Poster Session Abstracts

Poster sessions will be in the *Regency Ballroom S* held on:
Tuesday, February 10 from 4:30-6:30pm
Wednesday, February 11 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 (click to download).

Abstracts submitted prior to October 1, 2014 will be published in the February 2015 special "Lysosomes Issue" of Molecular Genetics and Metabolism (MGM). Articles and abstracts from this issue can be purchased individually from Elsevier. The journal is typically available in late January or early February. Registered attendee's will receive an electronic copy of the program and abstracts when they check in at the WORLD*Symposium* 2015 registration desk.

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Poster #	First Author	Abstract Title
1	Magy Abdelwahab	Abnormal behavioral features in Egyptian children with type III Gaucher disease
2	Magy Abdelwahab	Splenectomy in eight Egyptian patients with type III Gaucher disease: an 8 year prospective study
3	Alia Ahmed	Is somatic disease burden associated with brain disease phenotype in MPS I?
4	Suhail Alam	A new formulation for the treatment of neurological and systemic defects in Niemann-Pick disease type C
5	Taciane Alegra	Is melanogenesis disturbed in mucolipidosis II/III? A multicenter study based on clinical and genetic findings
6	Nadia Ali	Psychological health in adults with Morquio Syndrome
7	Einat Almon	Novel treatment for Fabry disease, IV administration of plant derived alpha-GAL-A enzyme phase 1/2 safety and efficacy study: interim clinical report
9	Marcio Andrade-Campos	Multiple myeloma and Gaucher disease share features of a cytokine profile
10	Carolina Aranda	Immunology of mucopolysaccharidosis
11	Andrea Atherton	Newborn screening for Fabry disease: is the A143T allele a pathogenic mutation or a pseudodeficiency allele
12	Andrea Atherton	The first two years of full population pilot newborn screening for lysosomal disorders: the Missouri experience
13	Christiane Auray-Blais	Galabiosylceramide isoforms/analogues as biomarkers for Fabry disease patients
14	Christiane Auray-Blais	Mass spectrometry multiplex analysis of urinary glycosaminoglycans for mucopolysaccharidose patients
15	Christiane Auray-Blais	Urine keratan sulfate (uKS) elevation in lysosomal disorders: comparison of uKS levels in Morquio/MPS IV versus non-Morquio lysosomal disorder
16	Stephanie Austin	Further expanding the phenotype of treated infantile onset Pompe disease
17	Lauren Bailey	Avascular necrosis in neuronopathic Gaucher despite high-dose enzyme replacement therapy
18	Laurie Bailey	Combination therapy (eliglustat + velaglucerase alfa) in a pediatric patient with Gaucher disease type 1 and hereditary spherocytosis
19	Guilherme Baldo	Elosulfase alfa decreases glycosaminoglycan storage in white blood cells from Morquio syndrome type A patients undergoing enzyme replacement

20	Edgar Barajas	Osteomyelitis in a breastfed child with Gaucher disease type I with an indistinguishable bone crisis
21	Miguel Barba-Romero	Fabry disease in untreated women with enzyme replacement therapy: symptomatology and clinical profile
22	Spyros Batzios	Alterations in gait pattern in Hunter disease patients undergoing enzyme replacement therapy as assessed by the GaitRite system: a six year follow up
23	Michael Beck	Long-term effectiveness of agalsidase alfa enzyme replacement in Fabry disease: a Fabry Outcome Survey analysis
24	David Bedwell	The nonsense suppression drug PTC124 restored alpha-l-iduronidase activity and reduces glycosaminoglycan accumulation in MPS IH mice carrying the Idua-W402X mutation
25	Peter Bell	Evaluating the impact of systemic AAV9.cIDUA administration on brain pathology in MPS I dogs
26	Nadia Belmatoug	Skeletal involvement in Gaucher disease at MRI: what long-term evolution can we expect under enzyme replacement therapy.
27	Marie-Françoise Ben Dridi	Clinical characteristics of type III Gaucher disease in children and adolescents enrolled in a trial of velaglucerase alfa
