# Poster Session Abstracts

Poster sessions will be in the Harbor Ballroom on:

Tuesday, February 14 from 4:30-6:30pm
Wednesday, February 15 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 14, 2017 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 sessions 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 15, 2017 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 sessions ends.

**All late-breaking abstracts** will be assigned to the poster session on Wednesday, February 15, 2017 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 sessions ends.

***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.

## Tuesday, February 14 – Poster Presentations

| Poster # | First Author | Abstract Title |
|---|---|---|
| **1** | Magy Abdelwahab | Cardiac assessment in Egyptian type 3 Gaucher disease and report of calcifications in homozygous L444P mutation |
| **2** | Patricio Aguiar | MicroRNAs in Fabry disease: role as biomarkers and a possible pathophysiological pathway |
| **3** | Patrício Aguiar | Increase synthesis of collagen type I: an early event in Fabry cardiomyopathy. New biomarkers? |
| **4** | Amelia Ahern-Rindell | Characterization of a secondary deficiency of neuraminidase activity in a unique ovine model of GM1 gangliosidosis |
| **5** | Alia Ahmed | Neurocognitive and behavioral longitudinal trajectory of Hurler-Scheie patients with L238Q mutation |
| **6** | Rebecca Ahrens-Nicklas | Small molecule therapies for juvenile neuronal ceroid lipofuscinosis |
| **8** | Kyrieckos Aleck | Urinary glycosaminoglycan levels in a mucopolysaccharidosis type II pediatric population receiving idursulfase therapy: data from the Hunter Outcome Survey (HOS) for patients aged <18 months |
| **9** | Nadia Ali | Preliminary validation of telecounseling for depression in patients with Fabry disease |
| **10** | Carlos Almeciga-DIaz | Human recombinant N-acetylgalactosamine-6-sulfate sulfatase (GALNS) produced in a glycoengineered Escherichia coli strain |
| **11** | Carlos Almeciga-Diaz | Understanding the metabolic consequences of human arylsulfatase A deficiency through a computational systems biology study |
| **12** | Carlos Almeciga-Diaz | Interaction between the human GALNS crystal structure and different ligands using computational molecular docking |
| **13** | Moeenaldeen AlSayed | High dose alglucosidase alfa to reverse ventilator dependence during clinical deterioration in a patient with Pompe disease |
| **14** | Moeenaldeen AlSayed | Response to high dose escalation therapy with sebelipase alfa in a patient with severe infantile lysosomal acid lipase deficiency (ILALD) and further insight into this disease |
| **15** | Nicole Anderson | Short-term longitudinal cognitive and neuroimaging studies of untreated fucosidosis |

| | | |
|---|---|---|
| **16** | Marcio Andrade-Campos | Assessing underlying chronic inflammation status in Gaucher disease type 1 patients (GD1): involvementof lipocaline (lcn2) and other monocyte cytokines |
| **17** | Kara Anstett | Incidental diagnosis of a 35 year old pregnant woman with presymptomatic Pompe disease by expanded carrier screening |
| **18** | Carolina Aranda | Hypersensitivity reactions to enzyme replacement therapy for lysosomal diseases in Brazil: outcomes and safety of 1140 rapid desensitizations |
| **19** | Christiane Auray-Blais | Diurnal variation of Fabry biomarkers during enzyme replacement therapy |
| **22** | Lauren Bailey | A newborn screening dilemma: when to treat Pompe disease with c.-32-13T>G IVS splice site mutation |
| **23** | Laurie Bailey | Could it be something else? Patients with Gaucher disease, chronic pain, and fatigue found to also meet diagnostic criteria for another inherited condition, Ehlers-Danlos syndrome, hypermobility type |
| **24** | Suelen Basgalupp | Relation between homocysteine and vitamin B12 levels in Brazilian patients with Gaucher disease |
| **25** | Brendan Beaton | The role of mesenchymal stem cells in the development of bone pathology and myeloma in Gaucher disease |
| **26** | David Bedwell | Identification of drugs to treat MPS I caused by nonsense mutations |
| **27** | Soraya Bekkali | Design and rational of the natural history study in preparation for the pivotal clinical trial for the treatment of Sanfilippo syndrome type A |
| **28** | Soumeya Bekri | Development, analytical validation and implementation of a next generation sequencing panel to assess lysosomal diseases |
| **30** | Bruno Bembi | Investigation on acute effects of ERT and influence of clinical severity on physiological variables related to exercise tolerance in late onset Pompe disease |
| **32** | Donna Bernstein | Rapidly progressive hepatosteatosis and liver failure five years after liver transplantation in a patient with lysosomal acid lipase deficiency |
| **34** | Brian Bigger | Neurological correction of mucopolysaccharidosis IIIB mice by haematopoietic stem cell gene therapy |
| **35** | Tobias Boettcher | Biochemical and genetic data in a large, worldwide Fabry cohort |
| **36** | Constanza Bondar | Osteocytes contribute to bone pathology in Gaucher disease |
| **37** | Line Borgwardt | Improvement in pulmonary function and serum immunoglobulin G in long-term enzyme replacement therapy with velmanase alfa (human recombinant alpha-mannosidase) in alpha-mannosidosis patients |
| **38** | Line Borgwardt | Improvement in fine and gross motor proficiency after long-term enzyme replacement therapy with velmanase alfa (human recombinant alpha mannosidase) in alpha-mannosidosis patients |
| **40** | Araceli Borja-Borja | Ophthalmologic findings in Fabry disease patients: relation to disease severity and to enzyme replacement therapy in a Mexican family |
| **41** | Gordon Brandt | Messenger RNA (mRNA) delivery to the liver corrects ornithine transcarbamylase deficiency in a mouse disease model |
| **42** | Heydy Bravo-Villalta | Investigation of newborns screened in a pilot program for four lysosomal diseases in Brazil |
| **43** | Joanna Brokowska | Sulfpraphane induces autophagy and reduces the level of mutated huntingtin in human fibroblasts |
| **44** | Alexander Broomfield | Rapid progressive leukodystrophy in early childhood: a new phenotype for treated infantile onset Pompe patients? |
| **45** | Alexander Broomfield | Combined therapeutic approach to protein losing enteropathy complicating type 3 Gaucher disease using eliglustat |
| **46** | Alexander Broomfield | A single center's 10-year experience of idursulfase in MPS II |
| **47** | Barbara Burton | Long-term benefit of sebelipase alfa over 76 weeks in children and adults with lysosomal acid lipase deficiency (LAL-D) (ARISE) |

