Printed posters will be displayed in the Exhibit Hall (Seaport Ballroom), with each presenter assigned a specific day. Daily poster receptions, including live Q&A with the presenters, will take place as outlined below.

Basic Science & Late-Breaking Science Posters (LB-01 to LB-34) will be presented on **Tuesday, February 4** from **15:30-17:30 PST**

Translational Research & Late-Breaking Science Posters (LB-35 to LB-62) will be presented on **Wednesday, February 5** from **15:30-17:30 PST**

Clinical Applications & Rapid-Fire Posters will be presented on **Thursday, February 6** from **15:30-17:30 PST**

*** **Contemporary Forum Posters** will be presented each day based on the abstract category. **Late-Breaking Science Posters** (poster numbers starting with an LB) will be presented on Tuesday, February 4th and Wednesday, February 5th.

ePosters will be available to all registered attendees via the WORLD*Symposium* mobile app beginning at 05:00 PST on Tuesday, February 4, 2025, and will remain accessible throughout the live meeting. On Demand registered attendees can access ePosters from February 12 to March 14, 2025.

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 WORLDSymposium. The first author of the submitted abstract will be listed as the presenting author on the Preliminary Program, Agenda, and Poster List.

Basic Science & Late-Breaking Science – Poster Session I – Tuesday, February 4, 15:30-17:30 PST

#	Author	Title	Kiosk
4	Pratikshya Adhikari	AAV9-based gene replacement therapy targeting the root cause for the treatment of MPS IIID in mice	1-A
5	Anusha Aditya	Direct reprogramming of oligodendrocytes from globoid-cell leukodystrophy patient primary fibroblasts	1-B
6	Noor Ul Ain	Investigating the genomic basis of phenotypic diversity among siblings in Gaucher disease	2-A
8	Julia Alton	Exploring the lived experiences and care perceptions of females living with Fabry disease in the US, Canada, and Japan	2-B
9	Julia Alton	Exploring the demographics of the diagnosed Fabry disease population in Canada	3-A
11	Isidro Arevalo-Vargas	Clinical variability in Gaucher disease associated with the c.1880T>G (D409H) variant in GBA1	3-B
12	Isidro Arevalo-Vargas	Lipid profiling in dried blood spots: A tool for diagnosing lysosomal diseases	4-A
13	Isidro Arevalo-Vargas	Validation of “in silico” predictors for analyzing variants of uncertain significance in lysosomal disorders	4-B
14	Jessica Arozqueta-Basurto	Hematopoietic stem cell transplantation for lysosomal disorders: Unraveling the mechanisms for CNS repopulation	5-A
15	Imane Assiri	First comprehensive identification of urinary sphingomyelin species in Niemann-Pick disease patients using UHPLC-MS/MS	5-B
18	José Avendaño-Ortiz	Could the microbiota contribute to the symptomatology of Sanfilippo syndrome? A report in two affected sibligs	6-A
19	Alireza Ayoubi	Imaging flow cytometry (IFC) a novel tool for automated and standardized quantification of urine podocytes and their globotriaosylceramide (GL3) content in Fabry disease	6-B
22	Françoise Piguet	Development and validation of an intravenous AAV gene therapy for mucopolysaccharidosis type IIIB in mouse and dog model of the pathology	7-A
30	Abdelaati Berrachid	The bioactive potential of Rosa damascena mill: Preventive chelating effects and the role of ecosystem dynamics in inherited diseases	7-B
38	Nika Breznik	RNA sequencing reveals the involvement of immune pathways in the Fabry disease nephropathy progression	8-A

