy	PK/PD modeling and simulation of recombinant human ASA in patients with metachromatic leukodystrophy: a preliminary evaluation

382	Takahiro Tsukimura	Comparison of GLA variants of unknown significance and the specific mutations causing moderate Fabry disease
383	Coleman Turgeon	Psychosine - a useful biomarker for newborn screening, follow up and monitoring of Krabbe disease
384	Kultigin Turkmen	The 2 year follow-up results of Lyso-GB3 levels in patients with Fabry disease
385	Alfredo Uribe Ardila	Confirmatory assays for alpha-glucosidase enzymatic values using glycogen: an improving test for the diagnosis of Pompe disease
386	Patricia Varela	In silico and in vitro analysis of a novel GLA missense mutation in patients with severe renal involvement
388	Erika Vucko	Outcomes of newborn screening for Gaucher disease in Illinois
389	Susanne Walls	Does infusion time have effect on the efficacy of the enzyme
390	Gustavo Wandalsen	Pulmonary repercussions in patients with Fabry disease
391	Hua Wang	Spectrum of lysosomal disorders at a medical genetics center in the central part of the United States
392	Raymond Wang	Sustained efficacy and safety of vestronidase alfa (rhGUS) enzyme replacement therapy in patients with MPS VII
393	Christoph Wanner	Therapeutic goals in Fabry disease: European expert consensus recommendations based on current clinical evidence
394	David Warnock	Enhanced pharmacokinetics profile of pegunigalsidase alfa (PRX-102) supports once-monthly 2mg/kg dosing for the treatment of Fabry disease
395	Allison Warwick	In Anderson-Fabry disease can early improvement in eGFR predict prognosis over 5 years?
396	Melissa Wasserstein	Systemic improvement in Niemann-Pick disease type B after liver transplantation
398	Nadav Weinstock	GALC ablation in Schwann cells produces a demyelinating peripheral neuropathy characterized by psychosine formation but lacking globoid cells
399	Chester Whitley	Genotype-phenotype correspondence in mucopolysaccharidosis type I and the implications for newborn screening
400	Chester Whitley	Final results of the first-in-human open-label study of intravenous SBC-103 in children with mucopolysaccharidosis type IIIB
401	Shanna Widera	Novel mutations for late onset Fabry disease are being identified from newborn screening in the state of Illinois
402	Miranda Williams	Enhancements to the rare diseases clinical research network contact registry
404	JIll Wood	"Mission: hide and help" - Sanfilippo awareness campaign
405	Michelle Wood	Toe walking in the mucopolysaccharides: does it matter?
406	Michelle Wood	Guided growth surgery for genu valgum in mucopolysaccharidosis type VI: timing is everything
407	Chen Wu	Combination of lysosphingomyelin, 7-ketocholesterol and bile acid W for diagnosis of Japanese patients with Niemann Pick disease type C by MS/MS
408	Su Xu	A next-generation Fabry enzyme replacement therapy: a proprietary human α-galactosidase A co-formulated with a pharmacological chaperone, AT1001, shows greater substrate reduction than standard of care in Fabry mice
409	Hiroyuki Yamakawa	For a patient of unidentified cardiomyopathy, NGS (nest gene sequense analysis) was useful for a diagnosis for Fabry disease
410	Chia-Feng Yang	Very early treatment for infantile-onset Pompe disease contributes to better outcomes: 10-year experience of nationwide NBS in Taiwan
411	Karen Yee	Projected Retained Ability Score (PRAS): a new methodology applied to DAS-II GCA scores for the longitudinal assessment of cognitive abilities in pediatric and adolescent patients with Hunter syndrome
412	Kwangchae Yoon	Optimizing cystine depleting therapy in nephropathic cystinosis: two case reports
413	Sarah Young	Plasma lyso-Gb3 as a diagnostic marker for Fabry disease

415	Natasha Zeid	Characterization of N409H/R535H (N370S/R496H) genotype in type 1 Gaucher disease
