Poster Session Abstracts

Poster sessions will be in the Harbor Ballroom on:
Tuesday, March 1 from 4:30-6:30pm
Wednesday, March 2 from 4:30-6:00pm.

No photos or videos are permitted of any oral or poster sessions. The only exception is at the official poster sessions if the author is present and gives permission.

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.

Poster #	First Author	Abstract Title
1	Magy Abdelwahab	Long-term follow up and sudden unexpected death in Gaucher disease type 3 in Egypt
2	Magy Abdelwahab	Ocular abnormalities in Egyptian Gaucher disease patients
3	Walter Acosta	Lectin-mediated delivery of α-L-iduronidase: a novel approach for MPS I enzyme replacement therapy
4	Elma Aflaki	iPSC-derived dopaminergic neurons from patients with Gaucher disease and Parkinsonism demonstrate the potential of a new glucocerebrosidase chaperone
5	Nicholas Agard	Evolving improved therapeutic proteins for treating Fabry disease
6	Patricio Aguiar	Prognostic model for hearing loss in Fabry disease
7	Patricio Aguiar	Urinary type IV collagen: better than albuminuria to identify incipient Fabry nephropathy
8	Alia Ahmed	Association of physical symptom score (PSS) with age and cognitive measures in attenuated mucopolysaccharidosis types I, II and VI
9	Pilar Alfonso	Mutations in the GBA1 gene in a Spanish population with Parkinson's disease and plasma miRNAs
10	Pilar Alfonso	Study of CYP2D6 genotyping in Spanish Gaucher disease type 1 patients
11	Nadia Ali	Attention difficulties and attention deficit / hyperactivity symptoms in adults with Fabry disease
12	Carlos Almeciga	Cell uptake evaluation of human recombinant lysosomal enzymes produced in *Pichia pastoris*
13	Walaa Alshuaibi	Immunomodulation in a Morquio syndrome type A patient treated with enzyme replacement therapy
14	Gheona Altarescu	Rapid noninvasive prenatal diagnosis of autosomal recessive founder mutations by free fetal cell DNA analysis: Gaucher disease as a proof of principle
15	Hernan Amartino	New measure to assess severity of MPS II: the disease severity score
16	Hernan Amartino	Aberrant mongolian spots as a clue to early diagnosis of Hunter syndrome
17	Hernan Amartino	Chagas and Fabry disease comorbidity in Argentinian patient: double threat for heart
18	Gizely Andrade	Multisensory processing in lysosomal disorders: a behavioral and high-density electrophysiology investigation in Niemann-Pick disease type C and cystinosis
19	Marcio Andrade Campos	Follow-up of bone disease in a large cohort of Gaucher disease patients
20	Marcio Andrade Campos	Gaucher disease type 3 in Spain: outcomes and characterization
21	Ida Annunziata	Chaperone mediated gene therapy for sialidosis type I

22	Mika Aoyagi-Scharber	Time- and dose-dependent normalization of pathological lysosomal storage and biochemistry in the mucopolysaccharidosis IIIB (MPS IIIB, Sanfilippo B) mouse model by intracerebroventricular enzyme replacement therapy with BMN 250, a NAGLU-IGF2 fusion protein
23	Carolina Aranda	Laronidase hypersensitivity and desensitization of mucopolysaccharidosis I patient
24	Carolina Aranda	Infusion related hypersensitivity reaction to enzyme replacement therapy for lysosomal diseases
25	Christiane Auray-Blais	Simultaneous analysis of glucosylceramide and galactosylceramide isoforms in mouse and human brain tissue samples using UPLC-MS/MS
26	Stephanie Austin	Three cases of familial pseudodominance in Pompe disease - are current practices missing diagnostic and treatment opportunities?
27	Stephanie Austin	Neuroimaging findings in infantile Pompe patients treated with enzyme replacement therapy
28	Jorge Ayala	Uptake, lysosomal activation, and disease correction in GM1 gangliosidosis cells by plant-made β-galactosidase lectin fusions
29	Anil Bagri	Histologic characterization of the progression of central nervous system pathology in the mucopolysaccharidosis IIIB (MPS IIIB, Sanfilippo B) mouse model and bio-distribution and efficacy of the intracerebroventricular enzyme replacement therapy, BMN 250, a NAGLU-IGF2 fusion protein
30	Anil Bagri	Immunohistochemical analysis of mannose 6-phosphate/insulin-like growth factor 2 receptor in murine wild-type and mucopolysaccharidosis IIIB mutant central nervous system vasculature and implications for trans-blood brain barrier (BBB) transport
31	Lauren Bailey	Late onset Pompe disease case review: severe isolated hypertrophic cardiomyopathy
32	Deeksha Bali	Krabbe and Niemann-Pick disorders: development of novel fluorimetric assays using dried blood spots
33	Bruce Barshop	Leukocyte cystine as a biomarker to monitor cystinosis
34	Michael Beck	Long-term outcomes with agalsidase alfa enzyme replacement therapy: analysis using deconstructed composite events
35	Nadia Belmatoug	The French Gaucher disease registry: clinical characteristics, complications and treatment of 616 patients
36	Lalitha Belur	Gene delivery and biodistribution following intravenous administration of AAV9/rh10 iduronidase in a murine model of MPS type I
37	Lalitha Belur	Intranasal gene delivery of AAV9 iduronidase: a non-invasive and effective gene therapy approach for prevention of neurologic disease in a murine model of mucopolysaccharidosis type I
38	Elfrida Benjamin	The validation of pharmacogenetics in the identification of Fabry patients for treatment with migalastat
39	Naomi Bergin	Unexpected resolution of cardiomyopathy in severe alpha-mannosidosis
41	Kathryn Berrier	Long-term immune tolerance of infantile Pompe disease with entrenched immune responses to ERT using bortezomib-based regimen
42	Lisa Berry	Long term outcome of bone marrow transplantation for Hunter syndrome: a case report
43	Debora Bertholdo	Structural changes in the brain of patients with Gaucher disease
44	Riddhi Bhavsar	Molecular characterization and identification of novel mutations in the PPT1 gene causing neuronal ceroid lipofuscinosis-1 (NCL1) in children from India
45	Meena Bolourchi	Aortic root dilatation in mucopolysaccharidosis
46	Lauren Boudewyn	Assessment of n-butyl-deoxynojirimycin as a therapeutic option for mucolipidosis type IV
47	Allison Bradbury	Natural history study and preliminary assessment of therapies in canine globoid cell leukodystrophy