| | | |
|---|---|---|
| **49** | Gustavo Cabrera | Long term enzyme replacement therapy for Fabry disease: effectiveness on heart, kidney and brain |
| **50** | Stephanie Cagle | A case report of a Hispanic male with mucolipidosis III gamma with mild disease in the presence of a homozygous nonsense mutation |
| **51** | Carolina Cardona | Iduronate-2-sulfatase proteome isolation from mouse brain and identification of changes in the proteomic profiling in murine model for Hunter syndrome |
| **52** | Fu-Pang Chang | Accumulation of globotriaosylceramide (GL3) in cardiomyocytes (CM) is progressive with age and inversely correlates with baseline alpha galactosidase A (AGALA) activity in enzyme replacement therapy (ERT)-naïve Fabry patients with IVS4 + 919G>A mutation |
| **53** | Parapoj Changsila | Mechanism and efficiency of delivery of therapeutic enzymes in different cell models of Fabry disease |
| **55** | Anu Cherukuri | Immunogenicity to cerliponase alfa, an enzyme replacement therapy for patients with CLN2 disease: results from a phase 1 /2 study |
| **56** | Yin-Hsiu Chien | Risk assessments in infants suspect having later-onset Pompe disease identified through newborn screening |
| **59** | Don Clarke | Development of a stem cell gene therapy for Sanfilippo syndrome type B |
| **60** | Jonathan Cooper | The glial contribution to pathogenesis differs between forms of Batten disease: implications for therapy |
| **61** | Jason Cournoyer | Six-plex MS/MS method to measure I2S, NAGLU, GALNS, ARSB, GUSB and TPP1 enzyme activities in dried blood spots |
| **62** | Timothy Cox | Maintenance of quality of life in adults with Gaucher disease type 1 previously stabilized on enzyme therapy who were switched to oral eliglustat: 4 year results of the ENCORE trial |
| **63** | Claudia Cozma | C26-ceramide is a new and sensitive biomarker for Farber disease |
| **64** | Claudia Cozma | Lyso-SM-509 is an easy-measurable and sensitive biomarker for Niemann-Pick disease: a two year study |
| **65** | Marco Curiati | The impact of the disease burden on the quality of life of patients with lysosomal diseases: preliminary data of adult patients under enzyme replacement therapy |
| **66** | Amauri Dalla-Corte | An algorithm to assess the need for CSF shunting in mucopolysaccharidosis patients |
| **69** | Talita de Carvalho | CRISPR/Cas9-directed genome editing of human MPS I fibroblasts |
| **70** | Patrick Deegan | Treatment patterns from 647 patients with Gaucher disease: an analysis from the Gaucher Outcome Survey |
| **71** | Russell DeKelver | ZFN-mediated in vivo genome editing results in phenotypic correction in murine MPS I and MPS II models |
| **72** | Dolores del Pino | PReFiNe project: strategic plan to improve knowledge & recognition of Fabry disease among Spanish nephrologists |
| **73** | Robert Desnick | The Fabry Disease genotype-phenotype database (dbFGP): an international expert consortium |
| **74** | Hung Do | Stabilized next-generation recombinant human acid alpha-glucosidase ATB200 clears accumulated glycogen and reverses cellular dysfunction to increase functional muscle strength in a mouse model of Pompe disease |
| **75** | Dana Doheny | Fabry disease: prevalence of affected males and heterozygotes with pathogenic GLA mutations identified by screening renal, cardiac, and stroke clinics |
| **76** | Aimee Donald | A retrospective review of the neurocognitive profile of patients with neuronopathic Gaucher disease in the UK: data from the GAUCHERITE study |
| **77** | Danielle Dong | Evolving observational registries: an evidence based approach to understanding data availability in the "Sanofi Genzyme Rare Disease Registries" |

| | | |
|---|---|---|
| **78** | Halil Dündar | In vitro translational readthrough by gentamicin and geneticin improves GLA activity in Fabry disease |
| **79** | Berendine Ebbink | Neuropsychological profile of long-term treated patients with classic infantile Pompe disease |
| **80** | N. Matthew Ellinwood | Preliminary findings of a twenty-six week or longer intracerebroventricular infusion study of BMN 250 administered once every 2 weeks in a canine model of mucopolysaccharidosis type IIIB |
| **82** | Deborah Elstein | Medical histories in Gaucher disease: a descriptive analysis from 852 patients in the Gaucher Outcome Survey (GOS) |
| **83** | Andressa Federhen | Sanfilippo disease type B: a review of patients diagnosed by the MPS Brazil Network |
| **84** | Ulla Feldt-Rasmussen | Efficacy and safety of migalastat, an oral pharmacologic chaperone for Fabry disease: results from two randomized phase 3 studies, FACETS and ATTRACT |
| **85** | Can Ficicioglu | Intrafamilial variability in the clinical presentation of mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS) |
| **86** | Veronica Finn | Cardiovascular mortality and potential predictors in patients with Fabry disease treated with agalsidase alfa |
| **87** | Michael Flanagan | Echocardiographic assessment of Morquio syndrome type A mice |
| **88** | Michael Flanagan | Impact of miglustat therapy on the hypophosphatasia mouse model |
| **90** | Joaquin Frabasil | Improving the detection of heterozygous females for Fabry disease |
| **91** | Mark Friedman | Effect of sebelipase alfa on survival to 3 years of age and liver function in infants with rapidly progressive lysosomal acid lipase deficiency |
| **92** | Andrea Frustaci | Immune mediated myocarditis in Fabry disease cardiomyopathy |
| **93** | Haiyan Fu | Global metabolomics profiling reveals profound metabolic impairments in patients with mucopolysaccharidosis III |
| **95** | Katie Gallagher | Fabry disease symptoms confined to the kidney in a large family with the M296V genotype |
| **96** | Jaya Ganesh | Late-onset Pompe disease with atypical presentation - what else is going on? |
| **97** | Jose Garcia Fernandez | Fluorinated chaperone-β-cyclodextrin formulations for neuronopathic Gaucher disease |
| **98** | Roselyne Garnotel | In vivo Raman micro-spectroscopy: a new way to screen Fabry disease |
| **99** | Michael Gelb | Newborn screening and post-screening diagnosis of lysosomal diseases |
| **100** | Dominique Germain | A Fabry genotype-phenotype working group initiative: classifying GLA mutations for male patients in the Fabry Registry |
| **101** | Dominique Germain | The phenotypic characteristics of the p.N215S Fabry disease genotype in male and female patients: a multi-center Fabry Registry study |
| **102** | Dominique Germain | Efficacy of migalastat in a cohort of male patients with the classic Fabry phenotype in the FACETS phase 3 study |
| **103** | Dominique Germain | Effects of treatment with migalastat on the combined endpoint of kidney globotriaosylcermide accumulation and diarrhea in patients with Fabry disease: results from the phase 3 FACETS study |
| **104** | Arunabha Ghosh | Plasma oxysterols as a putative biomarker for infantile onset lysosomal acid lipase deficiency (Wolman disease) |
| **105** | Arunabha Ghosh | Haematopoietic stem cell transplantation alongside enzyme replacement therapy in infantile onset lysosomal acid lipase deficiency (LAL-D, Wolman disease) |
| **106** | Vicente Giner-Galvañ | Home enzymatic therapy administration for Gaucher disease in Spain: first national experience |
| **107** | Vicente Giner-Galvañ | Proposal of a tool for the asessement of quality of Gaucher disease clinical management |
| **108** | Vicente Giner-Galvañ | Home enzymatic therapy administration program for lysosomal diseases in Spain: first national experience |