416	Mindy Zhang	Transcriptome analysis in muscle biopsies of late-onset Pompe patients treated with alglucosidase alfa or neoGAA
417	Xuling Zhu	Systemic mRNA therapy for the treatment of Fabry disease: preclinical studies in GLA-deficient mice and rats, and wild-type non-human primates
418	Ari Zimran	QCSI (quantitative chemical shift imaging) for assessment of bone marrow involvement in patients with Gaucher disease
LB-03	Lisa Berry	Intraoperative neuromonitoring for improved safety in children with Hurler syndrome undergoing non-spine surgery
LB-04	Lisa Berry	Understanding and managing complexity: surgical experience and anesthesia events in 52 individuals with mucopolysaccaridosis
LB-05	Aline Bochernitsan	GALNS mutations and haplotype study in 68 unrelated South American patients with mucopolysaccharidosis type IVA
LB-06	Mike Broeders	An in vitro model for cartilage defects in MPS VI based on induced pluripotent stem cells
LB-07	Valerie Byers	Caregiver perceptions about neurocognitive testing and factors affecting its accuracy in children with Sanfilippo syndrome (MPS III)
LB-08	Sheng-Kai Chang	Correction of the GLA IVS4+919 mutation with CRISPR deletion strategy in fibroblasts of Fabry disease
LB-09	Pasqualina Colella	Whole-body rescue of Pompe disease with adeno-associated virus vector-mediated liver gene transfer of secretable acid alpha-glucosidase
LB-10	Carley Corado	Exploration of disease biomarkers in a canine model of Krabbe disease
LB-11	Manuela Corti	Immune blockade in CNS gene therapy improves safety and clinical outcome.
LB-12	Mireia del Toro	Enzyme replacement therapy with elosulfase in pediatric Morquio syndrome type A patients: two year follow-up
LB-13	Karina Donis	Early initiation of elosulfase alpha is associated to better outcomes in mucopolysaccharidosis type IVA (MPS IVA).
LB-15	Michael Flanagan	Assessment of heart function in Morquio syndrome type A mice
LB-16	Michael Flanagan	Improvement of enzyme activity in vitro of GALNS-deficient fibroblasts by umbilical mesenchymal stem cell-mediated microvesicles
LB-17	John Gargus	Developing an algorithm to identify suspected cases of cystinosis using electronic health records and an analysis of progression of biomarkers
LB-18	Dominique Germain	Cardiac outcomes with long-term migalastat treatment in patients with Fabry disease: results from phase 3 trials
LB-19	Roberto Giugliani	Spectrum of genetic mutations in patients in the Fabry Outcome Survey
LB-20	Caio Gomes	Plasma Lyso-Gb3 measurement in a family carrying the new GLA variant p.Asn34Asp
LB-21	Simon Heales	Neuronal beta-hexosaminidase and oxidative stress; a further insight into Parkinson disease
LB-22	Nadene Henderson	Pompe and MPS I newborn screening in Pennsylvania - review of our experience
LB-23	Tania Hernández-Mojica	Quality of life in Fabry disease: introduction of a disease-specific tool
LB-24	Ting-Rong Hsu	Demographic characteristics and GLA mutations in Taiwanese patients with Fabry disease: An analysis of data from the Fabry Outcome Survey (FOS)
LB-25	Anita Inwood	A 12 year old male with Sanfilippo syndrome type A (MPS IIIA) who had refractory hip pain due to bilateral avascular necrosis of the femoral heads which required an intra-thecal infusion of bupivacaine and morphine for control
LB-26	Juliette Janson	Distribution to the central nervous system of chemically modified recombinant human sulfamidase after intravenous administration explored in preclinical species
LB-27	Lynne Jesaitis	Immunogenicity bioanalytical methods and assessment of anti−drug antibody impact

LB-28	Shih-hsin Kan	Biochemical evaluation of from-birth intracerebroventricular rhNAGLU-IgF2 enzyme replacement therapy treatment in mice with Sanfilippo syndrome type B
LB-29	Sungmi Kim	Impaired autophagy and mitophagy in MPS disease type II models