40	Laura Buch	Alpha mannosidosis: One center's experience with three patient's diagnoses, interventions, and biomarkers	8-B
50	Magdalena Ceron-Rodriguez	Elastoresonance as a surveillance tool in a patient with acid sphingomyelinase deficiency undergoing treatment with olipudase alfa	8-C
51	Magdalena Ceron-Rodriguez	Is ambroxol a real protector for neuronopathic Gaucher disease	8-D
66	Dominic D'Agostino	Ketone metabolic therapy to improve muscle function and coordination in a mouse model of infantile onset Pompe disease	7-C
68	Julia Dao	Age-related inflammatory markers and early-onset osteoporosis in women with Gaucher disease	7-D
74	Karolina Stepien	Secondary mitochondrial dysfunction in three lysosomal disorders	6-C
75	Gabrielle Dineck Iop	Screening of alpha-mannosidosis in patients with suspected mucopolysaccharidosis (MPS) who tested negative for MPS in a Brazilian reference center	6-D
81	Cathy Duong	Polycystic kidney disease complicates renal pathology in two families with Fabry disease	5-C
83	Hatim Ebrahim	Audit of patients screened for lysosomal disorders at the Royal Free London Hospital	5-D
84	Sana El Foutat	Assessment of trace elements in the serum of epileptic patients with inherited metabolic diseases: Focus on zinc and copper	4-C
89	Jacob Favret	Dissecting the role of oligodendroglial NFkB signaling in the pathogenesis of Krabbe disease	4-D
92	Allan Feng	A novel murine model for neuronopathic Gaucher disease	3-C
101	Jessica Gambardella	Pegunigalsidase alfa cellular uptake, stability, and potential impact on the energetics of Fabry disease cells	3-D
104	Michael Gelb	Toward newborn screening of cystinosis	2-C
113	Domingo Gonzalez-Lamuño Leguina	Reversible deficiencies of arylsulfatase B and alpha-galactosidase in Duchenne muscular dystrophy	2-D
114	Sarah Gosling	Co-design of a physical activity and sedentary behaviour intervention for adults with Fabry disease	1-C
116	Vincenza Gragnaniello	Inflammation and impaired autophagy in Gaucher disease type 1 infants diagnosed by newborn screening	1-D
120	Jacky Guerrero	Use of the polymer PP6D5 and CRISPR-nCas9 in gene therapy for Tay-Sachs disease	16-A
134	Angelica Maria Herreno Pachon	CRISPR/Cas9-edited hematopoietic stem cells rescue MPS IVA fibroblasts phenotype	16-B
145	Shih-Chang Hsueh	A cyclic oligosaccharide structure as a novel therapeutic strategy for Krabbe disease	15-A
152	Majdolen Joleen Istaiti	Excess risk of monoclonal gammopathy of undetermined significance (MGUS) in a screened cohort of patients with Gaucher disease	15-B
153	Margarita Ivanova	Inflammatory biomarkers underlying sex differences in Fabry disease related cardiomyopathy	14-A
154	Margarita Ivanova	Pilot study to assess immune biomarkers and growth factors related to bone pathology in pediatric patients with Gaucher disease	14-B
157	Maroua Jakani	Altered ganglioside pattern in the infantile form of GM1 gangliosidosis identified using reversed-phase chromatography combined with thin-layer chromatography	13-A
159	Xuntian Jiang	Secondary accumulation of lyso-platelet activating factors in lysosomal diseases	13-B
160	Eric Jones	Next-generation corrector therapy for Fabry disease	12-A
167	Neil Kasaci	Unraveling the cytotoxic effects of circulating Lyso-Gb1 and Lyso-Gb3: Linking immune activation and cellular dysfunction in Gaucher and Fabry diseases	12-B
179	Stefanie Krassnig	Compound muscle action potential as translational biomarker in a mouse model of Pompe disease	11-A
184	Meghana Kushwaha	Exploring the death and signaling of galactosylceramidase-deficient oligodendrocytes	11-B
194	Lucia Lavalle	Sex differences in alpha galactosidase protein processing and its impact on disease severity in Fabry disease	10-A

196	Andres Leal	Uncovering mitochondrial disturbances in MPS IVA chondrocytes	10-B
197	Chris Lee	Residual galactosylceramidase activity correlates with psychosine levels in GALC missense variants cell model of Krabbe disease	9-A
198	Dau-Ming Niu	Early potentially irreversible cardiac damage in Fabry disease precedes Gb3 inclusion body formation	9-B
201	Malte Lenders	Impact of neutralizing antibodies on the pharmacokinetics of pegunigalsidase alfa in patients with Fabry disease	9-C
203	Daniel Lewi	Spot the stop: A multi-channel campaign to improve early diagnosis of Tay-Sachs disease among healthcare professionals	9-D
204	Daniel Lewi	The impact of a clinical toolkit on post-diagnosis care for Tay-Sachs and Sandhoff Disease: A comparative study of family experiences	10-C
210	Tina Löffler	Longitudinal NF-L measurements in in vivo CSF samples for tracking pathology progression in mouse models of Gaucher and Niemann-Pick diseases	10-D
211	Tina Löffler	Comparative analysis of human and mouse fibroblast lines as in vitro models for lysosomal storage diseases: A focus on enzyme activity and lipid metabolism	11-C
212	Sergio Londono	Evaluating the impact of agalsidase beta on the cost of chronic kidney disease progression in Fabry disease	11-D
220	Angela Martin Rios	Clinical and biochemical phenotypes of β-mannosidosis, and a new association with white matter disease	12-C
226	Ana Méndez Silva	Functional panorama and evolution of Pompe disease patients in Latin America	12-D
242	Samira Najeh	Assessment of carnitine deficiency in Moroccan patients: A high sensitivity and specificity enzymatic approach	13-C
247	Katherin Niño-Traslaviña	Cellular and functional characterization of CRISPR/Cas9-generated NAGLU-deficient cell models for MPS IIIB	13-D
254	Petra Oliva	Importance of including α-mannosidosis in a combined testing protocol with MPS in patients suspected of a mucopolysaccharidoses	14-C
255	Petra Oliva	Update on high-risk population screening for neuronal ceroid lipofuscinoses (NCL1 and NCL2)	14-D
256	Petra Oliva	Acid sphingomyelinase deficiency (ASMD): Genotype-phenotype correlation and variant frequency	15-C
257	Andrew Olson	Remote technologies for monitoring gait changes due to neurodegenerative diseases in children: Evaluation, issues, and future promise	15-D
264	Magali Pettazzoni	Acid sphingomyelinase deficiency (ASMD) and Niemann-Pick disease type C: Cytologic features of bone marrow and peripheral blood smears can guide the diagnosis	16-C
266	Magali Pettazzoni	Kinetic evolution of plasma biomarkers in acid sphingomyelinase deficiency patients under enzyme replacement therapy: A French experience	16-D
267	Nathan Phan	Biomarker potential of allele-specific extracellular vesicles in Gaucher disease	17-A
271	Luisa Pimentel Vera	Correction of GD1 pathology by genome edited murine hematopoietic stem cell transplantation	17-B
284	Anna Reinelt	Advancing cardiac disease modeling in Fabry cardiomyopathy by utilizing patient-derived induced pluripotent stem cells, heart organoids, and engineered heart tissue	18-A
285	Anna Reinelt	Kidney organoids from patient-derived induced pluripotent stem cells for the investigation of Fabry nephropathy	18-B
290	Sonia Roca-Esteve	Understanding the role of neutrophil extracellular traps in vascular complications of lysosomal disorders	19-A
291	Candela Romano	Bilateral avascular necrosis: A rare complication in Fabry disease	19-B
296	Samreen Safdar	Missed diagnosis of Fabry disease leading to end-stage renal disease: A case report	20-A
311	Irene Serrano Gonzalo	Predictive model for estimating the risk of bone mineral loss in Gaucher disease	20-B