29	Lisa Berry	Support for siblings of children with lysosomal disorders
30	Debora Bertholdo	Correlation between brain MR spectroscopy and BMB score in type 1 Gaucher disease: is there any?
31	Marieke Biegstraaten	Consensus recommendations on initiation and cessation of enzyme replacement therapy in patients with Fabry disease
32	Olaf Bodamer	Correlation between birth weight, disease severity and outcomes in patients with Hunter syndrome: data from the Hunter Outcome Survey (HOS)
33	Daniel Borger	Impaired autophagy leads to inflammasome activation and a heightened inflammatory profile of macrophages in Gaucher disease
34	Noelle Brackett	The impact of bone marrow transplant treatment for mucopolysaccharidosis type IH on family functioning: social, emotional and financial factors
35	Alexander Broomfield	The UK experience of enzyme replacement therapy in patients with infantile onset Pompe disease
36	Anne-Sophie Brouard-Lapointe	Rare diseases and patient organization collaboration in the medical research: analysis of the issues with all the protagonists
37	Dakota Buhrman	Longitudinal assessment of autistic behaviors in children with Sanfilippo syndromes types A and B
38	Derek Burke	Lysosomal β-glucosidase (GBA1) and non-lysosomal β-glucosidase (GBA2), potential involvement in the pathogenesis of Gaucher disease/Parkinson disease
39	Thomas Burrow	Description of later onset presentations of neuronal ceroid lipofuscinosis due to mutations in CTSD
40	Barbara Burton	Results of a global phase 3, randomized, double-blind, placebo-controlled trial evaluating the efficacy and safety of sebelipase alfa as an enzyme replacement therapy in children and adults with lysosomal acid lipase deficiency
41	Sharon Byers	MPS GAG modulate mesenchymal stem cell differentiation
42	Stephanie Cagle	Clinical outcomes and biochemical data following hematopoietic stem cell transplantation in a patient with Hunter syndrome
43	Marli Camelier	Galactocerebrosidase assay on dried-leukocytes impregnated in filter paper for the detection of Krabbe disease
44	Fabrice Camou	Gaucher disease: lived experience of patients in the context of a French national patient therapeutic education program
45	Jorge Cebolla	Experience with 7-ketocholesterol and CCL18/PARC as surrogated biomarkers in a series of Spanish Niemann-Pick disease type C patients
46	Jorge Cebolla	New haplotype of Fabry disease among patients screened for left ventricular hypertrophy of unknown cause
47	Agnes Chen	A study of intrathecal enzyme replacement for cognitive decline in mucopolysaccharidosis I
48	Yehudit Chen Zion	Quality of life and Gaucher disease

49	Wei-Chieh Cheng	Combinatorial approach toward rapid synthesis of iminosugar-based libraries: development of new β-glucocerebrosidase inhibitors and pharmacological chaperones for Gaucher disease
50	Eun-Young Choi	Choroid plexus-directed viral gene therapy for α-mannosidosis, a prototypical lysosomal disease
51	Jou-Ku Chung	Pharmacokinetics and biodistribution of idursulfase in non-human primates after intrathecal-lumbar administration
52	Heather Church	Prediction of CRIM status in Pompe disease using cultured chorionic villi.
53	Nicholas Clayton	Antisense oligonucleotide-mediated suppression of muscle glycogen synthase 1 synthesis as an approach for substrate reduction therapy of Pompe disease
54	Maureen Cleary	Early predictors of neurological involvement scoring system in mucopolysaccharidosis type 2: does it help?
55	Lisa Coles	Repeated-dose oral N-acetylcysteine: effect on brain and blood glutathione concentrations
56	Timothy Cox	ENCORE, a randomized, controlled, open-label non-inferiority study comparing eliglustat to imiglucerase in Gaucher disease type 1 patients stabilized on enzyme replacement therapy: 24-month results
57	Renata Cravo	Taliglucerase alfa: Rio de Janeiro experience at Hemorio
58	Marco Curiati	Enzyme replacement therapy for mucopolysaccharidosis type II: experience from a Brazilian reference center
59	Sheng Dai	Rapid kinetics of beta-cyclodextrin entering and exiting cells: Implication of its mechanism on reduction of cholesterol accumulation in Niemann-Pick disease type C cells
60	Carolina de Souza	Broad clinical and laboratory spectrum found in 9 Niemann-Pick disease type C Southern Brazilian patients
61	Carolina de Souza	Diagnosing communicating hydrocephalus in mucopolysaccharidoses: correlation between cerebrospinal fluid flow imaging and lumbar pressure studies
62	Patrick Deegan	Predictors of fracture in treated Gaucher patients
63	Russell DeKelver	ZFN-mediated genome editing of albumin “safe harbor” in vivo results in supraphysiological levels of human IDS, IDUA and GBA in mice
64	Kathleen Delaney	Methods for cognitive assessment of children and adults with lysosomal diseases
65	Robert Desnick	Fabry disease: the α-galactosidase A (GLA) c.427G>A (A143T) mutation, effect of the 5'-10C>T polymorphism