| | | |
|---|---|---|
| **109** | Vicente Giner-Galvañ | Qualitative evaluation of home therapy administration of enzymatic treatment for patients with Gaucher and Fabry disease in the Comunitat Valenciana in Spain: point of view of patients and health care professionals |
| **110** | Vicente Giner-Galvañ | Home enzymatic infusion program for Gaucher and Fabry diseases: economic impact evaluation of the experience in the Comunitat Valenciana in Spain |
| **111** | Pilar Giraldo | Is there an increased incidence of neoplasia in Spanish Gaucher disease patients? |
| **113** | Roberto Giugliani | Long-term galsulfase treatment associated with improved survival of patients with mucopolysaccharidosis VI (Maroteaux-Lamy syndrome): 15 year follow-up from the survey study |
| **114** | Roberto Giugliani | Clinical outcomes after 3 years of idursulfase treatment in patients with MPS II: data from the Hunter Outcome Survey (HOS) |
| **115** | Hélène Gleitz | Whole body correction of severe mucopolysaccharidosis type II by lentiviral-mediated stem cell gene therapy with blood-brain barrier-crossing peptides |
| **116** | Esteban Gonzalez | Losartan improves cardiovascular disease in mucopolysaccharidosis type I |
| **118** | Alexandra Gossler | Natural history of siblings with mucopolysaccharidosis type IIIA - Sanfilippo syndrome type A |
| **120** | Richard Grey | Looking for diamonds in the rough: identifying differentially expressed modifier genes in mouse models for type 2 Gaucher disease |
| **121** | Karen Grinzaid | Impact of education and the facilitation of carrier screening in a population at increased risk for lysosomal diseases |
| **122** | Nicolas Guérard | Lucerastat, an iminosugar for substrate reduction therapy: pharmacokinetics, tolerability, and safety in subjects with mild, moderate, and severe renal function impairment |
| **123** | Nicolas Guérard | Lucerastat, an iminosugar for substrate reduction therapy: safety, tolerability, pharmacodynamics, and pharmacokinetics in adult subjects with Fabry disease |
| **124** | Nathalie Guffon | MPS I and carpal tunnel syndrome: analysis using the MPS I Registry |
| **125** | Punita Gupta | Early initiation of prophylactic immune tolerance induction and enzyme replacement therapy in prenatally diagnosed infantile onset Pompe disease with a CRIM-negative mutation |
| **126** | Punita Gupta | Infantile Tay-Sachs disease: a case report |
| **127** | Christine Ha | Outside the fiber: interstitial pathology of skeletal muscle in infantile Pompe disease |
| **128** | Aleksandra Hac | Functions of lysosomes are impaired during prolonged stress conditions in cells devoid of S6K1/2 |
| **130** | Alaa Hamed | A conceptual framework of patient-reported outcomes for Gaucher disease type 3 |
| **131** | Alaa Hamed | PRO instrument development for late-onset Pompe disease |
| **132** | Alaa Hamed | Modeling of changes in forced vital capacity and late-onset Pompe disease related outcomes |
| **133** | Sang-oh Han | Beneficial effects of carvedilol with enzyme replacement therapy in Pompe disease |
| **134** | Paul Harmatz | Characteristics of patients with mucopolysaccharidosis type II (MPS II) diagnosed aged <5 years: data from the Hunter Outcome Survey (HOS) |
| **135** | Paul Harmatz | A novel, randomized, placebo-controlled, blind-start, single-crossover phase 3 study to assess the efficacy and safety of UX003 (rhGUS) enzyme replacement therapy in patients with MPS VII |
| **137** | Coy Heldermon | Dietary effects on glycosaminoglycan storage in MPS IIIB |
| **138** | Nadene Henderson | The Pennsylvania newborn screening experience for Pompe disease |
| **139** | Chris Hendriksz | Identification of relevant clinical niches in rare inherited metabolic diseases: Niemann-Pick disease type C as a model |

| | | |
|---|---|---|
| **140** | Christian Hendriksz | Elosulfase alfa treatment and changes in physical functioning and disability in Morquio syndrome type A |
| **141** | Julia Hennermann | Arterial tortuosity is a new clinical feature in patients with mucopolysaccharidosis type IVA |
| **142** | Robert Hopkin | Burden of Fabry disease in young patients (≤30 years of age) who were initiated on enzyme replacement therapy with agalsidase beta: a Fabry Registry analysis |
| **145** | Mohammad Hossain | The severity of phenotype for a heterozygous Fabry female patient correlates to the methylation of non-mutated allele |
| **146** | Audrey Hou | Two-year postmarketing safety experience with oral eliglustat in adults with Gaucher disease type 1 |
| **147** | Jeffrey Huang | Generation of infantile- and late-onset Pompe disease models using CRISPR-Cas9 genome editing |
| **148** | Derralynn Hughes | Sub-analysis of long-term elosulfase alfa treatment outcomes in adults with Morquio syndrome type A |
| **149** | Derralynn Hughes | One-year follow up of Fabry disease patients treated by IV administration of a plant derived alpha-Gal-A enzyme: safety and efficacy |
| **150** | Derralynn Hughes | Response of patients with Fabry disease with the amenable GLA mutation p.N215S to treatment with migalastat |
| **151** | Marshall Huston | Liver-based expression of the human alpha-galactosidase A gene (GLA) in a murine Fabry model results in continuous supra-physiological enzyme activity and effective substrate reduction |
| **152** | Kohji Itoh | A transgenic silkworm overexpressing human lysosomal enzyme as a novel resource for producing recombinant glycobiologics and its application to development of enzyme replacement therapy for lysosomal diseases |
| **153** | Margarita Ivanova | Personalized medicine strategies in lysosomal diseases: cell models for in vitro screening in Gaucher disease |
| **154** | Marlene Jacobson | Patient derived phenotypic high throughput assay to discover treatments for Tay-Sachs disease |
| **155** | Era Jain | Injectable microgels development for sustained GALNS enzyme replacement therapy for Morquio syndrome type A |
| **156** | James Jarrett | The patient experience of elosulfase alfa treatment for MPS IVA (Morquio syndrome type A): beyond traditional measures |
| **157** | Prakrit Jena | Optical non-invasive detection of Niemann-Pick disease in vitro and in vivo |
| **158** | Rui-Ru Ji | Novel NAGLU variants associated with MPS IIIB and in vitro analysis of residual enzymatic activities |
| **159** | Franklin Johnson | Migalastat exposures in Japanese healthy volunteers and non-Japanese subjects provide evidence that they are similar to Japanese patients with Fabry disease |
| **161** | Olive Jung | Novel molecular tools to advance the evaluation of Gaucher disease therapeutics in live cells |
| **162** | Chanchala Kaddi | Quantitative systems pharmacology model of acid sphingomyelinase deficiency and the enzyme replacement therapy olipudase alfa is an innovative tool for linking pathophysiology and pharmacology |
| **163** | Ayfer Kahraman | The effects of sub-maximal aerobic exercise in adults with late-onset Pompe disease (LOPD) |
| **164** | Shih-hsin Kan | AAAV5-mediated gene therapy with choroid plexus-directed α-n-acetyl-glucosaminidase expression in Sanfilippo syndrome type B mice |
| **165** | Ilkka Kantola | PQ-interval and QRS duration increased in Fabry patients treated by enzyme replacement therapy for 12 years |
| **166** | Reena Kartha | Role of oxidative stress and inflammation in type 1 Gaucher disease (GD1): Potential use of antioxidant/anti-inflammatory medications |
| **167** | Yoo-Mi Kim | Successful treatment with eliglustat in unexpected gastrointestinal involvement in a patient with type 1 Gaucher disease |