312	Irene Serrano Gonzalo	Exploring the correlation between miRNA and osteonecrosis in Gaucher disease	21-A
313	Irene Serrano Gonzalo	Retrospective study to analyze and validate a set of potential predictive markers to identify lysosomal acid lipase deficiency	21-B
314	Irene Serrano Gonzalo	Evaluating lipocalin-2 as a biomarker for lysosomal acid lipase deficiency	22-A
316	Sonia Antonella Sgro	Real world reflections of the patient odyssey in alpha-mannosidosis: Insights and challenges in diagnosis from caregiver interviews	22-B
320	Yuki Shiro	Identification of a pathological association between neuronal ceroid lipofuscinosis type 10 and loss of anti-aggregate activity of pro-cathepsin D	23-A
323	David Smerkous	Quantification of globotriaosylceramide (GL3) in peritubular capillary endothelial cells (PTCEC) in kidney biopsies from patients with Fabry disease using machine learning	23-B
329	Zinandre Stander	HGSNAT - a 'chip-off-the-old-block', or a double entendre with an unusual clinical course?	24-A
335	Diego Suarez	Study of GM2 gangliosidosis by understanding the pathophysiology in a knockout cellular model generated by CRISPR-Cas9 technology	24-B
336	Leah Svarny	The utility of neurofilament light chain in conjunction with clinical biomarkers for early detection and prediction of disease burden and severity in neuronopathic Gaucher disease	24-C
337	Marcia Terluk	Investigating mitochondria-lysosome cross-talk in Gaucher disease cells	24-D
342	Cristian Verano Guevara	Establishment of a method for detecting glycosaminoglycans using liquid chromatography coupled with tandem mass spectrometry (LC-MS/MS) in Colombia	23-C
350	WM Subadra Wanninayake	Potential increased risk of renal carcinoma in Fabry disease	23-D
353	Rebecca Whiteley	A single centre review of pre-medications used in patients receiving enzyme replacement (ERT) infusions with evaluation of risk of adrenal insufficiency	22-C
354	Chester Whitley	PS Gene-editing system corrects the CNS with blood-brain barrier penetrant ApoE-enzymes	22-D
360	Shu Xing	Apoe-Abca1 axis is involved in the pathogenesis of Gaucher disease	21-C
361	Nirbhay Yadav	Non-invasive magnetic resonance imaging monitoring of glycogen accumulation in a mouse model of Pompe disease	21-D
365	Ewa Ziolkowska	Treatment of dysphagia associated pathologies in CLN3 deficient mice via gene therapy	20-C
LB-01	Hamdan Alshahrani	Successful management of juvenile idiopathic arthritis in a pediatric patient diagnosed with mucopolysaccharidosis type IIIC (Sanfilippo syndrome type C) using adalimumab: A case report	20-D
LB-02	Jose Alvarez Gonzalez	Oral enzyme therapy for lysosomal diseases	19-C
LB-03	Alireza Ayoubi	Imaging flow cytometry (IFC): A novel tool for automated and standardized quantification of urine podocytes and their globotriaosylceramide (GL3) content in Fabry disease	19-D
LB-04	Maryam Banikazemi	A phase 4 study to evaluate the safety and tolerability of higher infusion rates of agalsidase beta to shorten infusion duration in Fabry disease - interim analysis	18-C
LB-05	Conan Donnelly	Investigating the real-world experience of patients with Niemann-Pick disease type C (NPC) and their caregivers	18-D
LB-07	Jian Chen	Automated fluorescence enzymatic assays for the screening of lysosomal disorders	17-C
LB-08	Matthieu Colpaert	GAA-based therapeutics for the treatment of multiple glycogen storage diseases	17-D
LB-09	Kathleen Coolidge	The view from the patient advocate: Identifying and overcoming the challenges to collecting data from different LD patient communities	32-A
LB-10	Yanya Ding	Age related and sex specific progression of auditory neurophysiological deficits in the $Cln3^{-/-}$ mouse model	32-B
LB-11	Rana Dutta	Safety and tolerability of chenodeoxycholic acid in pediatric patients with cerebrotendinous xanthomatosis (RESTORE): An open-label phase 3 study	31-A
LB-13	Lucas Ferreira Teixeira	Gaucher disease type 3 and IgA vasculitis: A case report	31-B
LB-14	Lucas Ferreira Teixeira	Cholelithiasis in Gaucher Disease: A cohort study	30-A