48	Paiten Brand	Are Fabry disease patients at risk of osteoporosis?
49	Elizabeth Braunlin	Naglazyme® after hematopoietic cell transplant for MPS VI (Maroteaux-Lamy syndrome)
50	Catherine Breen	A facial gestalt in early onset lysosmal acid lipase deficiency - an evolving phenotype
51	Alberto Burlina	Newborn screening for lysosomal diseases: the Italian experience
52	Alessandro Burlina	Basilar artery remodeling in Fabry disease
53	Barbara Burton	Survival in idursulfase-treated and untreated patients with MPS II: data from the Hunter Outcome Survey (HOS)
54	Barbara Burton	Newborn screening for lysosomal diseases in Illinois
55	Vincent Carson	Natural history of metachromatic leukodystrophy
56	Margret Casal	Urinary glycosaminoglycan concentration as a biomarker for effectiveness of enzyme replacement and gene therapy in large animal models of mucopolysaccharidoses
57	Jorge Cebolla	Evaluation of different approaches to lysosomal acid lipase deficiency screening.
58	Jorge Cebolla	Dried blood spot screening of lysosomal acid lipase deficiency and confirmatory studies in Spanish suspected patients
59	magdalena Cerón	Adhesion to the enzymatic replacement therapy in Gaucher disease and the effect on the bone health
60	Fu-Pang Chang	Differential response of glomerular parietal epithelial cells and podocytes to enzyme replacement therapy in Fabry nephropathy
61	Erk Changsila	Gaucher disease is associated with lymph node reactive follicular hyperplasia with tangible body (M2) macrophages
62	Agnes Chen	Intrathecal enzyme replacement for cognitive decline in mucopolysaccharidosis I
63	Raul Chertkoff	Long-term efficacy and safety results of taliglucerase alfa through 5 years in adult treatment-naïve patients with Gaucher disease
64	Yin-Hsiu Chien	Slowly progressive myopathy in neonatal-treated patients with infantile-onset Pompe disease: a longitudinal prospective muscle MRI cohort study
65	Jou-Ku Chung	Biodistribution of idursulfase in cynomolgus monkeys after intrathecal-lumbar administration
66	Heather Church	Successful implementation of plasma oxysterol for screening of Niemann-Pick disease type C in Manchester UK
67	Jose Condori	Novel bioproduction and delivery strategies for MPS IIIA enzyme replacement therapeutics
68	Jonathan Cooper	Progressive neuropathology in the spinal cord of a mouse model of CLN1 disease and assessing the efficacy of intrathecal enzyme replacement therapy (ERT)
69	Jill Corkery	Implementation of newborn screening for lysosomal diseases − a clinician perspective
70	Gerald Cox	Functional performance in patients with late-onset Tay-Sachs and Sandhoff diseases
71	Timothy Cox	Four-year follow-up from the ENCORE trial: a randomized, controlled, non-inferiority study comparing eliglustat to imiglucerase in patients with Gaucher disease type 1 stabilized on enzyme replacement therapy
72	Marco Curiati	Agalsidase beta and infusion-related reactions: findings in Brazilian patients
73	Christine Dali	Intrathecal delivery of recombinant human arylsulfatase A in children with late-infantile metachromatic leukodystrophy
74	Cristin Davidson	Different cyclodextrins for the treatment of Niemann-Pick disease type C
75	Alessandra d'Azzo	Pathogenic cascade downstream of NEU1 regulated lysosomal exocytosis
76	Camila de Aragao	Synaptic dysfunction in Sanfilippo syndrome type C

77	Stephanie DeArmey	Electrocardiograph findings when screening for initiation of oral substrate reduction therapy with eliglustat for treatment of Gaucher disease
78	Stephanie DeArmey	Platelet response to enzyme replacement therapy and oral substrate reduction therapy in an adult with Gaucher disease
79	Patrick Deegan	Risk factors for fracture in imiglucerase-treated Gaucher disease type 1 patients in the ICGG Gaucher Registry
80	Russell DeKelver	ZFN-mediated in vivo genome editing results in supraphysiological levels of human iduronate 2-sulfatase and phenotypic correction in a murine MPS II model
81	Derrick Deming	The molecular basis of Pompe disease: crystal structure of acid alpha-glucosidase
82	Ankit Desai	Cross-reactive immunologic material positive infantile Pompe disease: characterization of immune responses in patient treated with enzyme replacement therapy
83	Robert Desnick	Evolution of cardiac pathology in type 1 classic Fabry disease: progressive cardiomyocyte enlargement leads to increased cell death and fibrosis, and correlates with severity of ventricular hypertrophy
84	Aimee Donald	Unexpected and paediatric death in UK patients with neuronopathic Gaucher disease
85	Chrissa Dwyer	Lysosomal degradation of heparan sulfate is required for normal development of the neural circuitry
86	Berendine Ebbink	Cognitive decline in classic infantile Pompe disease - an underacknowledged challenge
87	Florian Eichler	The role of complement system in GM2 gangliosidoses: implications for Tay-Sachs and Sandhoff disease
88	Julie Eisengart	Outcomes of enzyme replacement therapy in a 14-year-old female with Hurler syndrome
89	Sean Ekins	Virtual collaborations for developing Sanfilippo syndrome treatments on a shoestring
90	N Ellinwood	Twenty-six week or longer intracerebroventricular (ICV) infusion study of BMN 250 administered once every 2 weeks in a canine model of mucopolysaccharidosis type IIIB (MPS IIIB)
91	Deborah Elstein	Therapeutic goals and normal clinical values achieved within 4 years of initiating velaglucerase alfa in treatment-naïve patients with Gaucher disease in phase 3 studies
92	Deborah Elstein	The Gaucher Disease Outcome Survey: description of the population in an ongoing international observational disease registry
93	Deborah Elstein	Taliglucerase alfa during pregnancy for patients with Gaucher disease type 1
94	Deborah Elstein	Children with Gaucher disease type 1: changing profiles in the 21st century
95	Fatma Eminoglu	Mucolipidosis type II (I-cell disease) with pulmonary hypertension and difficult airway
96	Fatma Eminoglu	Case presentation: a girl with cholesterol ester storage disease
97	Niamh Finnigan	Experience of withdrawal of elosulfase alpha (VIMIZIM) from patients at an lysosomal disease referral centre in Northwest England
98	Brona Fletcher	Irish nursing experience of discontinuing enzyme replacement therapy
99	Katherine Foerster	Determination and validation of cutoff score for newborn screening of mucopolysaccharidoses
100	Katherine Foerster	Correlation levels of activities of daily living and disaccharide concentrations in mucopolysaccharidoses
101	José Franco	Screening for mucopolysaccharidoses in patients with short stature of unknown etiology