| | | |
|---|---|---|
| **168** | Kelly King | Age and social functioning in individuals with MPS I, II, and VI |
| **170** | Priya Kishnani | Sequence variants and genotypes among 898 patients with Pompe disease: data from the Pompe Registry |
| **171** | Anja Koehn | Clinical course in a MPS IIIA patient following hematopoietic stem cell transplantation: an eight-year follow up and comparison with the natural history in patients with identical mutations in the SGSH-gene |
| **172** | Tiago Koppe | Gaucher disease patients show increased levels of hepcidin - evidence for iron-mediated injury |
| **173** | Konstantinos Koulousios | Screening of the Greek population for Fabry disease via pedigree analysis |
| **174** | Francyne Kubaski | Glycosaminoglycan levels in dried blood spots of patients with mucopolyasccharidoses and mucolipidoses |
| **175** | Francyne Kubaski | Hematopoietic stem cell transplantation for patients with mucopolysaccharidosis type II |
| **176** | Francyne Kubaski | LC/MS/MS measurement of glycosaminoglycans in amniotic fluid of a MPS VII fetus |
| **177** | Francyne Kubaski | Newborn screening for mucopolysaccharidoses: a pilot study of measurement of glycosaminoglycans by tandem mass spectrometry |
| **178** | Gé-Ann Kuiper | Incomplete biomarker response in mucopolysaccharidosis type I after successful hematopoietic cell transplantation |
| **179** | Thomas Kukar | Lysosomal production of granulins: implications for the pathogenesis of neuronal ceroid lipofuscinosis and frontotemporal dementia |
| **180** | Ludmila Kuzenkova | Subdural hematomas in a boy with mucopolysaccharidosis type IIIB |
| **181** | Heechun Kwak | MPS II model cell line by CRISPR-Cas9 technique |
| **182** | Florian Lagler | Interprofessional simulation training for emergencies in mucopolysaccharidoses |
| **183** | Dawn Laney | Sudden, pronounced height increase for adolescent and young adult males with Fabry disease |
| **184** | Heather Lau | Reported outcomes of 453 pregnancies in patients with Gaucher disease: an analysis from the Gaucher Outcome Survey |
| **185** | Heather Lau | Profile of patients with mucopolysaccharidosis type II without cognitive impairment who started idursulfase treatment aged >20 years: data on late treatment initiation from the Hunter Outcome Survey (HOS) |
| **186** | Lucia Lavalle | Heterogeneity in Fabry disease |
| **187** | Steven Le | Corpus callosum white matter myelination by neuroimaging and myelin composition analysis in murine mucopolysaccharidosis type I |
| **188** | Malte Lenders | Immunosuppressive effect on ERT inhibition in transplanted patients with Fabry disease |
| **189** | Malte Lenders | Renal function at ERT-naïve baseline predicts long-term outcome in Fabry disease |
| **190** | Malte Lenders | Treatment and clinical survey of females with Fabry disease in Germany |
| **191** | Valerie Lew | Fibrointimal thickening, elastin fragmentation and activated macrophages observed in coronary vasculature of a human mucopolysaccharidosis type VII patient |
| **192** | Sarah Lewis | Neurocognitive profiles of untreated Hunter syndrome |
| **195** | Renuka Limgala | Alterations in alpha-synuclein and parkin expression within PBMCs in patients and carriers of Gaucher disease with Parkinsonism |
| **196** | Renuka Limgala | Role of dendritic cells and NK cells in immune hypersensitivity reactions in patients with Fabry disease |
| **197** | Renuka Limgala | Effect of two different therapeutic interventions: SRT in comparison to ERT on immune aspects and bone involvement in Gaucher disease |
| **198** | Gloria Lin | Diffusion of intrathecally injected AAV9 in canine MPS VII: a peripheral nerve study |

| | | |
|---|---|---|
| **199** | Ales Linhart | Prompt agalsidase alfa therapy initiation after symptom onset is associated with improved renal and cardiovascular outcomes in the Fabry Outcome Survey |
| **201** | Valynne Long | Identification of lysosomal acid lipase (LAL) deficiency patients using key existing clinical data points focused on BMI, type 2 hyperlipidemia, and/or fatty liver disease |
| **202** | Laura López de Frutos | Comparison between several splicing bioinformatic predictors |
| **203** | Laura López de Frutos | A clinical case with a new damaging variant associated to Niemann-Pick disease type C |
| **204** | Jan Lukas | Ambroxol and bromhexine derivatives as pharmacological chaperones for mutant glucocerebrosidase |
| **205** | Su Han Lum | Long term survival and cardiopulmonary outcome in children with Hurler syndrome after haematopoietic stem cell transplantation in Manchester |
| **206** | Su Han Lum | A 20-year review of the changing pattern of graft failure in cord blood transplant for Hurler syndrome |
| **207** | Yi Lun | A novel recombinant human acid alpha-glucosidase, ATB200, leads to greater substrate reduction and improvement in Pompe disease-relevant markers compared to alglucosidase alfa in Gaa KO mice |
| **208** | Allen Lund | Long-term enzyme replacement therapy with velmanase alfa (human recombinant alpha-mannosidase) improves mobility in alpha-mannosidosis patients |