LB-15	Lucas Ferreira Teixeira	Acid sphingomyelinase deficiency and lung fibrosis: A case report	30-B
LB-18	Esteban Gonzalez	Safety and tolerability of repeated nasal administrations of liposomes carrying the CRISPR-CAS9 system for mucopolysaccharidosis type I gene editing: A study in non-human primates	29-A
LB-19	Nadene Henderson	Lessons learned - transition from research studies to clinical care	29-B
LB-20	Niels Høeg Brandt-Jacobsen	Trends in loss of renal function following the initiation of Fabry-specific treatment 2001-2023: Sub-study from the National Danish Fabry Cohort	28-A
LB-21	Sunhee Kang	Effect of GC200, a novel orally available pharmacological chaperone (PC), on GM1-ganglioside reduction in the brains of hR201C Tg, Glb1KO/ mice as a promising preclinical candidate	28-B
LB-22	Jin Young Kim	Beneficial effects of HM15421, a novel long-acting alpha-galactosidase A analog, following a switch from the conventional enzyme replacement therapy in a symptomatic Fabry mouse model	27-A
LB-23	Sujeong Kim	Intracerebroventricular administration of recombinant human heparan-N-sulfatase (GC1130A) demonstrates effective brain biodistribution in rodents	27-B
LB-24	Wonki Kim	Comparative efficacy of conventional ERT drugs and HM15421, a novel long-acting alpha A analog, in a symptomatic Fabry mouse model	26-A
LB-25	Tudor Lazaruc	Unmasking Fabry disease in a young boy: A multisystem enigma	26-B
LB-26	Kathleen Coolidge	Aligning stakeholder terminologies and priorities in clinical trials to enhance outcomes for lysosomal disease (LD) patients	25-A
LB-27	Daniel Lewi	The Global LSD Collaborative - uniting advocacy to elevate care for Lysosomal communities worldwide	25-B
LB-28	Hsiang-Yu Lin	Newborn screening for mucopolysaccharidosis type I: Nine years' experience in Taiwan	25-C
LB-29	Charles Lourenco	Double the trouble: Dual diagnosis in Fabry disease may contribute to atypical phenotypes	25-D
LB-30	Charis Ma	Analysis of GBA1 isogenic iPSC-derived dopaminergic neurons and microglia to investigate the pathogenesis of GBA1-associated Parkinson disease	26-C
LB-31	Clarisa Maxit	Expanding the spectrum of manifestations in infant-onset Pompe disease: Involvement of the central nervous system	26-D
LB-32	Pramod Mistry	Peripheral enzyme replacement therapy in type 2 Gaucher disease: From mouse models to clinical translation in the era of newborn screening	27-C
LB-33	Kazuki Miyauchi	Treatment with a transferrin receptor-targeted β-hexosaminidase A, prolongs life span of GM2 gangliosidosis mice	27-D
LB-34	Daisy Ng-Mak	Natural history and burden of disease among patients with late-onset GM2 gangliosidoses in France	28-C

Translational Research & Late-Breaking Science – Poster Session II – Wednesday, February 5, 15:30-17:30 PST

#	Author	Title	Kiosk
1	Magy Abdelwahab	Long term follow up of bone mineral density in Egyptian Gaucher disease type 3 patients on enzyme replacement therapy	1-A
7	Gheona Altarescu	Preimplantation genetic testing (PGT) to reduce the risk for GBA-related Parkinson disease: Expanding of applications for embryo selection	1-B
16	Christiane Auray-Blais	Feasibility of newborn screening of Triple H syndrome using dried urine spots analyzed by flow-injection mass spectrometry	2-A
21	Françoise Piguet	Cell-based device provides effective therapeutic strategy to treat the metachromatic leukodystrophy	2-B
23	Nicholas Bascou	Parent-reported disease burden in Krabbe disease: Evaluating outcomes of hematopoietic stem cell transplant	3-A
24	Karen Bean	Exploring the net monetary benefit of implementing newborn screening for metachromatic leukodystrophy in California	3-B
25	Eliane Beauregard-Lacroix	Phenotypic and genotypic expansion of mucopolysaccharidosis type II: A case with IDS c.817C>T variant detected through newborn screening	4-A
34	Brian Bigger	Long-term HSC gene therapy in mucopolysaccharidosis type IIIB mice corrects disease with no evidence of insertional mutagenesis despite high vector copy numbers	4-B
35	Nidal Boulos	Audiology assessment of participants in CAMPSIITE®, a phase I/II/III study of investigational RGX-121 in neuronopathic MPS II	5-A
36	Elizabeth Braunlin	Transitioning individuals with MPS to adult care	5-B
39	Amy Brooks	Five years of newborn screening for mucopolysaccharidosis type I in Virginia	6-A
41	Alberto Burlina	Neonatal screening for Fabry disease and long-term follow-up: The role of plasma globotriaosylsphingosine (LysoGb3) assay	6-B
43	Barbara Burton	Evaluation and follow-up of newborns screening positive for mucopolysaccharidosis type II: results from an international modified Delphi consensus	7-A
48	Betul Celik	All-in-one lentiviral vectors improving bone abnormalities in MPS IVA mice	7-B
49	Betul Celik	In vivo direct bone targeting lentiviral gene therapy for MPS IVA murine model	8-A
52	Dau-Ming Niu	Applications of a rapid real time analysis system for whole genome/exome sequencing in newborn screening	8-B
53	Chong Kun Cheon	Evaluation of potential drug development and therapeutic approaches for mucopolysaccharidosis type II	8-C
56	Megan Clarke	Screenplus: An assay-based multi-tiered testing model for expanded NBS	8-D
57	Kristin Clinard	North Carolina experience with newborn screening for mucopolysaccharidosis type II (Hunter syndrome)	7-C
59	Kristin Clinard	Challenges of disease severity prediction in an MPS II asymptomatic individual with a novel IDS variant: A unique NBS case with familial mosaicism	7-D
60	Pasqualina Colella	Genome-edited autologous stem cell transplantation with enhanced brain conditioning to correct progranulin deficiency	6-C
LB-62	Tanya Collin-Histed	Global collaborative effort for Gaucher disease: A step towards improving healthcare	6-D
64	Gerson Da Silva Carvalho	Newborn screening for six lysosomal diseases in the Brazilian population: The first year of experience in the Unified Health System of the Federal District, Brazil	5-C
67	Uma Ramaswami	Gaucher disease - correlation of lyso-Gb1 with biochemical therapeutic goals	5-D
69	Nicole Miller	Mucopolysaccharidosis type VII (MPS VII): A novel, online GUSB gene variant database	4-C
71	Patrick Deegan	Reaching consensus on comprehensive and achievable monitoring for adults with late-onset Pompe disease in the UK	4-D