66	Colleen Doyen	Home infusion or infusion center decision criteria
67	Karen Dublan-García	Fabry disease in a male patient: de novo mutation, germline mosaicism or both?
68	Shaalee Dworski	Spatial distribution of brain ceramides in an acid ceramidase deficient murine model: subsequent histological manifestations and functional deficits
69	Jonathan Dyke	Comparison of cortical thinning in late infantile neuronal ceroid lipofuscinosis with a normative pediatric population using magnetic resonance imaging
70	Deborah Eastwood	The management of thoracolumbar deformity in the mucopolysaccharidoses
71	Johanneke Ebbink	Long−term neuropsychological follow−up in a patient with α−mannosidase
72	Julie Eisengart	Clinical outcomes of Hurler syndrome treated exclusively with enzyme replacement therapy from a young age
73	Sean Ekins	Phoenix Nest: starting a small company focused on Sanfilippo syndrome
74	Stuart Ellison	Pre-clinical workup of lentiviral mediated stem cell gene therapy for mucopolysaccharidosis type IIIA
76	Deborah Elstein	The need for disease-specific patient-reported outcome measures for lysosomal disorders
78	Jeffrey Esko	Intranasal enzyme replacement therapy in mice
79	Fatih Ezgü	Importance of family screening in Fabry disease: reaching the bottom of the iceberg

80	Andressa Federhen	MPS I and MPS II: minimal estimated incidence in Brazil and comparison to the rest of the world
81	Niamh Finnegan	Project to empower young type III Gaucher disease patients to shape the future
82	Qi Gan	Newborn screening for mucopolysaccharidoses: determination of sensitivity, specificity and cut-off score
83	José Garcia-Ortiz	Biochemical diagnosis of mucopolysaccharidoses in Mexico: preliminary results in a reference center
84	Scott Garman	Pharmacological chaperoning in Fabry and Schindler diseases
85	Michael Gelb	Newborn screening for lysosomal disease by tandem mass spectrometry: from development to worldwide implementation
86	Dominique Germain	A 10-year study documenting the long-term effectiveness of agalsidase-beta treatment in 52 adult patients with classic Fabry disease
87	Javier Gervas-Arruga	Complex intronic haplotype in Fabry disease
88	Arunabha Ghosh	Use of enzyme replacement therapy prior to haematopoietic stem cell transplantation for severe mucopolysaccharidosis I, a 10 year, 2-center retrospective review
89	Arunabha Ghosh	Haematopoietic stem cell transplantation in 3 patients with attenuated mucopolysaccharidosis type I with homozygous p.Leu490Pro mutation
90	Janine Gilkes	Evaluation of biodistribution and transduction profiles of novel AAV8 capsid mutated variants as a therapeutic candidate for the treatment of MPS IIIB
91	Roberto Giugliani	Oxysterol measurement in plasma: a potentially useful tool for the screening of Niemann-Pick disease type C
92	Ozlem Goker-Alpan	Effect of time of initiating enzyme replacement therapy on immune dysfunction in patients with Gaucher disease
93	Ashley Gonzalez	Modeling the association between Gaucher disease and Parkinson disease using in vivo mouse models
94	Russell Gotschall	Novel recombinant human acid α-glucosidase with optimal glycosylation is significantly better than standard of care enzyme replacement for glycogen clearance in skeletal muscles of GAA knock-out mice
95	Shane Grace	Age-dependent gene expression profile analysis in Morquio syndrome type A mouse cartilage tissue
96	Sue Graham	The Morquio A Registry Study (MARS): improving the understanding of Morquio syndrome type A
97	Perry Hackett	Non-viral gene therapy by liver-directed hydrodynamic delivery of Sleeping Beauty transposons to treat MPS in dogs
98	Bryan Hall	Microwave assisted CLARITY for whole organ surveillance
99	Takashi Hamazaki	Successful cord blood transplantation for Hurler syndrome patient with high-titer neutralizing antibody against α-l-iduronidase
100	Rick Hamler	Accurate quantitation of plasma globotriaosylsphingosine (lyso-Gb3) in normal individuals and Fabry patients by liquid chromatography-tandem mass spectrometry (LC-MS/MS)
101	Sang-Oh Han	Propranolol decreases the efficacy from enzyme replacement therapy in Pompe disease
102A	Paul Harmatz	Impact of elosulfase alfa on pain in patients with Morquio syndrome type A
102B	Katie Harvey	A comparison of plasma, leucocyte and dried blood spot α-galactosidase assays as first line diagnostic tests for Fabry disease
103	Katie Harvey	Prognostic utility of rapid leucocyte-based assay of α-glucosidase cross-reactive immunological material (CRIM) patterns in patients with Pompe disease
104	Alev Hasanoglu	The results of enzyme studies in the diagnosis of lysosomal diseases: 8 years experience of Gazi University, Ankara, Turkey
105	Christian Hendriksz	Antibody response to investigational intrathecal enzyme replacement therapy with idursulfase-IT in pediatric Hunter syndrome patients with cognitive impairment
106	Christian Hendriksz	The evaluation of psychological therapy interventions to improve emotional wellbeing in patients with lysosomal disorders.

107	Eric Herbig	Sleeping Beauty engineered human B lymphocytes express therapeutic levels of human iduronidase: a new approach for mucopolysaccharidosis type I
108	Stacy Hewson	Abdominal pain and mucosal hyperplasia of the gallbladder leading to a diagnosis of metachromatic leukodystrophy (MLD)
109	Wendy Heywood	Urine biomarker discovery using label free proteomics reveals novel markers for the monitoring of treatment for mucopolysaccharide disorders
111	Robert Hopkin	Risk factors for severe clinical events and the incidence of these events in male and female patients with Fabry disease treated with agalsidase beta