102	Mark Friedman	Safety findings from 3 trials of treatment with sebelipase alfa in children and adults with lysosomal acid lipase deficiency
103	Haiyan Fu	Functional benefits of systemic rAAV9-HIDS gene delivery in MPS II mouse model
104	Maria Fuller	Manipulation of regional brain bis(monoacylglycero)phosphate in the MPS I mouse by dietary fatty acid supplementation
105	Jose Garcia-Ortiz	Biochemical diagnosis of Sanfilippo syndrome type A and B in a Mexican reference center
106	Michael Gelb	A pilot study for newborn screening of metachromatic leukodystrophy
107	Dominique Germain	Natural history of Fabry disease in male and female patients with the N215S genotype
108	Rasa Ghaffarian	ICAM-1 targeting by direct conjugation enhances gastrointestinal transcytosis and encapsulation enables gastric protection and controlled released for oral enzyme delivery
109	Arunabha Ghosh	Haemophagocytic lymphohistiocytosis and other immunological anomalies in infantile onset lysosomal acid lipase deficiency
110	Arunabha Ghosh	IDUA mutational profile and genotype-phenotype correlations in mucopolysaccharidosis type I
111	Maria Giovinale	Morphological and functional alterations in the non-coronary arterial circulation in Fabry disease
112	Pilar Giraldo	Chronic myeloid leukemia during enzyme replacement therapy in Gaucher disease
113	Luciana Giugliani	Disease duration and survival in Brazilian Niemann-Pick disease type C patients: Preliminary data on potential impact of miglustat
114	Roberto Giugliani	Clinical outcomes in idursulfase-treated patients with MPS II: 3-year data from the Hunter Outcome Survey (HOS)
115	Ozlem Goker-Alpan	Caveolae-mediated uptake of a-galactosidase A in Fabry disease in vitro systems
116	Circe Gómez-Tenorio	Clinical-pathological correlation in Fabry nephropathy in Mexican children based on ISGFN
117	Heather Gray-Edwards	Long term survival after gene therapy in a feline model of Sandhoff disease
118	Martin Grootveld	One- and two-dimensional NMR investigations of the urinary excretion and metabolism of miglustat and valproate in patients with Niemann-Pick disease type C1
119	Anita Grover	Intracerebroventricular administration of BMN 250 to cynomolgus monkeys results in elevated tissue levels and superior biodistribution in the central nervous system in comparison to intravenous delivery
120	Nathalie Guffon	Pregnancy, enzyme replacement therapy and mucopolysaccharidosis: successful outcome
121	Nathalie Guffon	Weekly enzyme replacement therapy: a French patient with infantile onset Pompe disease
122	Allicia Gunderman	OpenIDUA: a novel software for simplifying reports of IDUA variants and their clinical significance in MPS I
123	Sihoun Hahn	52-week efficacy and safety profile of alglucosidase alfa produced at 4000 L scale in US patients with Pompe disease: ADVANCE, a phase 4 open-label prospective study
124	Christine Haller	A novel approach to characterization and categorization of infusion reactions associated with ERT using adverse physiology related groups
125	Sang-oh Han	Minimum effective dose for immune tolerance induction with an adendo-associated virus vector in Pompe disease
126	Paul Harmatz	Impact of elosulfase alfa in patients with Morquio syndrome type A who have limited ambulation: an open-label, phase 2 study