72	Patrick Deegan	Algorithmic case finding approaches for Gaucher disease type 1 in primary care records	3-C
77	Hung Do	What is in store for Pompe disease therapy?	3-D
78	Aimee Donald	Two hundred and fifty cases of "Gaucher disease type 2": A novel system of clinical categorization and evidence of genotype:phenotype correlation	2-C
79	Taraka Donti	Measuring non-reducing terminal glycosaminoglycan fragments increases specificity and differentiates mucopolysaccharidosis type I (MPS I) from mucopolysaccharidosis type II (MPS II)	2-D
88	Martha Faraguna	A new insight in Pompe disease: Pharmacokinetic variability after recombinant alpha-glucosidase infusion	1-C
95	Kathleen Flynn	Engaging patients and Patient Advocacy Organizations (PAOs) in the rare disease drug development process	1-D
96	Kathleen Flynn	"Fact checking" with the patient community: An industry-advocacy collaboration for optimizing trial design	16-A
97	Andrew Freiman	Enhancing family experiences and emergency protocols: Integrating genetic counseling in lysosomal disease clinics	16-B
100	Ellen Fung	Baseline data from the proof of concept study to evaluate the safety and early efficacy of vosoritide for the treatment of growth deficits in MPS IVA and VI	15-A
106	Gisela Linthorst	Seizures. Significant co-morbidity for patients and families living with NPC	15-B
115	Russell Gotschall	M021: A novel drug candidate for Pompe disease	14-A
117	Christina Grant	Cardiomyopathy and reduced volume enzyme replacement therapy in patients with MPS I	14-B
123	Gwen Gunn	Newborn screening outcomes for MPS I in Georgia	13-A
128	Caroline Hastings	Genotypic heterogeneity in GM1 gangliosidosis	13-B
139	Robert Hopkin	Miglustat: A first-in-class enzyme stabilizer for late-onset Pompe disease	12-A
143	Kerren Hosking	Data priorities of an international community to support research and improve Sanfilippo syndrome outcomes	12-B
148	Aya Ibrahim	Fabry disease in the Republic of Ireland: Insights from the newly established multidisciplinary team clinic at Mater Misericordiae University Hospital	11-A
156	Skyler Jackson	Rare disease difference maker program	11-B
158	Elizabeth Jalazo	Inconclusive endogenous nonreducing end GAG analysis in an infant detected on NBS for MPS I	10-A
162	Deepthy Joseph	Clinical depression and impact on patients with lysosomal diseases (LDs): Service evaluation from a specialist centre using the Beck's Depression Inventory (BDI-II)	10-B
166	Audrey Kao	Serial brain MRI volumetrics and tractography of atypical CLN2 patients receiving ICV cerliponase alfa treatment	9-A
174	Ibrahim Khoja	The impact of newborn screening on the age of patients referred for testing and with confirmed diagnoses of Pompe disease and mucopolysaccharidosis type I	9-B
178	Tracy Kornafel	Physical therapy outcomes in young children with late-onset Pompe disease identified by newborn screening	9-C
180	Stefanie Krassnig	Application of preclinical gene therapy approaches	9-D
181	Akhil Kulkarni	A novel AAV-based gene therapy strategy reverses lethality in a murine model of neuronopathic Gaucher disease	10-C
182	Francyne Kubaski	Inconclusive urine glycosaminoglycan results in MPS I and MPS II newborn screening cases using both methanolysis and endogenous NRE GAG methods	10-D
183	KeriAnn Kuperman	Newborn screening for Pompe disease: The 7 year Washington, DC experience and the emerging phenotype of juvenile late onset Pompe disease	11-C
190	Kyle Landskroner	Nizubaglustat, a novel GCS and NLGase inhibitor significantly increases survival in a mouse model of GM1 gangliosidosis	11-D