112	Mia Horowitz	Presence of mutant GBA allele leads to ER stress and development of Parkinson disease
113	Ting-Rong Hsu	Taiwanese patients with the Chinese IVS4+919G>A mutation who underwent endomyocardial biopsy: data from the Fabry Outcome Survey (FOS)
114	Derralynn Hughes	Fabry Disease: impact of ERT on renal function. Single-centre 5-year results
115	Derralynn Hughes	Long-term efficacy and safety of migalastat compared to enzyme replacement therapy in Fabry disease: phase 3 study results
116	Pilar Irún	Proteomic profile of osteoclasts in Gaucher disease patients according to the severity of bone manifestations
117	Margarita Ivanova	Impact of enzyme replacement therapy on lysosomal function in Gaucher disease
118	Matilda Jackson	α-L-iduronidase transduced mesenchymal stem cells improve the behavioural deficits in mucopolysaccharidosis type I mice
119	Roland Jaussaud	Patients' need to design a patient education program in Fabry disease (Filigrane)
120	Simon Jones	Effect of sebelipase alfa on survival and liver function in infants with rapidly progressive lysosomal acid lipase deficiency
121	Simon Jones	Enzyme replacement therapy (ERT) for mucopolysaccharidosis VII (MPS VII; Sly Syndrome) reduces lysosomal storage in a 36-week phase 1/2 clinical study
122	Amel Karaa	Expanding the clinical spectrum of the lysosomal disorders with whole exome sequencing
123	Nesrin Karabul	Ophthalmological manifestations in Fabry disease children in the Fabry Outcome Survey
124	Nesrin Karabul	Pedigree analysis: a call to action to raise awareness of Fabry disease and the importance of family history evaluation
125	Zoheb Kazi	Proteomics to identify signature proteins in patients likely to mount an immune response to enzyme replacement therapy in infantile Pompe disease
126	Maria Keever	A study to identify individuals at risk to be affected with late-onset Pompe disease with previous non-specific diagnoses
127	Jenny Kim	Gaucher disease and Parkinsonism: clinical course and prognosis
128	Kellie King	Average age at diagnosis for Sanfilippo syndrome: a case for newborn screening
129	Kelly King	Neurobehavioral outcomes in Sanfilippo syndrome type B compared to type A
130	Sandra Kingma	A study on the influence of glycosaminoglycan and growth factor interaction in mucopolysaccharidosis type I bone disease
131	Sandra Kingma	Adverse effects of genistein in mucopolysaccharidosis type I cell and mouse models
132	Masafumi Kinoshita	The characterization of mouse model of mucopolysaccaridosis type II
133	Priya Kishnani	Prophylactic immume modulation in infantile Pompe disease; collective experience treating CRIM-positive and negative patients in the naive setting
134	Priya Kishnani	The Pompe Registry: 10 years of data
135	Keisuke Kitakaze	Development of protease−resistant modified human β−hexosaminidase B and evaluation of intracerebroventricular replacement effects on GM2 gangliosidosis model mice
136	Nilima Kolli	Molecular basis of sialidosis and its treatment
137	Francyne Kubaski	Analysis of C6S/C4S ratio in Morquio syndrome type A patients

138	Francyne Kubaski	Chondroitin 6-sulfate as a novel biomarker for mucopolysaccharidosis IVA and VII
139	Francyne Kubaski	Di-sulfated keratan sulfate as a novel biomarker for mucopolysaccharidosis IVA
140	Francyne Kubaski	Noninvasive pulmonary function test on Morquio syndrome type A patients
141	Sandra Kyosen	Natural history of mucopolysaccharidosis in a referral center
142	Jean Lachowicz	Systemic administration of a brain-penetrant peptide-iduronidase conjugate results in brain IDUA activity in MPS I mice
143	Dawn Laney	Comparison of clinical practice guidelines and actual clinical practice in Fabry disease diagnosis
144	Eveline Langereis	Progression of hip dysplasia in MPS I patients (Hurler syndrome) after successful hematopoietic stem cell transplantation
145	Heather Lau	Multiple mechanisms of ophthalmologic involvement in attenuated Hunter syndrome: a case report
146	Heather Lau	Tremor and peripheral neuropathy are infrequent and non-serious events in Gaucher type 1 patients treated with eliglustat
147	Christine Lavery	Pedigree analysis in patients with Fabry disease: evaluating changes in referral and diagnosis over successive generations
148	Christine Lavery	Raising awareness of lysosomal diseases amongst British medical students
149	Alexandria Lee	Development of a less immunogenic protein for enzyme replacement therapy of Morquio syndrome type A disease
150	Malte Lenders	Thromboembolic events in Fabry disease and the impact of factor V Leiden
151	Paul Levy	Experiences from setting up a 5 year longitudinal, prospective, natural history study of patients with Sanfillipo syndrome types C or D (MPS IIIC or MPS IIID)
152	Lishu Li	Two masters of lysosomal and autophagosomal biogenesis, TFEB and TFE3, and their potential therapeutic value in Pompe disease
154	Renuka Limgala	Gastrointestinal manifestations of immune dysregulation and Gaucher disease: mesenteric lymphadenopathy and enteropathy with profound T cell defects
155	Renuka Limgala	Role of non-classical monocytes in Gaucher disease severity
156	Emily Lisi	Do the benefits outweigh the harms? Views of patients with later onset LSD on newborn screening
157	Valynne Long	Trabeculae bone structure analysis in individuals affected by type 1 Gaucher disease using micro magnetic resonance imaging
158	Yan Long	Cellular distribution and mechanism of delta-tocopherol on reduction of lysosomal cholesterol accumulation in cells with Niemann-Pick disease type C