## Wednesday, February 15 – Poster Presentations

| Poster # | First Author | Abstract Title |
|---|---|---|
| **210** | Bernardus Machielse | VTS-270 for the treatment of Niemann-Pick type C disease |
| **211** | Christoffer Madsen | Fabry disease: renal function during long-term enzyme replacement therapy evaluated by gold standard GFR 51Cr-EDTA clearance |
| **213** | Elina Makino | Discovery of novel endogenous biomarkers in MPS I |
| **215** | Samantha Marcellus | Detection of three families with GLA p.A143T mutation and low α-galactosidase levels by newborn screening for Fabry disease |
| **216** | Reid Martin | Receptor-independent mechanisms of RTB lectin-mediated ERT delivery provide unique advantages in enzyme uptake capacity, transcytosis, and lysosomal correction |
| **217** | Ana Maria Martins | Natural history data from 182 female patients with Fabry disease in Latin America: A Fabry Registry analysis of disease burden |
| **219** | Ryuichi Mashima | Enzyme activities of six lysosomal diseases in a Japanese neonatal population |
| **220** | Maria Mattera | Validation of the shortened "Hunter Syndrome-Functional Outcomes for Clinical Understanding Scale" (HS-FOCUS) questionnaire |
| **222** | Casey McKenna | Expansion of the N215S phenotype in Fabry disease: a report of a large family with multi-organ symptoms in young adulthood |
| **223** | Benjamin McMahon | Impaired phagosome maturation in Gaucher macrophages provides a new target for therapeutic intervention target |
| **224** | Blanca Medrano Engay | Health outcomes research study on patients with type 1 Gaucher disease under substrate reduction therapy (SRT) |
| **225** | Julio Medrano-Montes de Oca | Pregnancy in a Fabry patient on enzyme replacement therapy - a case report |
| **226** | Atul Mehta | A global Delphi consensus initiative to facilitate early diagnosis of Gaucher disease type 1 and type 3: what are the phenotypic commonalities? |
| **227** | Olga Meijer | Processing and trafficking of N-acetyl-α-glucosaminidase in fibroblasts of patients with mucopolysaccharidosis type IIIB |
| **228** | Estrella Lizbeth Mellin Sanchez | Successful bone marrow transplantation treatment for a 10-year-old patient with late onset Krabbe disease |

| | | |
|---|---|---|
| **229** | Roman Melnyk | A novel platform to deliver lysosomal enzymes |
| **230** | Jaromir Mikl | Health care resource utilization by patients with Hunter syndrome in the UK hospital episode statistics (HES) database |
| **231** | Jaromir Mikl | The challenge of using Hospital Episode Statistics (HES) to identify a Hunter syndrome cohort in the UK |
| **232** | James Miller | The early stages of Fabry disease in an α-galactosidase A-deficient rat model |
| **233** | Weston Miller | Post-transplant intravenous laronidase augmentation for Hurler syndrome: anti-drug antibody, biochemical and donor hematopoietic graft response |
| **234** | Pramod Mistry | Long-term results of ENGAGE: a phase 3, randomized, double-blind, placebo-controlled, multi-center study investigating the efficacy and safety of eliglustat in adults with Gaucher disease type 1 |
| **235** | Takashi Miyajima | Lysosomal acid lipase deficiency high-risk screening of patients with fatty liver and dyslipidemia using DBS in Japan |
| **236** | Derek Moen | Scale-up processing of recombinant human glucosamine (n-acetyl)-6-sulfatase for the treatment of mucopolysaccharidosis type IIID |
| **237** | Lina Moreno | Pathogenic mutation at IDS gene on a newborn girl with clinical symptoms of Hunter syndrome (MPS II) |
| **238** | Lina Moreno | Short term impact from enzyme replacement therapy on patients with attenuated Hunter syndrome (MPS II) showing complex heart disease |
| **239** | Nina Movsesyan | Intravenous and intrathecal treatment with HP-β-CD reduced disease biomarkers in a patient with Niemann-Pick disease type C1 |
| **240** | Joseph Muenzer | A long-term extension study evaluating intrathecal idursulfase-IT in children with Hunter syndrome and cognitive impairment |
| **241** | Neslihan Mungan | Successful cardiovascular surgery experience and enzyme replacement therapy in type 3C Gaucher disease |
| **243** | Behzad Najafian | Podocyte globotriaosylceramide (GL-3) content strongly impacts age-dependent podocyte loss in ERT-naïve male Fabry patients |
| **244** | Kimitoshi Nakamura | High-risk screening trial of Gaucher disease for patients with neurological symptoms |
| **245** | Leyla Namazova-Baranova | 14 novel mutations of GLA gene as a result of selective Fabry disease screening in Russian Federation |
| **246** | Juana Navarrete | Multiple lysosomal diseases - newborn screening in a Mexican cohort of patients |
| **247** | Igor Nestrasil | White matter alterations in mucopolysaccharidosis type I assessed by automated DTI analysis |
| **248** | Stephanie Newman | Defective macrophage phagocytosis and particle clearance in Niemann-Pick disease type C1 mice |
| **249** | Kim Nickander | Oligosaccharidurias profiles by MALDI-TOF mass spectrometry and post-analytical interpretation using multivariate pattern recognition software |
| **250** | Stephanie Nijmeijer | Phenotypic severity in MPS IIIA strongly correlates with increase in residual SGSH activity in skin fibroblasts cultured at 30<sup>o</sup>C |
| **251** | Dau-Ming Niu | Reevaluate current routine histopathologic examinations for Fabry disease-not sensitive enough to identify early globotriaosylceramide accumulation in cardiomyocytes |
| **253** | Albina Nowak | Plasma lyso-Gb3: a useful biomarker for the diagnosis and treatment of Fabry disease heterozygotes |
| **256** | Burcu Öztürk Hişmi | Investigation of total oxidant status, antioxidant status and thiol/disulphide homeostasis in mucopolysaccharidosis type III patients |
| **258** | Samantha Parker | From testimonials to qualitative research embedded in an MPS IIIA observational study |
| **259** | Livia Paskulin | Taliglucerase alfa and type 1 Gaucher disease: a south Brazilian experience |
| **260** | Gabriela Pasqualim | MicroRNA and gene expression studies provide novel information to understand heart disease in women with Fabry disease |