195	Lucia Lavalle	Exploring the clinical impacts of genetic variants of unknown significance in Fabry disease	12-C
202	Daniel Lewi	Understanding ancestry in the diagnostic process for Tay-Sachs disease	12-D
207	Alaena Lim	Further characterization of the GLA p.A143T phenotype through leveraging the genomic database, All of Us	13-C
208	Hsiang-Yu Lin	Newborn screening for mucopolysaccharidosis type IVA: Four years' experience in Taiwan	13-D
215	Troy Lund	CSF GAG reduction is associated with a decrease in inflammation	14-C
227	Caitlin Menello	Newborn screening for Gaucher disease: The New Jersey experience	14-D
244	Igor Nestrasil	Phenotype differentiation and white matter preservation by early hematopoietic cell transplantation - free-water diffusion tensor MRI study of the brain in mucopolysaccharidosis type I patients	15-C
245	Sarah Neuhaus	A phase 1/2 study of LY3884961 (PR001) an AAV9-based gene therapy for Gaucher disease type 2 - a clinical update from the PROVIDE trial	15-D
246	Sarah Neuhaus	Phase 1/2 dose-finding study to evaluate systemic administration of an AAV9-based gene therapy for peripheral manifestation of Gaucher disease - The PROCEED study	16-C
251	Ilyas Okur	Expanded pilot newborn screening for lysosomal disorders: First results from Turkey	16-D
259	Katrina Paleologos	Screenplus parental perspectives on universal and consented newborn screening based on treatability	17-A
260	Francis Pang	Validation of quality-of-life states in late-infantile and early-juvenile metachromatic leukodystrophy	17-B
261	Marzia Pasquali	Challenges in the diagnosis of mucopolysaccharidosis type IVA	18-A
262	Siddhant Patel	Association of GBA1 genotype with disease severity in treatment-naïve patients with Gaucher disease type 1	18-B
263	Lucie Perillat	A conceptual model and practical guidance for the development, administration, and evaluation of individualized therapies	19-A
269	Agustin Pijierro Amador	Project for the education and diagnosis of Gaucher disease and acid sphingomyelinase deficiency (PREDIGA-2)	19-B
274	Edina Poletto	Investigational new drug-enabling studies for genome-edited hematopoietic stem cells to treat mucopolysaccharidosis type I	20-A
277	Michael Przybilla	Penetrating the blood-brain barrier: Utilizing the PS gene editing system to encode a novel fusion β-galactosidase for the treatment of GM1 gangliosidosis	20-B
278	Michael Przybilla	Improving blood-brain barrier penetration in Hurler syndrome using an IDUA-ApoE fusion enzyme delivered via the PS Gene Editing System	21-A
280	Karthikeyan Rajagopal	In vitro development and in vivo evaluation of intra-articular GUSB mRNA therapy for mucopolysaccharidosis type VII	21-B
281	Deepa Rajan	Natural history of hearing loss in children with mucopolysaccharidoses type II (Hunter syndrome)	22-A
282	Renuka Raman	Pre-clinical development of an enzyme replacement therapy for CLN1 Batten disease: Process development, stability and uptake studies	22-B
289	Jorge Cebolla	Marklald: Plasma biomarkers as key indicators for monitoring patients with lysosomal acid lipase deficiency treated with sebelipase alfa. Preliminary findings from a multicentre cohort study in Spain	23-A
292	Aviva Rosenberg	Challenges of Gaucher disease type 2 and 3 patients to access treatment due to FDA label	23-B
293	Sam Ruesing	Stability of rhGALNS enzyme in buffers for sustained release applications	24-A
295	Krystyna Rytel	A genome-wide CRISPR activation screen to identify beta-glucocerebrosidase modifiers	24-B
299	Sampurna Saikia	Immune modulation for AAV-9 gene therapy by oral administration of peptides for GALNS enables the vector re-administration in MPS IVA	24-C
304	Roselena Schuh	Evaluation of off-target events after an intravenous injection of liposomal CRISPR/Cas9 complex in vivo	24-D

307	Kathryn Schwan	What's the plan? A single center's experience navigating through the challenges of newborn screening identified attenuated mucopolysaccharidosis type I (MPS I) cases	23-C
308	Markus Schwarz	Differential diagnosis for mucopolysaccharidoses (MPSs) and subtype prevalence in Europe and Middle East	23-D
315	Caroline Sevin	Oral leriglitazone as an effective therapeutic strategy to treat neuro inflammation in metachromatic leukodystrophy	22-C
317	Hadeel Shammas	Arimoclomol upregulates expression of genes belonging to the coordinated lysosomal expression and regulation (CLEAR) network	22-D
321	Heather Shorten	Pompe disease community members face 2.5x higher risk of mental health issues compared to the general population	21-C
325	Sean Ekins	Development and preliminary analysis from the Batten disease CLN1 registry	21-D
326	Fabrício Soares	Early diagnosis and pre-symptomatic treatment of infantile-onset Pompe disease: First report from public newborn screening in Brazil	20-C
333	Giovanni Stracquadanio	Next-generation, engineered enzyme replacement therapies for Fabry disease	20-D
334	Emily Stuebing	Cardiac MRI findings among Fabry disease patients undergoing higher intensity therapy	19-C
339	Sophie Thomas	Transition in lysosomal diseases: Results of a UK patient and carer survey	19-D
340	Lindsay Torrice	Exploring the optimal timing of treatment for attenuated MPS II: Learning from a late-diagnosed symptomatic adult vs asymptomatic infant identified by newborn screening	18-C
341	Karmen Trzupek	Outcomes of an expert working group in Pompe disease: Recommended patient reported outcome measures to support an online research platform	18-D
343	Aalap Verma	Quantitative Systems Pharmacology (QSP) modeling based head-to-head comparison of agalsidase-beta and migalastat therapy in a virtual population of Fabry disease patients	17-C
348	Andrew Wang	Clinical, neurological, and neuroradiological outcomes of intravenous AAV9 gene therapy in infantile GM1 gangliosidosis patients	17-D
356	Anna-Maria Wiesinger	Irritable bowel syndrome manifestation in pediatric Fabry disease patients	32-A
357	Anna-Maria Wiesinger	A suspicion index tool (SIT) to aid diagnosis and treatment of ASMD	32-B
358	Tobias Willer	Stable: A protein engineering platform for enhancing stability and delivery of alpha-galactosidase A for Fabry disease therapy	31-A
LB-06	Karen Bean	Treatment effect of atidarsagene autotemcel (arsa-cel) in age-matched treated vs. untreated sibling pairs with early-onset metachromatic leukodystrophy (MLD)	31-B
LB-12	Merve Emecen Sanli	Gene replacement therapy for MPS IIIC with AAV9/HGSNAT vector	30-A
LB-16	Carolina Fischinger Moura De Souza	Two year update from the first-in-human intracisternal dosing of TTX-181 investigational AAV9 gene therapy in a child with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2)	30-B
LB-17	Brad Garrison	The Accelerating Medicines Partnerships® (AMP®) Bespoke Gene Therapy Consortium (BGTC) regulatory playbook	29-A
LB-35	Miriam Nickel	Assessment of cognitive function in CLN3 patients: Application of the Vineland Adaptive Behavior Scale	29-B
LB-36	Miriam Nickel	Inter-rater reliability of the Hamburg iCRS scale: Quantitative scoring of disease progression in a cohort of infantile CLN1 patients	28-A
LB-37	Albina Nowak	Resilience in classic and later-onset patients with Fabry disease	28-B
LB-38	Christina Ohnsman	Updated interim results from the first-in-human clinical trial of TTX-381, an investigational gene therapy for the treatment of ocular manifestations of CLN2 Batten disease	27-A
LB-39	Paul Orchard	Safety and initial activity of autologous human B cells genetically engineered to express human iduronidase using the Sleeping Beauty transposon system: Results from a first-in-human clinical trial in subjects with MPS I	27-B