159	Monica Lopez-Rodriguez	Alpha-mannosidosis and compassionate use of alpha-mannosidase (Lamazym™): two case reports
160	Charles Lourenco	“Night, night, sleep tight?”: sleep disorders in Fabry disease, recognizing an overlooked feature of a complex lysosomal disorder
161	Fernanda Ludwig	Updates in biochemical and molecular diagnosis of Brazilian patients with mucolipidosis II/III alpha/beta
162	Yi Lun	Histological examination of the effect of a highly phosphorylated proprietary recombinant human acid alpha-glucosidase on glycogen reduction in disease-relevant muscles of Pompe mice
163	Alexandra Malinowski	Evaluation of United States schools and colleges of pharmacy curriculum to assess education on lysosomal diseases
164	Thorsten Marquardt	Cystinosis: missing the diagnosis for more than 50 years
165	Thorsten Marquardt	Cystinosis treatment: kinetics of different cysteamine formulations and fluctuations of intracellular cystine levels
166	John Marshall	Evaluation of a novel substrate reduction therapy with CNS access in mouse models of neuronopathic Gaucher disease
167	Ana Martins	Outcome of pregnancy in Gaucher disease patients treated and not treated with imiglucerase
168	Paul McIntosh	Characterization of gait in late onset Pompe disease

169	Douglas McKechnie	Long term clinical outcomes in patients with Fabry disease receiving enzyme replacement therapy
170	Casey McKenna	Pregnancy in an adult with Maroteaux-Lamy syndrome: a case report
171	Blanca Medrano Engay	Gastrointestinal disturbances, lactose intolerance and Gaucher disease
172	Olga Meijer	N-acetyl-α-glucosaminidase activity in fibroblasts of patients with Sanflippo disease type B cultured at 30°C is associated with phenotypic severity
173	Matthew Metcalf	Characterization of a potential next generation enzyme replacement molecule for the treatment of Fabry disease
174	Langis Michaud	Vascular tortuosities of the upper eyelid: a new clinical finding in Fabry patient screening
175	Kevin Mills	The development of a multiplexed, rapid, mass spectrometry-based test for new and existing biomarkers to identify and monitor kidney disease in pediatric Fabry disease patients
176	Pramod Mistry	ENGAGE -- a phase 3, randomized, double-blind, placebo-controlled, multi-center study to investigate the efficacy and safety of eliglustat in adults with Gaucher disease type 1: results after 18 months
177	Ken Momosaki	Newborn screening of Pompe disease in Japan: 2 years experience
178	Isabelle Morin	Ear, nose and throat and hernia surgeries in children with Hunter syndrome: data from the Hunter Outcome Survey (HOS)
179	Joseph Muenzer	Long-term biomarker and cognitive follow up of children with Hunter syndrome receiving intrathecal enzyme replacement therapy
180	Behzad Najafian	Enzyme replacement therapy in Fabry disease reduces podocyte globotriaosylceramide (GL3) content within a year
181	Behzad Najafian	Mosaicism of podocyte involvement is related to podocyte injury in females with Fabry disease
182	Diana Najarian	An inter-laboratory comparison study of detection and characterization of anti-velaglucerase Alfa antibodies
183	John Naleway	Novel live cell screening platform for small molecules to enhance enzyme activity in Gaucher disease
184	John Naleway	Targeted chaperone therapy agents for Gaucher disease
185	Luba Nalysnyk	Imiglucerase treatment associated with reduction of bone claims in Gaucher patients: analysis of US claims data
186	Leyla Namazova-Baranova	Haemostatic system at diseases of the cardiovascular system in children
187	Juana Navarrete	Genotype-phenotype correlation in lysosomal diseases detected by lysosomal newborn screening in Mexico
188	Jazmin Navarro Munguia	Mucopolysaccharidosis I enzyme replacement treatment: experience of 3 cases in a 3rd level hospital, Hospital Infantil de Mexico Federico Gomez
189	Igor Nestrasil	Brain volumes and cognitive function in MPS IIIB (Sanfilippo Syndrome Type B): Cross-sectional study
190	Matthew Nguyen	Development of a novel neuronal cell model for investigating the link between glucocerebrosidase and Parkinson disease
191	Dau-Ming Niu	When is the best time to start enzyme replacement therapy in patients with cardiac-type Fabry disease? Experience from Taiwan, an area highly prevalent in this cardiac phenotype
192	Claire O'Leary	Development of an adeno-associated viral vector for mucopolysaccharidosis IIIC
193	Ilyas Okur	Identification of novel mutations and prevalence for Fabry disease (FD) via screening studies using dried blood samples (DBS) among hemodialysis patients in Turkey
195	Aida Oliván-Viguera	Characterization of monocyte / macrophage K_{Ca}3.1 channels in Gaucher disease
196	Alberto Ortiz	Occurrence of severe clinical events by time on agalsidase beta among patients with Fabry disease
197	Luying Pan	A comparison study of methods for detection and characterization of anti-idursulfase antibodies

198	Samantha Parker	AAVrh10-SGSH intracerebral gene therapy corrects the defect and improves the health status in mucopolysaccharidosis type IIIa
199	Marzia Pasquali	Urine karatan sulfate (uKS) in Morquio syndrome type A patients measured via LC-MS/MS method: improved KS detection as compared to dye-based methods and report of age-specific uKS reference ranges
200	Sun Peck	Failed vertebral bone formation in mucopolysaccharidosis VII dogs is associated with impaired chondrocyte hypertrophic differentiation
201	Maria Pedroso	High prevalence of liver diseases in patients with type I Gaucher disease in a specialized center: Is there an association with other genetically-determined liver disorders?