127	Hassan Hazari	Comparison of echocardiography and cardiac magnetic resonance imaging in the determination of left ventricular mass index in Fabry disease
128	Coy Heldermon	MRI findings reveal corollaries in brain pathology between murine and human MPS IIIB brains
129	Christian Hendriksz	Impact of long-term elosulfase alfa treatment on pulmonary function in patients with Morquio syndrome type A
130	Julia Hennermann	Treatment with pentosan polysulphate in patients with mucopolysaccharidosis type I: results from an open label, randomized, monocentric phase 2 study
131	Wendy Heywood	Proteomic analysis of urine reveals potential markers for CLN2 - Batten disease
132	Aki Hietaharju	Adult-onset Niemann-Pick disease type C patients treated with miglustat: Finnish experience
133	Amanda Hodgkins	Impact of social media use in Fabry and Gaucher diseases
134	Robert Hopkin	Improvement of Fabry disease related gastrointestinal symptoms in a significant proportion of female patients treated with agalsidase beta
135	Mia Horowitz	Presence of mutant GBA allele leads to ER stress and development of Parkinson's disease
136	TingRong Hsu	Detection of the first manifestation of the very young children with classical Fabry disease: a study based on newborn screening
137	Derralynn Hughes	Phenotype of Fabry disease in patients with mutations amenable to migalastat
138	Derralynn Hughes	Novel treatment for Fabry disease - IV administration of plant derived alpha-GAL A enzyme safety and efficacy interim report
139	Tim Hutchin	Technical difficulties in the diagnosis of Krabbe leucodystrophy by enzyme analysis
140	Jackie Imrie	Cause of death in patients with attenuated acid sphingomyelinase deficiency: comprehensive literature review and report of new cases
141	Michal Inbar-Feigenberg	Bone marrow transplantation treatment for a 4 year old asymptomatic patient with metachromatic leukodystrophy (MLD)
142	Margarita Ivanova	Autophagy lysosome pathway and mitochondrial crosstalk in Gaucher disease
143	Emanuela Izzo	Expert recommendations for the laboratory diagnosis of neuronal ceroid lipofuscinosis type 2 (CLN2 disease): diagnostic algorithm and best practice guidelines for a timely diagnosis
144	Maryam Jamil	Initial characterization of a murine model of Sanfilippo type IIID shows similar pathology to other Sanfilippo murine models
145	Jeanine Jarnes Utz	Immune tolerization to treat inhibitors to enzyme replacement therapy in Hunter syndrome using a non-immunosuppressing regimen
146	Nicholas Jeffery	Validation of a kinematic assessment of cerebellar dysfunction in the canine model of mucopolysaccharidoses type IIIB (MPS IIIB)
147	Xuntian Jiang	Improved diagnostics for Niemann-Pick disease type C based on a novel bile acid biomarker
148	Zhirui Jiang	Reduced chondrocyte proliferation and hypertrophy contribute to delayed endochondral bone formation in murine mucopolysaccharidosis VII
149	Franklin Johnson	Comparison of integrated white blood cell alpha-galactosidase A activity exposure between every-other-day orally administered migalastat and biweekly infusions of agalsidase beta or agalsidase alfa
150	Simon Jones	Effect of sebelipase alfa on survival and liver function in infants with rapidly progressive lysosomal acid lipase deficiency: 2-year follow-up data
151	Daniel Julien	B- and T-cell immune suppression in a Native American infant with Wolman disease
152	Mustafa Kamani	Reduced glucocerebrosidase activity improves acid ceramidase deficient mice

153	Ilkka Kantola	Enzyme replacement therapy carries for 4-7 years in Fabry disease thereafter quality of life decreases
154	Amel Karaa	Retrospective survey of late onset Tay-Sachs disease
155	Zahra Karimian	Characterizing infusion reactions to intravenous enzyme replacement therapy: establishing temporal relationships for effective management strategies
156	Reena Kartha	Characterization of oxidative stress and inflammation in Gaucher disease type 1: method evaluation and preliminary analysis
157	Zoheb Kazi	Prophylactic immune modulation in infantile Pompe disease using low-dose methotrexate induction: a safe, inexpensive, widely accessible, and efficacious strategy
158	Zoheb Kazi	Differentially expressed proteins in infantile Pompe disease: prediction of patients likely to mount an immune response to enzyme replacement therapy
159	Katherine Keating	Pseudodeficiency of alpha-iduronidase is a common finding identified from newborn screening in the state of Illinois
160	Richie Khanna	Co-administration of the pharmacological chaperone AT2221 with a proprietary recombinant human acid alfa-glucosidase leads to greater plasma exposure and substrate reduction compared to alglucosidase alfa
161	Tammy Kielian	Adeno-associated virus 9 gene therapy for juvenile neuronal ceroid lipofuscinosis
162	Bridget Kiely	Natural history of Hurler syndrome
163	Kelly King	Emotional-behavioral functioning in individuals with MPS I: a longitudinal approach
164	Thomas Kirkegaard	Development of heat shock protein based therapies for lysosomal diseases
165	Priya Kishnani	Clinical characteristics and genotypes in the ADVANCE baseline dataset, a comprehensive cohort of US children and youth with Pompe disease
166	Francyne Kubaski	Bone mineral density in Morquio syndrome type B
167	Francyne Kubaski	Hematopoietic stem cell transplantation and ERT for Hunter syndrome
168	Francyne Kubaski	Newborn screening for mucopolysaccharidoses: a pilot study of 2640 samples
169	Francyne Kubaski	Hematopoietic stem cell transplantation in Morquio syndrome type A
170	Dawn Laney	The impact of Fabry disease on reproductive fitness
171	Kanut Laoharawee	AAV9 mediated correction of iduronate-2-sulfatase deficiency in the central nervous system of mucopolysaccharidosis type II mice
172	Yvonne Latour	Development of isogenic human cerebral organoids with beta-galactosidase deficiency
173	Heather Lau	Enzyme replacement therapy with investigational rhGUS in an infant with non-immune hydrops fetalis and mucopolysaccharidosis type VII
174	Heather Lau	Evidence of early bone response after initiation of enzyme replacement therapy in a 3 year old patient with MPS VII
175	Heather Lau	Wandering spleen in 2 Gaucher patients on enzyme replacement therapy: diagnosis and management
176	Jonathan LeBowitz	Utilizing activity assays and population-wide allele frequencies to assess the contribution of novel mutations in NAGLU to MPS IIIB incidence
177	Rebecca Lehmann	Accumulation of MPS GAG influences stem cell differentiation
178	Malte Lenders	Fabry patients after enzyme replacement therapy dose reduction and treatment switch - renal impairment after 24 months follow-up
179	Malte Lenders	Antibody-mediated inhibition of enzyme replacement therapy in Fabry Disease results in impaired cardiac and renal function
180	Yedda Li	Combination therapy increases lifespan and improves clinicobehavioral performance in the murine model of globoid cell leukodystrophy