LB-40	Yan Ouyang	Interim results from a phase 2 trial of the GCS inhibitor AL01211 in treatment naïve, classic male Fabry disease patients	26-A
LB-41	Cho Rong Park	A novel long-acting alpha-galactosidase A analog, HM15421, mitigates progressive renal impairment in Fabry disease via enhanced renal targeting and prolonged cellular retention	26-B
LB-42	Nishitha Pillai	Exploring the relationship between clinical phenotype and biochemical parameters in MPS I: Insights from the MPS I Registry	25-A
LB-43	Nishitha Pillai	Differentiation of juvenile-onset Pompe disease among children found by newborn screening: Insights from Minnesota Pompe Consortium	25-B
LB-44	Guillem Pintos Morell	Individual and health-care system socioeconomic impact of lysosomal disorders in Spain	25-C
LB-45	Gabriela Ponte de Mattos	A novel homozygous variant in MAN2B1 causing alpha-mannosidosis	25-D
LB-46	Carlos Prada	Eliglustat use in pediatric Gaucher disease: An 8-year experience	26-C
LB-47	Carly Rasmussen	Outcomes of newborn screening for Gaucher disease: Insights from a single-center experience	26-D
LB-48	Lara Ruhberg	Long-term and large scale analysis of NfL as biomarker in CLN2 patients treated with cerliponase alfa: Strengths and limitations	27-C
LB-49	Volha Skrahina	Genetic stratification for Parkinson disease subjects for future personalized trails and therapies - Sidransky syndrome a new entity	27-D
LB-50	Ying Sun	New mouse models to study GBA1 mutation-associated diseases with multiple organ involvement	28-C
LB-51	Nahid Tayebi	Neurotinib, a brain penetrant c-Abl inhibitor, prevents activation of c-Abl kinase, TFEB, and α-synuclein	28-D
LB-52	Rithika Thampy	Beyond detection: Comparing state-based NBS methods for effective MPS I diagnosis	29-C
LB-53	Sophie Thomas	Evolving mental health needs in lysosomal disease communities. Findings from the UK LSD collaborative surveys	29-D
LB-54	Assel Tulebayeva	Clinical case of mucopolysaccharidosis type VI patient with airway crisis	30-C
LB-55	Ali Tunç Tuncel	Ambroxol - a game changer?	30-D
LB-56	Ali Tunç Tuncel	Changing therapeutic landscapes: The first case of HSCT for galactosialidosis	31-C
LB-57	Christine Waggoner	The GM1 Natural History Data Sharing Collaborative: A roadmap for accelerating rare disease research	31-D
LB-58	Lena Marie Westermann	Real-world presymptomatic treatment in CLN2 disease: Learnings from families with multiple affected children	32-C
LB-59	Lena Marie Westermann	Dysregulation of peripheral blood monocytes in CLN2 disease	32-D
LB-60	Lena Marie Westermann	Longitudinal analysis of anti-drug antibody response against cerliponase alfa in CLN2 patients	33-A
LB-61	Knut Wittkowski	Oral alpha-cyclodextrins (αCD) as novel substrate reduction (co-) treatments (SRTs) against a hallmark of lysosomal (LY) diseases (LDs)	33-B