202	Jeff Peng	Improved respiratory function in a mouse model of Pompe disease treated with BMN 701
203	Jordi Pérez-López	Epidemiological assessment of Spanish patients with type 1 Gaucher disease using the therapeutic goals MAP Tool®
204	M. Judith Peterschmitt	Clinical response to eliglustat in treatment-naive patients with Gaucher disease type 1: post-hoc comparison to imiglucerase in a real-world setting
205	Rebecca Pleat	Early treatment with laronidase improves clinical outcomes in patients with attenuated MPS I: analysis of eight sibling pairs
206	Lynda Polgreen	Impact of laronidase on shoulder, elbow, and hip range of motion in children with Hurler syndrome after hematopoietic cell transplantation
207	Juan Politei	Fabry disease and ERT experience in 12 classic patients: different formulations, different outcome?
208	Juan Politei	Fabry disease: Late onset variant in proteinuria and dialysis screening: be prepared for more cases, and more questions
209	Laura Pollard	Clinical validity of beta-glucosidase and alpha-iduronidase enzyme analysis in dried blood spots
210	Katherine Ponder	Intrathecal injection of lentiviral vector results in high expression in the brain of mucopolysaccharidosis VII dogs but the pattern of expression is different than for AAV9 or AAV-rh10
211	Sean Prater	Consideration of increased dosing of alglucosidase alfa to achieve improved clinical outcomes in infantile Pompe disease
212	Helen Prunty	Glucose tetrasaccharide (Glc4) instability in urine, resolved by use of a special collection tube
213	Helen Prunty	Improved method for the analysis of urinary glucose tetrasaccharide (Glc4) by high pressure liquid chromatography (HPLC)
214	Alexey Pshezhetsky	Brain disease in mucopolysaccharidosis IIIC mouse: neuroinflammation, mitochondrial defects and neurodegeneration
215	Matthew Reed	Changes in peripheral blood osteoclast cultures in relation to therapeutic effects in Gaucher disease
216	Janine Reunert	Improved diagnostics of Niemann-Pick disease type C by the analysis of plasma oxysterols
217	Richard. Rogers	Screening an orthopedic population for mildly-affected individuals with Morquio syndrome type A and Maroteaux-Lamy syndrome
218	Sandra Rojas-Caro	Effect of sebelipase alfa after 2 years in adults with lysosomal acid lipase deficiency
219	Hanna Rosenbaum	The role of fibrosis in Gaucher disease
220	Vivian Rotman	Transitory elastography (TE) in patients with Gaucher disease
221	Paula Rozenfeld	Effect of glucocerebrosidase (GC) deficiency in osteoblasts on mineralization and osteoclastogenesis: implications for bone pathology in Gaucher disease
222	Adeel Safdar	Therapeutic potential of exosomes in Pompe disease: treatment of tomorrow, today for lysosomal diseases
223	Hitoshi Sakuraba	Determination of the structure of human α-L-iduronidase and structural basis of mucopolysaccharidosis type I
224	Saikat Santra	Desensitisation to galsulfase for the treatment of recurrent infusion association reactions in a child with MPS VI
225	Saikat Santra	Levomepromazine as a treatment for non-epileptic movement disorder in advanced neurodegenerative lysosomal disorders

226	Saikat Santra	Very early umbilical cord blood transplantation delays but does not prevent neurodegeneration in infantile Sandhoff disease
227	Imre Schene	Therapeutic options for patients with neuraminidase deficiency
229	Raphael Schiffmann	A prospective 10 year study of individualized, intensified enzyme replacement therapy in advanced Fabry disease
230	Raphael Schiffmann	Improvement in gastroinstestinal symptoms observed in the phase 3 FACETS (AT1001-011) study of migalastat in patients affected with Fabry disease
231	Erica Schindewolf	"Who is the deciding factor?" Analysis of parental perspectives regarding discontinuation of elaprase in children with MPS II
232	Joseph Schneider	Prevalence of hypothyroidism in adult-onset Pompe disease
233	Edward Schuchman	Novel use of the lysosomal enzyme acid ceramidase for the treatment of inflammatory lung diseases, including cystic fibrosis
234	Becky Schweighardt	Immunogenicity of elosulfase alfa, an enzyme replacement therapy in patients with Morquio syndrome type A: results from MOR-004, a phase 3 trial
235	C. Ronald Scott	Identification of newborn infants at risk for a lysosomal disease by tandem mass spectrometry
236	Rosângela Silva	Exercise in lysosomal diseases
237	Angela Simcox	Treatment decision making for parents of children with lysosomal disorders in the era of rapidly advancing medical options
238	Calogera Simonaro	Pentosan polysulfate: new mechanistic insights and treatment of the mucopolysaccharidoses
239	Ernestas Sirka	The development of a rapid, multiplexed UPLC-MS/MS assay for quantitation of lyso-Gb3 and Gb3 in dried blood spots
240	Siyamini Sivananthan	Disease course after cessation of enzyme replacement therapy (ERT) in 5 patients with type II mucopolysaccharidosis (MPS II)
241	Siyamini Sivananthanan	Hematopoietic cell transplant in mucopolysaccharidosis type I: single centre review of age of diagnosis and time to transplant
242	Melanie Sivley	Conjunctival lymphangiectasias and cysts persist despite long-term enzyme replacement therapy in males with Fabry disease
243	Hatice Serap Sivri	Two adult siblings with progressive walking difficulty and visual disturbances
244	Elizabeth Smith	A patient with mucopolysaccharidosis type I diagnosed at 38 of age with only one identifiable mutation: a case report from the newborn screening perspective
245	Alexander Solyom	Acid ceramidase deficiency: clinical implications of an emerging phenotypic spectrum and potential therapies
247	Eser Sozmen	Lekocyte cell surface antigens in Gaucher disease: new implications for B-cell proliferation and pathogenesis of myelomatosis
248	Richard Steet	Small molecule modulation of CI-MPR-dependent uptake of therapeutic enzymes in patient fibroblasts
249	Karolina Stepien	A 4-year follow up study of 24 patients with late onset Pompe disease treated with alglucosidase alfa enzyme replacement therapy at a single centre
250	Fiona Stewart	Study of an extended 4-generation family with A143T Fabry mutation: presentation of variable phenotypes including very mildly affected individuals