| | | |
|---|---|---|
| **261** | Maria Pedrose | What would be the real prevalence & etiopathogeny of chronic liver diseases in patients with type 1 Gaucher disease? |
| **262** | Jordi Pérez-López | Clinical characteristics of patients with lysosomal diseases in a Spanish cohort of patients of 499 adult patients with inborn errors of metabolism from university hospitals |
| **264** | Karolina Pierzynowska | Stimulation of autophagy by genistein reduces levels of mutant huntingtin in cellular models of Huntington disease |
| **266** | Lynda Polgreen | Pilot study of the effect of adalimumab on pain, physical function, and musculoskeletal disease in mucopolysaccharidosis types I and II |
| **267** | Juan Politei | Chronic intestinal pseudo-obstruction: consider Fabry disease? |
| **268** | Juan Politei | Is joint pain a criteria for ERT indication in adult patients with MPS IVA and VI, even without significant visceral involvement? |
| **269** | Juan Politei | Dose-dependent response in Fabry treatment: decrement in plasma lyso-Gl3 after switching from alfa to beta |
| **270** | Laura Pollard | Novel UPLC-MS/MS oligosaccharide analysis for the diagnosis and monitoring of patients with glycoproteinoses |
| **271** | Fabiano Poswar | Aortic root dilatation in patients with mucopolysaccharidoses and the effect of enzyme replacement therapy |
| **272** | Anna Potier | Long-term stability of I2S, NAGLU, GALNS, ARSB, GUSB and TPP1 enzymes in frozen dried blood spots |
| **273** | Brianna Pruniski | Impact of newborn screening on families in the case of Pompe disease |
| **274** | Michael Przybilla | Quantification of ganglioside in brain tissue in the murine model of Hurler syndrome |
| **276** | Yana Puckett | Epidemiology of mucopolysaccharidoses (MPS) in the United States: challenges and opportunities |
| **277** | Ana Cristina Puga | Olipudase alfa for the treatment of acid sphingomyelinase deficiency (ASMD): safety and efficacy in adults treated for 30 months |
| **279** | Cathy Qin | Genotype/phenotype relationships in Fabry disease: developing an online interactive web-based data collection system |
| **281** | Uma Ramaswami | A randomized, phase 3B, open-label, parallel-group study of agalsidase beta in treatment-naive male pediatric patients with Fabry disease without severe symptoms (FIELD study): GL-3 clearance from superficial skin capillary endothelium |
| **282** | Uma Ramaswami | Non-neuronopathic Gaucher disease: a retrospective review comparing therapeutic outcomes of 2-week and 4-week enzyme replacement therapy |
| **283** | Uma Ramaswami | A randomized, phase 3B, open-label, parallel-group study of agalsidase beta in treatment-naive male pediatric patients with Fabry disease without severe symptoms (FIELD study): GL-3 clearance from kidney cells |
| **284** | Vanessa Rangel Miller | ConnectMPS: the global patient insights network for MPS, ML and related disorders |
| **285** | John Reynders | Novel LIPA mutations resulting in lysosomal acid lipase deficiency |
| **286** | Ross Ridsdale | Newborn screening (NBS) for metachromatic leukodystrophy (MLD): results from a study of 100,000 deidentified NBS samples |
| **287** | Charlotte Roberts | Multi-stakeholder engagement leading to access to treatment for MPS IVA (Morquio syndrome type A) - a model for the ultra rare disease community |
| **289** | Alexander Rodríguez-López | Cell uptake evaluation of human recombinant N-acetylgalactosamine-6-sulfate sulfatase (GALNS) produced in Pichia pastoris |
| **290** | Arndt Rolfs | Glucosylsphingosine concentration in the blood of Gaucher patients reflects the severity of GBA mutations |
| **291** | Tamanna Roshan Lal | Type 2 Gaucher disease - Where do we go from here? |
| **292** | Alejandra Rozenberg | Early intrathecal gene therapy extends lifespan and improves quality of life in a mouse model for infantile neuronal ceroid lipofuscinosis |
| **293** | Frank Rutsch | Expression of the lysosomal membrane protein LMBD1 is essential for the initiation of gastrulation |

| | | |
|---|---|---|
| **296** | Valentina Sanghez | Characterization on the cellular distribution of enzyme replacement therapy (ERT) under the influence of humoral immune response in MPS I mouse model |
| **298** | Andrea Schenone | Performance evaluation of alpha iduronidase assay in dried blood spots for the diagnosis of mucopolysaccharidosis type I |
| **299** | Raphael Schiffmann | Migalastat improves diarrhea in patients with Fabry disease: results from the FACETS double-blind, placebo-controlled phase 3 study |
| **300** | Raphael Schiffmann | Is Fabry disease frequently undiagnosed in patients with common heart disease? |
| **303** | Ida Vanessa Schwartz | Characteristics of 27 patients with type 3 Gaucher disease: a descriptive analysis from the Gaucher Outcome Survey |
| **304** | Susanne Seemann | Pharmacological intervention of ERAD partially restores the activity of mutant α-galactosidase A in Fabry disease |
| **305** | Caroline Sevin | Intrathecal delivery of recombinant human arylsulfatase A in children with late-infantile metachromatic leukodystrophy: an update following extended treatment |
| **308** | Adam Shaywitz | Design and rationale of ongoing observational and treatment studies for BMN 250: a novel enzyme replacement therapy for Sanfilippo syndrome type B |
| **309** | Rania Sheikh | Unrecognized immune thrombocytopenia in patients with Gaucher disease on long term therapy |
| **310** | Jin-Song Shen | Tetrahydrobiopterin deficiency in the pathogenesis of Fabry disease |
| **311** | Morgan Simmons | ThinkGenetic: identification of misinformation and educational gaps using an innovative and interactive website |
| **312** | Calogera Simonaro | Evaluation of pentosan polysulfate in mucopolysaccharidosis type IIIA mice |
| **313** | Cynthia Smith | Improvement of Fabry disease pain with calcium gluconate infusions given at time of agalsidase beta therapy |
| **314** | Elizabeth Smith | Please proceed with caution: a case series of the perils and pitfall of making a Fabry diagnosis |
| **316** | Alexander Solyom | Farber disease (acid ceramidase deficiency) epidemiology: literature review and patient cohort data indicate moderate and attenuated phenotypes are likely underrepresented in the medical literature and are underdiagnosed |
| **317** | Alexander Solyom | Farber disease: design of the first observational and cross-sectional cohort study capturing retrospective and prospective data on the natural history and phenotypic spectrum of patients, including novel methodologies for assessment of disease-specific symptoms |
| **319** | Hiroyuki Sonoda | Correction of central nervous system deficits in the mouse model of Hunter syndrome by recombinant iduronate 2-sulfatase crossing the blood-brain barrier |
| **320** | Russell Soon Jr. | Development of an electrochemiluminescent assay to measure LAMP2 in canine cerebrospinal fluid samples |
| **321** | Eser Sozmen | False positive diagnosis of lysosomal disease with dried blood spot (DBS) sample; leukocyte number as a challenging factor |
| **323** | S. Kaye Spratt | Comparison of systemic routes of administration for CLN3 delivery via scAAV9 |
| **324** | Karolina Stepien | A review of serum lipid profile in patients with mucopolysaccharidoses |
| **325** | Alta Steward | Sibling pairs with Gaucher disease discordant for Parkinsonism |
| **326** | David Stockton | Impact of earlier treatment on respiratory function in patients with late-onset Pompe disease: data from the Pompe Registry |
| **327** | Dean Suhr | Measuring sulfatide in blood enables newborn screening for metachromatic leukodystrophy (MLD) |
| **328** | Esteban Symonds | Fabry disease in adult dialysis patients in a community hospital |