LB-30	Virginia Kimonis	Antisense oligonucleotide treatment targeting glycogen synthase (GYS1) in a mouse model of Pompe disease
LB-31	Heather Lau	From clinical trial to real-life practice: long-term analysis of 65 velaglucerase alfa clinical trial patients with Gaucher disease who entered the Gaucher Outcomes Survey (GOS) real-life registry
LB-32	Gwenaël Le Guyader	Mucolipidosis type IV: about 3 cases and review of the literature
LB-33	George Lopez	Developing a stem cell therapy for mucopolysaccharidosis type IIIB using CRISPR/Cas9 technology
LB-34	Maryana Marins	New variants in the MAN2B1 gene in Brazilian population
LB-35	Maryana Marins	Ten years of molecular analysis of mucopolysaccharidosis types I, II and VI genes in Brazilian population
LB-36	R. Scott McIvor	Adoptive transfer of human B cells expressing human iduronidase using the *sleeping beauty* transposon system: correction of lysosomal disease in MPS I mice
LB-37	John Mitchell	Clinical characteristics and response to elosulfase alpha in a large population of patients with Morquio syndrome type A in Quebec
LB-38	Tahseen Mozaffar	Updated results from ATB200-02: a first-in-human, open-label, phase 1/2 study of ATB200 co-administered with AT2221 in adults with Pompe disease
LB-39	Evangelia Ntounousi	Novel pathogenic GLA mutation revealed in a Greek family
LB-40	Carol Ogg	Decreasing the diagnosis odyssey of patients living with Fabry disease via a patient-centered innovative web-based platform
LB-41	Esmee Oussoren	Craniosynostosis occurs in the majority of mucopolysaccharidosis patients
LB-42	William Pardridge	Reduction in brain heparan sulfate with systemic administration of an IgG-Trojan horse-sulfamidase fusion protein in the mucopolysaccharidosis type IIIA mouse
LB-43	Sun Peck	Impaired bone formation in mucopolysaccharidosis (MPS) type VII dogs during postnatal growth is associated with lower serum levels of bone-specific alkaline phosphatase
LB-44	Faruk Pekgül	Targeting metachromatic leukodystophy: subtyping of patients with laboratory diagnosis and diagnosing presymptomatic cases
LB-45	Loren Pena	Neo1 and NEO-EXT: long-term safety of repeat neoGAA (avalglucosidase alfa) dosing in late-onset Pompe disease patients for 3.5 years
LB-46	Federico Pieruzzi	Clinical correlations of native myocardial T1 value in Fabry disease patients without overt cardiac involvement
LB-48	Bryony Ryder	Cardiomyopathy in attenuated alpha-mannosidosis
LB-49	Gerben Schaaf	Reactivation of muscle regeneration activity reverses muscle wasting phenotype in Pompe disease
LB-50	Kristen Skvorak	Improving ERTs with CodeEvolver® protein engineering technology to improve protein stability and in vivo half-life, as well as reduce immune response
LB-51	Barbara Smith	Coordination of breath and stimulator initiation in diaphragm-paced individuals with Pompe disease
LB-52	Sara Smith	Simultaneous measurement of lysosomal enzyme activities associated with MPS II, MPS IIIB, MPS IVA, MPS IVB, MPS VI, MPS VII, and CLN 2 disorders from a dried blood spot via LC-MS/MS
LB-53	Katherine Spurlock	Cognitive outcomes in Hurler syndrome following transplant before age 12 months
LB-55	Joe Trometer	Comparison between Perkin-Elmer QSight® 210md and Waters® Acquity® TQD when measuring six-plex FIA-MS/MS assay of ABG, ASM, GAA, GALC, GLA and IDUA enzyme activities

LB-56	Bethann Valentine	A GalNAc transferase/aspartylglucosaminuria double knockout mouse model experiences rapidly accelerated disease over the single aspartylglucosaminuria knockout model
LB-57	Cláudia Vanzella	Clinical, medical and social aspects of a Brazilian series of 5 MPS I (Scheie syndrome) patients
LB-58	Suresh Vijay	Successful desensitization of infusion associated reactions in infantile onset LAL-D treated with sebelipase alfa
LB-59	Ryuji Yamamoto	Evaluation of JR-051 as a biosimilar to agalsidase beta for Fabry disease in non-clinical studies