251	Akemi Tanaka	Genotype of mucopolysaccharidosis type II severe form and the efficacy of enzyme replacement therapy or hematopoietic stem cell transplantation on cognitive function
252	Noboru Tanaka	Novel quantification methods for globotriaosylceramide and globotriaosylsphingosine as biomarkers of Fabry disease
253	Brittany Taylor	Dietary intake of individuals with late onset Pompe disease: a review and comparison to current diet recommendations
254	Alison Thomas	Gaucher disease results in an acquired mucocutaneous bleeding disorder treatable with enzyme replacement therapy
255	Alison Thomas	The N215S mutation results in a distinct subtype of Fabry disease
256	Beth Thurberg	A phase 4 prospective study in patients with adult Pompe disease treated with alglucosidase alfa

257	Beth Thurberg	Hepatic pathology of acid sphingomyelinase deficiency: Clearance of sphingomyelin with recombinant human acid sphingomyelinase adminstration is associated with improvement in pro-atherogenic lipid profiles
258	Adviye Tolun	Diagnostic value of a microfluidic based fluorometric enzyme assay platform using dried blood spots for a lysosomal disorder, Fabry disease
259	Shunji Tomatsu	Activity of daily life in patients with Hunter syndrome: impact of enzyme replacement therapy and hematopoietic stem cell transplantation
260	Shunji Tomatsu	Establishment of glycosaminoglycan assays for mucopolysaccharidoses
261	Shunji Tomatsu	Impact of enzyme replacement therapy and hematopoietic stem cell therapy on growth in patients with Hunter syndrome
262	Shunji Tomatsu	Long term follow up of post-hematopoietic stem cell transplantation for Hurler syndrome: clinical biochemical and pathological improvements
263	Shunji Tomatsu	Novel heparan sulfate assay by using automated high throughput mass spectrometry: application to monitoring and screening for mucopolysaccharidoses
264	Shunji Tomatsu	Therapies for the bone in mucopolysaccharidoses
265	Camilla Tøndel	Foot process effacement is an early marker of nephropathy in young classic Fabry patients without albuminuria
266	Takahiro Tsukimura	Comprehensive study of Fabry disease: gene mutation, GLA activity, GLA protein and globotriaosylsphingosine
267	Leyla Tumer	Isovaleric acidemia and Niemann-Pick disease type C coexistence and new mutation for Niemann-Pick disease type C
268	Kelly Turner	Sanfilippo syndrome type B: reprogramming cultured skin fibroblasts to induced pluripotent stem cells using non-integrating sendai virus vector
269	Sandrine Turpault	CYP2D6 phenotype-based dosing of eliglustat
270	Jeanine Utz	Biomarkers of central nervous system inflammation in infantile and juvenile gangliosidoses
271	Filippo Vairo	Gastrointestinal disorders and miglustat therapy: a case report
272	Filippo Vairo	Inflammasome during pregnancy in a Gaucher disease patient
273	Filippo Vairo	Osteopontin: a potential biomarker of Gaucher disease
274	Filippo Vairo	Taliglucerase alfa to type I Gaucher disease: a south Brazilian experience
275	Ans van der Ploeg	Anti-alglucosidase alfa antibodies and infusion-associated reactions in 73 treated adult Pompe disease patients
276	Suresh Vijay	Evaluation of blood-brain barrier integrity and structural abnormalities in MPS IIIb patients using cerebrospinal fluid/serum albumin index (CSF-AI) and multimodal MRI
277	Charles Vite	Intracisternal cyclodextrin ameliorates neurological dysfunction, increases survival time, and stops Purkinje cell death in feline Niemann-Pick type C1 disease
278	Amy Wakumoto	Amygdalar volumes and acquired autistic symptoms in MPS IIIA
279	Susanne Walls	A new approach to follow-up of Finnish Fabry patients patient-based care
280	Jen-Hon Wang	Rapid synthesis of pyrrolidine-based iminosugars to develop pharmacological chaperones for disease
281	Raymond Wang	Carotid intima-media thickness and arterial stiffness of pediatric mucopolysaccharidosis patients are increased compared to both pediatric and adult populations
282	Raymond Wang	Human mucopolysaccharidosis IIIa patients do not demonstrate postprandial hypertriglyceridemia, but have increased carotid intima-media thickness
283	Raymond Wang	The mucopolysaccharidosis type IIIA murine model demonstrates increased brown adipose activity and energy demand, resulting in postprandial hypertriglyceridemia
284	Katie Warner	The role of the psychologist in the metabolic team: a patient-lead approach

285	Melissa Wasserstein	An open-label, multicenter, ascending-repeat-dose study of the tolerability and safety of recombinant human acid sphingomyelinase (rhASM) in patients with ASM deficiency (ASMD)
286	Richard Welford	Plasma lysosphingomyelin demonstrates great potential as a diagnostic biomarker for Niemann-Pick disease type C in a retrospective study
287	James Wilson	Adeno-associated virus vector-mediated gene therapy can effectively treat CNS and cardiac lesions and induce immune tolerance to the therapeutic enzyme in large animal models of mucopolysaccharidosis type I
288	Robert Wood	Effect of enzyme replacement therapy on airway abnormalities in patients with Hunter syndrome
289	Chia Feng Yang	Very early enzyme replacement therapy is “The earlier; the better” for infantile Pompe disease: experience of nationwide newborn screening program in Taiwan
290	Fabian Yu	Impaired lung function in the acid ceramidase deficient mouse
291	Aysel Yuce	Skeletal manifestations of children with Gaucher disease type I and type III
292	Ari Zimran	Comparison of taliglucerase alfa 30 u/kg and 60 u/kg in treatment-naïve pediatric patients with Gaucher disease
293	Ari Zimran	Long-term safety and efficacy of taliglucerase alfa in pediatric patients with Gaucher disease who were treatment-naïve previously treated with immiglucerase
294	Ari Zimran	Markers of bone turnover in patients with type 1 Gaucher disease receiving long-term velaglucerase alfa enzyme replacement therapy
LB-1	Marcio Andrade-Campos	Prospective study of plasma biomarkers associated with the inflammatory response in type 1 Gaucher disease patients treated during one year with velaglucerase alpha.