181	Olivier Lidove	Clinical characteristics and demographics in patients 50 years and older in the Fabry Outcome Survey (FOS)
182	Renuka Limgala	Selective large scale screening for lysosomal disease in minority groups shows higher incidence rates
183	Emily Lisi	Genetic counseling dilemma in neuronal ceroid lipofuscinosis associated with variants of unknown significance in whole exome sequencing: a case report
184	Valynne Long	Baffling brain MRI findings in a patient with Gaucher disease
185	Laura López de Frutos	Evaluation of suspicion index and plasma biomarkers as efficient tool in the diagnosis of Niemann-Pick disease type C
186	Laura López de Frutos	Descriptive report of the variant adult visceral form non-neuronopathic of Niemann Pick disease type C in a Spanish series
187	Lucia Lopez-Vasquez	Acid ceramidase deficiency leads to multiple skin abnormalities in a mouse model of Farber disease
188	Eda Machado	Regulated lysosomal exocytosis mediates cancer progression
189	Sarah Macklin	Examining the psychosocial impact of carrying the p.A143T variant in the GLA gene
190	Gustavo Maegawa	Screen and identification of small molecules therapies to reduce elevated psychosine levels in globoid-cell leukodystrophy
191	Alexandros Makis	Gaucher disease and heterozygous beta-thalassemia: a rare case of coinheritance
192	Raffaele Manna	The impact of fever and dystermia in Fabry diagnosis: a retrospective analysis
193	Rachel Manthe	Enhanced lysosomal enzyme delivery across the blood-brain barrier by modulating the valency of ICAM-1-targeted nanocarriers
194	Ana Martins	Alternative laronidase dose regimen for patients with mucopolysaccharidosis I
195	Ryuichi Mashima	Rapid detection system of glycosaminoglycans with small amount of urine samples for high-risk screening of mucopolysaccharidoses
196	Francesca Mazzacuva	Identification of new biomarkers suitable for an early diagnosis of Niemann-Pick disease type C1
197	Kim McBride	Design of a phase I/II gene transfer clinical trial of rAAV9.CMV.hNAGLU for mucopolysaccharidosis type IIIB
198	Aaron Meadows	Functional correction of mucopolysaccharidosis I in adult mice by a systemic rAAV9-IDUA gene delivery
199	Blanca Medrano Engay	Deficiency of intestinal disacharidases and miglustat in Gaucher disease patients
200	Olga Meijer	Quantity and structure of stored heparan sulfate may affect the nature and course of neuronopathic disease in mucopolysaccharidosis type I and mucopolysaccharidosis type III
201	Björn Mellgard	Effects of velaglucerase alfa on bone-related pathology in patients with Gaucher disease type 1: design and methods of a 2-year open-label phase 4 study
202	Christina Mikulka	Reducing neuroinflammation in globoid cell leukodystrophy by targeting the JAK/STAT pathway
203	Bradley Miller	Longitudinal changes in linear growth and BMI in the mucopolysaccharidoses
204	James Miller	The α-galactosidase A-deficient rat: characterization of a new animal model of Fabry disease
205	Kaitlyn Miller	Factors that influence an individual's or parent's willingness to follow a prescribed treatment plan for Fabry disease
206	John Mitchell	Farber disease: Implications of anti-inflammatory treatment
207	Derek Moen	Uptake and lysosomal delivery of recombinant human alpha-n-acetylglucosamine-6-sulfatase to mucopolysaccharidosis IIID fibroblasts

208	Ken Momosaki	2-year experience of newborn screening of Pompe disease in a Japanese region
209	JOSE MONTE NETO	Fabry disease and membranous nephropathy: case report
210	Mari Mori	Identification of modifier genes of Pompe disease phenotype by variant analysis of whole exome sequencing data
211	Joseph Muenzer	A phase II/III intrathecal enzyme replacement therapy clinical trial for MPS II patients with cognitive impairment
212	Simona Murko	Multiplexed enzyme testing as differential diagonosis in cases of suspected Gaucher disease - Niemann-Pick disease A/B
213	Simona Murko	Possibilities for an early diagnosis of CLN2 disease
214	Nicole Muschol	Clinical outcome following hematopoietic stem cell transplantation in two Hunter patients
215	Murtaza Nagree	Differential acyl chain storage of multiple glycosphingolipids in Fabry mice
216	Behzad Najafian	Podocyte globotriaosylceramide (GL-3) content in male adult patients with Fabry disease reduces following 6-12 months of treatment with migalastat
217	Kimitoshi Nakamura	Find-GEMS (Gaucher disease in Epilepsy and Myoclonus): screening for Gaucher disease with enzyme assay among patients with early-onset seizures
218	Aya Narita	pH-responsive pharmacological chaperones for lysosomal disease
219	Juana Navarrete	Follow-up of lysosomal disease patients detected by neonatal screening
220	Igor Nestrasil	Brain MRI patterns in MPS IIIB (Sanfilippo syndrome type B): longitudinal study
221	Stephanie Newman	Impaired Fc-gamma and complement receptor mediated phagocytosis in Niemann-Pick disease type C (NPC) macrophages
222	Dau-Ming Niu	Revisited later-onset cardiac type Fabry disease - cardiac damages progressed in silence - experiences from an extremely high prevalent area, Taiwan
223	Torayuki Okuyama	Phenotype-genotype correlation in Japanese patients with mucopolysaccharidosis type II
224	Aida Oliván-Viguera	Reduced KCa3.1 channels functions in monocytes/macrophages in Gaucher disease
225	Neslihan Onenli Mungan	Chanarin Dorfman syndrome: a case report
226	Kenji Orii	Safety study of sodium pentosan polysulfate for adult patients with mucopolysaccharidosis type II
227	Li Ou	Enzyme replacement therapy with α-l-iduronidase and lectin RTB fusion protein in treating murine Hurler syndrome
228	Li Ou	Selection of the optimal lentiviral vector for treating mucopolysaccharidosis type I
229	Li Ou	ZFN-mediated correction of murine MPS I model by expression of the human IDUA cDNA from the albumin “safe harbor” locus
230	Pablo Ovando-Seymour	The c.253G>A mutation in Fabry disease: evidence of pathogenicity in a Mexican male patient
231	A Pal	Sleep disordered breathing in treated mucopolysaccharidosis I patients correlates with worsening metabolic biomarkers and inhibitory antibodies
232	Manoj Pandey	Immune cells attack and neurodegeneration in Gaucher disease
233	Rebecca Parsons	Use of the T-maze to assess cognition in normal and MPS IIIB affected dogs
234	Rebecca Parsons	Use of a radial arm maze to assess cognition in normal and MPS IIIB affected dogs
235	Livia Paskulin	The impact of taliglucerase alfa in the bone of Gaucher disease type 1
236	Sarah Pass	The impact of treatment availability on reproduction in Fabry disease