| | | |
|---|---|---|
| **330** | Mireille Tallandier | In vitro and in vivo activity of IVA336, a potential substrate reduction therapy, in mucopolysaccharidosis models |
| **331** | Daisy Tapia | Variable clinical features in patients with Fabry disease |
| **334** | Abdellah Tebani | Implementation of an untargeted liquid chromatography ion mobility-mass spectrometry-based metabolomics method for inherited metabolic diseases investigation |
| **336** | Beth Thurberg | Autopsy pathology of infantile neurovisceral ASMD (Niemann-Pick disease type A): clinicopathologic correlations of a case report and review of the literature |
| **337** | Shunji Tomatsu | Activities of daily living for patients with Morquio syndrome type A |
| **338** | Shunji Tomatsu | Etiology of mucopolysaccharidoses |
| **339** | Takahiro Tsukimura | From diagnosis to follow-up of Fabry patients receiving enzyme replacement therapy in Japan |
| **340** | Anirudh Ullal | Flexible digital microfluidic platform to multiplex various combinations of enzymatic assays for newborn screening of Pompe, mucopolysaccharidosis types I and II, biotinidase deficiency and galactosemia disorders |
| **341** | Jeanine Utz | Infantile gangliosidoses: candidate outcome measures for future treatment trials |
| **342** | Filippo Vairo | The prevalence of lysosome-related diseases in a cohort of undiagnosed patients |
| **343** | Dora Vallejo Ardila | Regulatory units reconstruction approach identifies candidates for genetic risk of associated HCC in Gaucher disease |
| **344** | Ans van der Ploeg | Long-term efficacy of alglucosidase alfa in late-onset Pompe disease |
| **345** | Patricia Varela | Presence of the mutation c.1-769G>C in the promoter region of the GLA gene could be responsible for the α-galactosidase-A deficiency in Fabry patients |
| **347** | Corey Vural | A novel α-galactosidase A mutation in Fabry disease |
| **348** | Michael Wajnrajch | Occurrence of adverse bone events in adults and children with Gaucher disease treated with taliglucerase alfa |
| **349** | Stephen Waldek | New and improved therapy for cystinosis |
| **350** | Vanessa Wall | Carotid intima-media thickness and arterial stiffness are altered in patients with mucopolysaccharidosis type IVA |
| **351** | Susanne Walls | Improving home infusion follow-up |
| **352** | Hua Wang | Diagnosis, management and follow up of mucopolysaccharidosis: a five year review from a single mid-level institute |
| **353** | Ping Wang | Different ARSB variants causing mucopolysaccharidosis type VI in dogs |
| **354** | Ping Wang | Canine GM2-gangliosidosis (Sandhoff disease) caused by a 3 base pair deletion in the HEXB gene |
| **355** | David Warnock | PRX-102 for treating Fabry disease - immunogenicity and PK results from a phase 1-2 study |
| **357** | Melissa Wasserstein | Consensus recommendation on a diagnostic guideline for acid sphingomyelinase deficiency |
| **359** | Neal Weinreb | Transformation in pre-treatment presentations of Gaucher disease during the first two decades of imiglucerase enzyme replacement therapy: a report from the International Collaborative Gaucher Group Gaucher Registry |
| **360** | Richard Welford | Lucerastat, an iminosugar for substrate reduction therapy in Fabry disease: preclinical evidence |
| **362** | Kristen Wigby | Diagnosis of alpha mannosidosis after incidental finding of foamy cells on surgical specimen |
| **363** | Gisela Wilcox | Conventional measures of body composition may be less predictive in adult mucopolysaccharidoses patients - a small case series |
| **364** | Adina Wise | Parkinson’s disease penetrance in obligate carriers of SMPD1 mutations |