LB-2	Daniel Bichet	Migalastat reduces left ventricular mass index in Fabry patients naïve to ERT and previously treated with ERT
LB-3	Barbara Burton	Impact of elosulfase alfa on exercise capacity and muscle strength and safety in patients with Morquio syndrome type A
LB-4	Yin-Hsiu Chien	A high incidence of Fabry disease variants argues against newborn screening: outcomes of the Taiwan pilot
LB-5	Jason Cournoyer	An FIA MS/MS method to simultaneously measure ABG, ASM, GAA, GALC, GLA and IDUA activity in dried blood spots
LB-6	Cristin Davidson	Combinatorial therapy for Niemann-Pick disease type C: treatment of an NPCI murine model with 2-hydroxypropyl-beta-cyclodextrin and miglustat
LB-7	James DiPerna	A new MS/MS method to measure MPS IVA and MPS VI enzyme activities in dried blood spots
LB-8	Sergio Figueroa-Sauceda	Charcot arthropathy in a patient with Fabry disease
LB-9	Haiyan Fu	Restoration of NAGLU activity by a single systemic rAAV9 vector delivery led to the correction of widespread metabolomic dysfunction in MPS IIIB mice
LB-10	Rabia Gowa	Fabry disease and pain control
LB-11	Sue Graham	Impact of mucopolysaccharidosis on daily living, employment, general health and parenthood of adult patients
LB-12	Christian Hendriksz	Risks of long-term port use in the mucopolysaccharidosis patient population
LB-13	Patricia Kane	Optimizing neurometabolic function in lipid storage disorders by addressing epigenetics with phospholipid therapy
LB-14	Yan Long	Niemann-Pick disease type A: induced pluripotent stem cells for disease modeling and compound screening
LB-15	Juan Montes-Ramírez	Fabry disease in a Mexican family: a novel GLA gene mutation and the relevance of extended family tree investigation
LB-16	David Palmer	Effective gene therapy in ovine CLN5 Batten disease
LB-17	Ester Pereira	Generation of Fabry disease kidney cell lines using genome editing by CRISPR/Cas9
LB-18	Rebecca Pleat	Concomitant medication use and comorbidities in adult patients with Gaucher disease type 1: results from the MarketScan™ database

LB-19	Adrian Quartel	Pulmonary function predictors (VC, FVC, MIP, MEP) of respiratory insufficiency in late-onset Pompe disease
LB-20	Enzo Ranieri	The feasibility of using an MS/MS based method with Perkin Elmer lysosomal disease reagents to implement a newborn screening test for six lysosomal disorders
LB-21	Jane Roberts	Unexpected issues around the temporomandibular joint in patients with MPS
LB-22	Juan Romero-Trejo	Ventricular tachyarrhythmias in Fabry disease: relevance of enzyme replacement therapy dose apropos of a case
LB-23	Stephan Rust	A variant in NPC type 2 disease, that is more frequent than NPC1
LB-24	Annalisa Sechi	Acute effect of enzyme replacement therapy on exercise tolerance in late-onset Pompe patients
LB-25	Jinsong Shen	Establishment of immortalized endothelial cell lines from Fabry mouse aorta
LB-26	Zdenek Spacil	Newborn screening for metachromatic leukodystrophy in dried blood and urine spots
LB-27	Zdenek Spacil	Pilot studies of tandem mass spectrometry newborn screening for mucopolysacharidoses type II, IIIA, IIIB, and VI
LB-28	Adrian Todd	Improving neuromuscular junction pathology using AAV9 gene therapy in Pompe disease
LB-29	Sara Turner	Respiratory-related motoneurons are the first to show histopathology in Pompe mice
LB-30	Valerie Vernot	Volume compensation to inspiratory loads improves after gene therapy for Pompe disease
LB-31	Jesus Villarrubia	Correlation between the genotype and the phenotype in type in Gaucher disease in Spanish patients
LB-32	Robert Spiegel	Rationale for ataluren as a potential new treatment in patients with nonsense mutation mucopolysaccharidosis type I (nmMPS I)
LB-33	Walter Acosta	Lectin-mediated ERT delivery: correcting lysosomal disease pathologies using novel cell uptake mechanisms