237	Gregory Pastores	Insight into the pre-diagnosis period of 212 patients with Gaucher disease: results of the OnePath® US patient survey
238	Loren Pena	Phase 1 exploratory efficacy of the novel enzyme replacement therapy neoGAA in treatment-naïve and alglucosidase alfa-treated late-onset Pompe disease patients
239	Iliana Peña-Gomar	c.1898C>G/p.Ser633Trp IDUA mutation cause an intermediate to severe phenotype in mucopolysaccharidosis type I patient: clinical and computational approach
240	Ester Pereira	Generation of Fabry disease kidney cell lines using genome editing by CRISPR/Cas9
241	Ester Pereira	Podocyturia in Fabry disease
242	Ester Pereira	Exploring signaling pathways in a novel kidney cell model of Fabry disease
243	Vanessa Pereira	Low alpha-galactosidase A activity among haemodyalisis patients: does it correlate with Fabry disease mutations?
244	Jordi Pérez-López	Bone quality in patients with Gaucher disease type 1: validation and preliminary results with microindentation
245	Joao Pesquero	A Brazilian profile of GLA gene mutations - a review of ten years experience with diagnosis of Fabry disease
246	M. Judith Peterschmitt	Evaluation of glucosylsphingosine as a biomarker of the eliglustat treatment response in patients with Gaucher disease type 1 (GD1)
247	Dawn Phillips	Functional outcome measures in pediatric therapeutic intervention: application and issues in a rare disease
248	Luis Pineda-Galindo	Beneficial effect of agalsidase beta on long term evolution of patients with Fabry disease and kidney transplant
249	Lynda Polgreen	High bone turnover is associated with lower bone density in Hurler-Scheie and Hunter syndromes treated with enzyme replacement therapy
250	Juan Politei	Recommendations for enzyme replacement therapy in classical phenotype of Fabry disease in Latin America
251	Giulia Polo	Lysosphingolipids in dried blood spots as biomarkers for lysosomal diseases
252	Giulia Polo	High level of oxysterols in neonatal cholestasis: a pitfall in analysis of biochemical markers for Niemann-Pick disease type C
253	Forbes Porter	Phase 1/2 evaluation of intrathecal 2-hydroxypropyl-β-cyclodextrin for the treatment of Niemann-Pick disease, type C1
254	Anna Potier	Multiplex MS/MS method to measure MPS II, MPS IIIB, MPS IVA, MPS VI enzyme activities in dried blood spots
255	Vincent Puy	Alteration of cerebral iron metabolism in Sanfilippo syndrome
256	Xueying Qiao	Lower extremity edema is common in Fabry disease and diuretics are not an effective treatment
257	Nina Raben	Pompe disease: from pathophysiology to therapy and back again
258	Uma Ramaswami	Cardio-renal outcomes with long-term agalsidase alfa enzyme replacement therapy: a 10-year Fabry outcome survey analysis
259	Uma Ramaswami	Vitamin D deficiency in Fabry disease: a single centre, retrospective review
260	Dallas Reed	Clinical diagnosis of Gaucher disease type 3 with a rare genotype
261	Matthew Reed	Potential roles of glucosylceramide and glucosylsphingosine in bone metabolism and multiple myeloma
262	Ricardo Reisin	Looking for predictors of mortality in patients with Fabry disease on enzyme replacement therapy
263	Nathan Rodgers	Thirty year follow-up in Hurler syndrome after hematopoietic cell transplantation - the University of Minnesota experience
264	Adeel Safdar	Exosome-mRNA (EXERNA) therapy for Pompe disease

265	Hitoshi Sakuraba	Comparative study on α-galactosidase A (GLA) genetic variants with unknown clinical significance
266	Gabriela Salas-Pérez	The risks of positive findings while screening for Fabry disease in high risk populations
267	Adalberto Sanchez	Transitional mutations cluster C>T and T>C on a Colombian population affected with mucopolysaccharidosis
268	Adalberto Sanchez	Exome sequencing thechnology for diagnosis of mucopolysaccharidosis in Colombian populations
269	S. Pablo Sardi	Glucosylceramide synthase inhibition reduces α-synuclein pathology and improves cognition in murine models of synucleinopathy
270	Andreas Schaaf	Moss-aGal: preclinical evaluation of a plant made enzyme replacement for Fabry disease
272	Raphael Schiffmann	A 5 year neurological natural history of mucolipidosis type IV
273	Edward Schuchman	Cartilage and bone disease in a mouse model of Farber lipogranulomatosis and response to treatment
274	Angela Schulz	Expert opinion on the management of CLN2 disease
275	C. Ronald Scott	The performance characteristics of a 6-plex assay for the detection of six lysosomal diseases and preliminary data for the detection of MPS II, MPS IVA and MPS VI
276	Volkan Seyrantepe	Deletion of sialidase NEU3 causes progressive neurodegeneration in Tay-Sachs mice
277	Suma Shankar	Eye findings in Fabry disease and correlation with disease severity
278	Elsa Shapiro	Importance of common data elements (CDEs) for rare disease clinical trials
279	Emily Shawgo	Psychosine as a biomarker for Krabbe disease
280	Adam Shaywitz	Design and rationale of the study programs for BMN 250, a novel enzyme replacement therapy (ERT) for Sanfilippo syndrome type B
281	Jin-Song Shen	Sortilin expression and uptake of α-galactosidase A: a general mechanism of endocytosis in Fabry disease cell types
282	Calogera Simonaro	Pentosan polysulfate and neuroinflammation in mice with mucopolysaccharidosis type IIIa
283	Serap Sivri	Two cases with mucopolysaccharidosis type VII
284	Elizabeth Smith	An examination of the current metabolic education curriculum across genetic counseling training programs in the United States and Canada
285	Alexander Solyom	Farber disease is characterized by typical features but a broad phenotypic spectrum: selected information from a cohort of 37 patients
286	Archana Soni-Jaiswal	Attenuated mucopolysaccharidosis II; parental beliefs about the impact of disease on the quality of life of their children
287	Archana Soni-Jaiswal	Hearing loss in mucopolysaccharidosis and its impact on quality of life; a review of the literature
288	Archana Soni-Jaiswal	Mucopolysaccharidosis I; parental beliefs about the impact of disease on the quality of life of their children
289	Rebecca Southall	One year data from the Space4u2talk Emotional Wellbeing Programme
290	Srividya Sreekantam	Clinical features and outcomes in multiple sulfatase deficiency: a single centre experience
291	Richard Steet	Cathepsin-mediated alterations in TGF-β related signaling underlie the cartilage and bone defects associated with impaired lysosomal targeting
292	Fiona Stewart	Pregnancy in individuals with mucopolysaccharidosis: a case series
293	Fiona Stewart	Management of fertility and pregnancy in individuals with mucopolysaccharidosis (MPS)
294	Kanagaraj Subramanian	Quantitative analysis of the proteome response to histone deacetylase inhibitor in Niemann-Pick disease type C1