| | | |
|---|---|---|
| **365** | JIll Wood | How our approach for Sanfilippo syndrome could be a prototype for industrializing rare disease drug discovery and development |
| **366** | Michelle Wood | Guided growth surgery for genu valgum in mucopolysaccharidosis type VI |
| **367** | Michelle Wood | Assessment of functional mobility post haematopoietic cell transplant (HCT) in mucopolysaccharidosis type I using validated measures |
| **368** | Michelle Wood | Adjuvant oral salbutamol in treatment of juvenile Pompe disease: novel outcome assessment tool and initial report one-year efficacy in single case. |
| **369** | Kara Woolgar | Premedication and dose regimen for Gaucher type 1 patient with severe ERT intolerance |
| **370** | Amy Yang | Clinical characterization of variants of uncertain significance discovered through newborn screening for Pompe disease at one referral center |
| **371** | Shoji Yano | Detection of early end-organ damage by endothelial dysfunction with reactive hyperemia-digital peripheral arterial tonometry in patients with Fabry disease |
| **372** | Kwangchae Yoon | Identifying unmet needs for patients with Fabry disease through motivational interviewing |
| **373** | Elena Zaklyazminskaya | Prevalence of lysosomal diseases in the group of cardiac hypertrophy patients requiring cardiac surgery |
| **375** | Ari Zimran | Taliglucerase alfa: safety and efficacy across 6 clinical studies in children and adults with Gaucher disease |
| **376** | Ari Zimran | Pharmacokinetics, safety, and efficacy of rapid infusions of velaglucerase alfa in adult patients with Gaucher disease |
| **377** | Karim Zuberi | Cervical spine surgery in MPS IVA: a single center experience |
| **LB-01** | Walla Al-Hertani | Effect of ambroxol chaperone therapy on lyso-Gb1 levels in two Canadian patients with type 3 Gaucher disease |
| **LB-02** | Jeff An | Messenger RNA therapy for MUT methylmalonic acidemia (MMA): preliminary in vitro and in vivo efficacy |
| **LB-03** | Erika Augustine | Short-term administration of mycophenolate mofetil is well-tolerated in juvenile neuronal ceroid lipofuscinosis (CLN3) disease |
| **LB-04** | Edgar Barajas | Kidney injury in a family with Fabry disease due to mutation c.352c ≥ t(p.arg118cys) |
| **LB-05** | Elizabeth Braunlin | Cardiovascular risk in severe mucopolysaccharidosis type I after hematopoietic cell transplantation |
| **LB-07** | Selda Bulbul | Relationship between risk perception and treatment compliance |
| **LB-06** | Selda Bulbul | Identification of mutations in Turkish patients with Fabry disease |
| **LB-08** | Sharon Byers | Hepatic IGF1 production but not growth plate IGF1 production is altered in MPS VII mice in response to growth hormone stimulation |
| **LB-09** | Lisa Chalmers | The Australian ATYOURSIDE patient support experience |
| **LB-10** | Anthony Conway | In vivo genome editing via non-viral delivery of zinc finger nucleases results in supra-physiological levels of human iduronate 2-sulfatase in adult mice |
| **LB-11** | Glyn Dawson | Quantum dot delivery of enzyme for Batten disease therapy |
| **LB-12** | Maria de Castro Lopez | Preliminary safety and pharmacodynamic response data from a phase 1/2 study of ICV BMN 250, a novel enzyme replacement therapy for the treatment of Sanfilippo syndrome type B (MPS IIIB) |
| **LB-13** | Mireia del Toro | Unfarovable evolution of intracranial hypertension in pediatric patients with cystinosis |
| **LB-14** | Derrick Deming | Structural basis for the rational design of pharmacological chaperones for Pompe disease |
| **LB-15** | Shivakumar Devaiah | The RTB-lectin delivery module provides broad in vivo biodistribution and correction in multiple lysosomal disease mouse models |
| **LB-16** | Jordi Díaz-Manera | Quantitative muscle MRI is useful to follow-up late onset Pompe patients treated with enzyme replacement therapy |

| | | |
|---|---|---|
| **LB-17** | Ayse Eren | Pharmacological chaperone search for arylsulfatase A : in silico comparison of wild type and mutant structures |
| **LB-18** | Kaoru Eto | Neuronal ceroid lipofuscinosis (NCL) type 1 associated with type 2 diabetes |
| **LB-19** | Natalie Fraser | The Australian ATHOMETM infusion service experience |
| **LB-20** | Caio Gomes | PSAP and SCARB2 genes analysis as a differential diagnosis for Gaucher disease |
| **LB-21** | Abigail Hata | Gait analysis in children and adults with MPS IVA |
| **LB-22** | Wendy Heywood | Targeted proteomics of bloodspots for disease stratification of lysosomal storage disorders |
| **LB-23** | Sharon Hrynkow | Intravenous cyclodextrin trials and compassionate use in Niemann-Pick disease type C |
| **LB-24** | Pilar Irún | Comparison of glucosylsphingosine concentration and chitotriosidase activity as surrogated biomarkers in Gaucher disease, the Spanish experience |
| **LB-25** | Jinlong Jian | Progranulin recruits HSP70 to beta-glucocerebrosidase and is therapeutic against Gaucher disease |
| **LB-26** | Franklin Johnson | First-in-human preliminary pharmacokinetic and safety data on a novel recombinant acid-α-glucosidase, ATB200, co-administered with the pharmacological chaperone, AT2221, in ERT-experienced Pompe patients |
| **LB-27** | Nesrin Karabul | First German experiences with migalastat treatment in Fabry disease |
| **LB-28** | Zoheb Kazi | A prediction model to identify patients at high-risk of developing significant anti-drug antibodies (ADA): experience with infantile Pompe disease (IPD) on alglucosidase alfa utilizing acid α-glucosidase (GAA) mutations and HLA-type |
| **LB-29** | Jorge López | First report of late onset Pompe disease in an adult Mexican mixed-race patient |
| **LB-30** | Kira Lukina | Inherited thrombophilia in kidney transplant patient with Fabry disease and multiple thrombotic complications |
| **LB-31** | Maryana Marins | MAN2B1 gene analysis as a differential diagnosis for mucopolysaccharidosis types I, II and VI in the Brazilian population |
| **LB-32** | Sergey Moiseev | Clinical features of Fabry disease in dialysis patients |
| **LB-33** | Vagishwari Murugesan | Glycoprotein non-metastatic melanoma B as a novel biomarker to assess disease severity in Gaucher disease |
| **LB-34** | Jun Okada | A Fabry male patient associated with recurrent giant skin ulcer in lower extremity |
| **LB-35** | Guillem Pintos-Morell | Evaluation of the effects of a ketogenic diet on mucopolysaccharidosis type IIIB mice |
| **LB-36** | Daniel Rob | Uric acid as a marker of mortality and morbidity in Fabry disease |
| **LB-37** | Volkan Seyrantepe | Abnormal brain ganglioside accumulation triggers neuroinflammation in early onset Tay-Sachs disease mice model |
| **LB-38** | Joseph Shen | A perplexing case of presumed lysosomal acid lipase deficiency - phenotypic and biochemical information, and response to enzyme replacement therapy, are consistent with this diagnosis, but enzymatic studies are equivocal |
| **LB-39** | Lachlan Smith | Targeting wnt/β-catenin signaling to enhance bone formation in mucopolysaccharidosis type VII dogs |
| **LB-40** | Denize Souza | Cognitive findings of a series of patients with mucopolysaccharidosis |
| **LB-41** | Ying Sun | Tissue localization of glycosphingolipid accumulation in a Gaucher disease mouse brain by LC-ESI-MS/MS and high resolution MALDI imaging mass spectrometry |
| **LB-42** | Kazuya Tsuboi | Efficacy and safety of enzyme replacement therapy with agalsidase alfa in 36 treatment-naïve Fabry disease patients |
| **LB-43** | Alfredo Uribe Ardila | Enzymatic assays for alfaglucosidase in patients with Pompe disease, results from ten years of high risk screening in Colombia |
| **LB-44** | Christine White | Multidisciplinary approach to diagnosis and clinical management of Gaucher patients transitioning to adult care |

| | | |
|---|---|---|
| **LB-45** | Chester Whitley | First-in-human clinical trial of intravenous SBC-103 in patients with mucopolysaccharidosis type IIIB: safety, tolerability, cerebrospinal fluid, heparan sulfate levels, brain structural MRI, and neurocognitive evaluations after 1 year of the open-label phase I/II study |
| **LB-46** | Hunter Wilson | Characterization of the early cardiac phenotype in Fabry disease |