295	Baodong Sun	New perspectives for ERT in Pompe disease: extending the action of the enzyme to cytosolic targets
296	Stefan Svensson Gelius	Reversal of lysosomal storage and inflammation in brain of adult MPS IIIA male mice with intravenous glycan modified sulfamidase
297	Gyani Swift	Swallow prognosis and follow up protocol in infantile Pompe disease
298	Azza Tantawy	Velaglucerase alfa enzyme replacement therapy in children and adolescents with Gaucher disease type 3: results of a 12-month multicenter, open-label phase 1/2 study
299	Mark Tarnopolsky	Exosome-mRNA therapy for Gaucher disease
300	Mark Tarnopolsky	Exosome-mRNA and exosome-protein therapy for Niemann-Pick disease type C
301	Beth Thurberg	Meningioma in Fabry disease: three case reports and a review of the literature on malignancies in patients with metabolic lipid disorders
302	Tadayasu Togawa	Measurement of glycosphingolipids by means of tandem mass spectrometry and evaluation of them as a biomarker of Fabry disease
303	Shunji Tomatsu	Morquio syndrome type A airway and anesthetic considerations
304	Shunji Tomatsu	Activity of daily living for Morquio syndrome type A
305	Shunji Tomatsu	Obstructive airway in mucopolysaccharidosis IVA
306	Shunji Tomatsu	Hematopoietic stem cell transplantation for Morquio syndrome type A
307	Shunji Tomatsu	Surgical reconstruction for severe tracheal obstruction in Morquio syndrome type A
308	Amanda Toupin	Biomarker analysis of Fabry patient cell fractions using tandem mass spectrometry
309	Kristen Truxal	Prospective natural history study of mucopolysaccharidosis types IIIA and IIIB (Sanfilippo syndrome)
310	Takahiro Tsukimura	Anti-α-galactosidase A antibodies and serum-mediated inhibition in Fabry disease
311	Coleman Turgeon	Combined analysis of glucosylsphingosine, lyso-sphingomyelin, cholestane-3β,5α,6β-triol, and 7-ketocholesterol in plasma for Gaucher and Niemann-Pick disease types A, B and C
312	Karen Tylee	Screening for Fabry disease using dried blood spots in clinically relevant patient populations over six years
313	Anna Tylki-Szymańska	Characteristics of patients with MPS II diagnosed at a very young age: data from the Hunter Outcome Survey (HOS)
314	Anna Tylki-Szymańska	Molecular analysis of 22 patients with mucopolysaccharidosis IVA from Poland, Belarus and Kazakhstan identifies 6 novel GALNS mutations
315	Geoffrey Urbanski	Pigmentary retinopathy as first manifestation in two cases of Scheie syndrome
316	Filippo Vairo	CCL22 chemokine and platelets in Gaucher disease
317	Vassili Valayannopoulos	Obstructive sleep apnea syndrome after hematopoietic stem cell transplantation in children with mucopolysaccharidosis type I
318	Ans van der Ploeg	Phase 1 safety and pharmacokinetics of the novel enzyme replacement therapy neoGAA in treatment-naïve and alglucosidase alfa-treated late-onset Pompe disease patients
319	Nato Vashakmadze	Rare defect of plasma hemostasis in patients with mucopolysaccharidosis
320	Elena Verrecchia	Globotriaosylsphingosine (lyso-GB3) as useful marker for monitoring initial therapeutic outcomes of enzyme replacement therapy for Fabry patients
321	My Linh Vu	Patient iPS-derived neural stem cells and neuronal cells as a cell-based model system for Tay-Sachs disease
322	Erika Vucko	Role development for nurse practitioners as primary clinician for the initial evaluation for an abnormal newborn screen

323	Susanne Walls	Delivering care to the patients - satisfaction among the Finnish Fabry patients
324	Crista Walters	Enzyme replacement therapy rate escalation in infantile onset Pompe disease
325	David Warnock	Male patients with Fabry disease treated with enzyme replacement therapy: renal progression rates reflect averaged urine protein to creatinine ratios
326	David Warnock	Anti-proteinuric therapy and Fabry nephropathy; factors associated with preserved kidney function during agalsidase-beta therapy
327	Connie Wehmeyer	A single center's experience with hematopoietic stem cell transplant in MPS IH from 2002-2015: a retrospective review of variables that may influence transplant outcome and complications
328	Marie Wencel	Utility of routine respiratory function testing in a tertiary neuromuscular clinic: a 10-year experience
329	David Whiteman	Insights into the management of patients with MPS II: key findings following 10 years of the Hunter Outcome Survey (HOS)
330	Alison Wilson	The experiences and information requirements of women with mucopolysaccharidosis (MPS) or related condition during pregnancy, birth and the postnatal period
331	Tim Wood	Measurement of urinary glycosaminoglycans via UPLC-MS/MS provides increased sensitivity for the diagnosis and therapeutic monitoring of MPS patients
332	Gouri Yogalingam	Evaluation of myeloperoxidase as a targeted enzymatic approach for the elimination of retinal A2E in Stargardt disease
333	Gouri Yogalingam	Glycosylation independent lysosomal targeting of alpha-n-acetylglucosaminidase confers highly efficient enzyme uptake into critical cellular targets of disease pathogenesis in mucopolysaccharidosis type IIIB
334	Wen-Chung Yu	A Fabry Outcome Survey (FOS) analysis of cardiac biomarkers and left ventricular hypertrophy in Taiwanese patients with the Chinese hotspot IVS4+919G>A mutation or classical Fabry mutations
335	Aysel Yuce	Niemann-Pick disease type C in the newborn period: reflections from 10 patients
336	Haoyue Zhang	Quantification of dermatan sulfate and heparan sulfate in cerebrospinal fluid using liquid chromatography-tandem mass spectrometry for therapeutic monitoring of patients with mucopolysaccharidoses
337	Wen Zhang	Clinical, biochemical and molecular analysis of five Chinese patients with Sandhoff disease
338	Ari Zimran	Significant and continuous improvement in quantitative chemical shift imaging (QCSI) in patients with Gaucher disease treated with taliglucerase alfa during the early access program
LB-01	Daniel Bichet	Persistence of positive renal and cardiac effects of migalastat in Fabry patients with amenable mutations following 30 months of treatment in the ATTRACT study
LB-02	Stephanie Cagle	Clinical trials for the AGT-181 and AGT-182 drugs in adults with mucopolysaccharidosis (MPS): recruitment challenges and solutions
LB-03	Jessica Cohen Pfeffer	Expert opinion on the management of intracerebroventricular (ICV) drug delivery
LB-04	James Davison	Use and utility of neuroimaging in paediatric mucopolysaccharidosis type 2 (Hunter syndrome): single-centre review of current practice
LB-05	Kristin Dorfman	Projecting the prevalence of lysosomal storage disorders over 2015-2025
LB-06	Scott Garman	An engineered monomeric variant of human α-galactosidase
LB-07	Roberto Giugliani	A fifteen-year perspective of the Fabry Outcome Survey (FOS)

LB-08	Cyril Goizet	Atypical presentations of pain in Fabry disease
LB-09	Olga Gundobina	Bone complications in child with Gaucher disease
LB-10	Alaa Hamed	Patient and caregiver experience with late-onset Tay-Sachs and Sandhoff diseases
LB-11	Virginia Kimonis	Variable clinical features in Pompe disease associated with novel mutations
LB-12	Julia Kofler	Neuropathologic findings in two long-term survivors of Krabbe's disease with and without umbilical cord transplantation
LB-13	Vish Koppaka	Recombinant human protective protein/cathepsin A: an update on the development of an enzyme replacement therapy for galactosialidosis
LB-14	Roger Lawrence	New chondroitin sulfate derived non-reducing end (NRE) biomarker for the diagnosis and measurement of therapeutic response in Morquio syndrome type A (MPS IVA)
LB-15	Ulrike Löbel	Brain volumetry and clinical scoring in patients with CLN2 disease: an objective tool to monitor disease progression
LB-16	Charles Lourenco	Different presentations of lysosomal acid lipase deficiency across ages: unraveling the clinical phenotypes of an under-recognized genetic disorder
LB-17	Vagishwari Murugesan	The utility of lysoglucocerebroside as a pathophysiologically relevant biomarker of Gaucher disease
LB-18	Leyla Namazova-Baranova	Total Fabry disease screening among dialysis centers in Russian Federation
LB-19	Barbara Natke	Development and validation of a cross-reactive immunologic material assay for mucopolysaccharidosis II (Hunter syndrome)
LB-20	Miriam Nickel	Natural history of CLN2 disease: quantitative assessment of disease characteristics and rate of progression
LB-21	Angela Schulz	Intracerebroventricular cerliponase alfa (BMN 190) in children with CLN2 disease: interim results from a phase 1/2, open-label, dose-escalation study
LB-22	Daesung Shin	Trafficking and processing of GALC mutants in globoid cell leukodystrophy regulates disease severity and is modulated by cis-polymorphisms
LB-23	Lachlan Smith	Impaired WNT signaling contributes to delayed chondrocyte differentiation in mucopolysaccharidosis VII dogs
LB-24	Lachlan Smith	Progression of vertebral bone disease in mucopolysaccharidosis VII dogs from birth to skeletal maturity
LB-25	Dean Suhr	RUSP Roundtable examines perspectives and potential changes for newborn screening
LB-26	Ying Sun	Modulating ryanodinereceptors by dantrolene attenuated neuropathic phenotype in Gaucher disease mice
LB-27	Kazuya Tsuboi	Efficacy of enzyme replacement therapy with agalsidase alfa in 36 naïve Fabry disease patients
LB-28	Melissa Wasserstein	Long-term safety and efficacy of olipudase alfa in patients with acid sphingomyelinase deficiency (ASMD)
LB-29	Christine White	Gaucher disease diagnostic algorithm for health practitioner in Canada
LB-30	Miao Xu	Tocopherolenters cell through the endocytic pathway and regulates lysosomal exocytosis
LB-31	Tatsuyoshi Yamamoto	PK study with novel recombinant alpha-galactosidase A (jr-051) as a biosimilar to Fabrazyme in healthy Japanese volunteers
LB-32	Angela Zanette	Gaucher disease: endemic diseases as differential diagnosis in Bahia, Brazil
LB-33	Chester B. Whitley	Initial, 24 week results of heparan sulfate levels in cerebrospinal fluid (CSF) and serum in an open label, phase I/II, first-in-human clinical trial of intravenous SBC-103 in mucopolysaccharidosis IIIB