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3	Laura Adang	Characterizing diagnostic delays in metachromatic leukodystrophy: A real-world data approach	1-B
20	Daniel Bailey	Feasibility, acceptability, and safety of a physical activity and sedentary behaviour intervention (ACTIVE-FAB) for adults with Fabry disease	2-A
26	Michal Becker-Cohen	Prodromal Parkinsonian features in carriers of variants in GBA1: Who is at risk for developing Sidransky syndrome?	2-B
27	Michal Becker-Cohen	Will rapid administration of enzyme replacement therapy impact patients' satisfaction?	3-A
29	John Bernat	Extending the interval between pegunigalsidase alfa infusions in patients with Fabry disease: Five-year interim results from the ongoing BRIGHT51 study	3-B
31	Elizabeth Berry-Kravis	Arimoclomol for the treatment of Niemann-Pick disease type C in a real-world setting: Long-term data from an expanded access program in the United States	4-A
32	Elizabeth Berry-Kravis	Qualitative assessment of the validity and standardization of the swallow domain in the 5-Domain Niemann-Pick Disease type C (NPC) Clinical Severity Scale (5DNPCCSS) and analysis in an NPC clinical trial data set	4-B
33	Pablo Bianculli	Prospective observational study to assess the long-term safety of olipudase alfa effect in pediatric patients less than 2 years of age with acid sphingomyelinase deficiency: Study design	5-A
37	Tatiana Bremova-Ertl	Long-term findings of N-acetyl-L-leucine for Niemann-Pick disease type C	5-B
42	Alessandro Burlina	Reduced incidence of stroke in patients with Fabry disease treated with agalsidase beta: A matched analysis from the Fabry Registry	6-A
44	Barbara Burton	Unmet needs in the treatment and care of somatic manifestations in people with mucopolysaccharidosis type II (Hunter syndrome): A targeted literature review	6-B
45	Barbara Burton	Mucopolysaccharidosis type II sibling pairs study: A global retrospective chart review of effectiveness of idursulfase in infants and very young children with MPS II	7-A
46	Barry Byrne	Cipaglucosidase alfa plus miglustat in late-onset Pompe disease: two non-ambulatory patients switching from high-dose, high-frequency alglucosidase alfa	7-B
47	Melissa Calton	Non-clinical evaluation of 4D-310 in combination with rituximab/sirolimus: A translational study to support adoption of a novel prophylactic immunomodulation regimen in clinical trials in adults with Fabry disease	8-A
54	Chloe Cheung	Correlation of plasma LGL1 levels with clinical phenotype and treatment decisions in patients with Gaucher disease: Single site experience	8-B
55	Claudia Church Smith	Effective coordination of family screening to facilitate early diagnosis of Fabry disease	8-C
58	Kristin Clinard	Fatal intracranial hemorrhage in acid sphingomyelinase deficiency: A case report highlighting the role of managing comorbidities	8-D
61	John Collyer	Neurodevelopmental benefits of early hematopoietic stem cell transplantation in mucopolysaccharidosis type II	7-C
62	Sandra Cowie	Exploring global access challenges for Niemann-Pick disease therapies: Insights from an INDPA survey	7-D
63	Marco Curiati	Severe leukopenia and thrombocytopenia in an acid sphingomyelinase deficiency patient with dengue virus infection - case report	6-C
65	Christine I Dali	Safety of arimoclomol in a pediatric sub-study of Niemann-Pick disease type C patients aged 6 to 24 months at study enrolment	6-D
70	Stephanie DeArmey	Outcomes of a pediatric patient with late-onset Pompe disease switching from high-dose, high-frequency alglucosidase alfa to standard-dose cipaglucosidase alfa plus miglustat	5-C

73	Javier Adolfo de las Heras Montero	Successful desensitization protocol to alglucosidase and avalglucosidase alfa in a patient with infantile-onset Pompe disease	5-D
76	Tama Dinur	Mild Gaucher disease genotype is associated with more pronounced progress of pathologic midbrain sonography in the Sidransky syndrome	4-C
80	Lucy Dougherty De Miguel	Long-term outcome of mucopolysaccharidosis type I (Hurler syndrome) patients after hematopoietic cell transplantation	4-D
82	Consuelo Durand	Late diagnosis of Pompe disease in a patient with Down syndrome	3-C
85	Robin Ely	Exploratory survey study on adjunctive use of medical cannabis for Gaucher disease	3-D
86	Emily Eshraghian	Long term enzyme replacement therapy after hematopoietic stem cell transplant results in immune tolerance and improved biochemical outcomes	2-C
87	Martha Faraguna	Epilepsy is part of the central nervous system phenotype in classic infantile Pompe disease	2-D
90	Ulla Feldt-Rasmussen	Treatment satisfaction in patients with Fabry disease: Patient-reported outcomes from the followME Fabry Pathfinders registry and the SATIS-Fab study	1-C
91	Ulla Feldt-Rasmussen	FollowME Fabry Pathfinders Registry: Patient-reported outcomes in a cohort of patients on migalastat treatment for median 4 years	1-D
93	Giulia Ferrera	Neurological outcome following unrelated donor hematopoietic stem cell transplantation in children with juvenile globoid cell leukodystrophy: A long term study	16-A
94	Can Ficicioglu	Arimoclomol safety profile in the treatment of Niemann-Pick disease type C in a real-world setting: Long-term safety data from an expanded access program in the United States	16-B
99	Francesca Fumagalli	Atidarsagene autotemcel (autologous hematopoietic stem cell gene therapy) preserves cognition, language, and speech and slows brain demyelination and atrophy in early-onset metachromatic leukodystrophy	15-A
102	Jamie Gault	Patient perspective of participation in clinical studies in LOPD: Structured exit interviews from cipaglucosidase alfa plus miglustat studies	15-B
103	Michael Gelb	Massively parallel biochemical annotation of VOUS for lysosomal disorders	14-A
105	Dominique Germain	Erysipelas in patients with classic Fabry disease: A case series	14-B
107	Pilar Giraldo	Efficacy of eliglustat administered with and without imiglucerase in pediatric participants with Gaucher disease type 1 or type 3: The ELIKIDS study	13-A
108	Roberto Giugliani	Gangliosidoses: Understanding disease evolution in GM1 and GM2	13-B
109	Laura López de Frutos	Application of a natural history study on an investigational drug clinical development	12-A
110	Roberto Giugliani	Rainbow study: Phase 2 study of nizubaglustat as an investigational treatment for Niemann-Pick disease type C and GM2 gangliosidosis	12-B
111	Gulden Gokcay	Safety and pharmacokinetics of eliglustat administered with and without imiglucerase in pediatric participants with Gaucher disease type 1 and type 3: the ELIKIDS study	11-A
112	Jennifer Goldstein	Pseudodeficiency: A poorly defined and misunderstood term in an era of precision medicine	11-B
118	Christina Grant	Characterization of patients with mucopolysaccharidosis type VII (MPS VII) in the disease monitoring program (DMP)	10-A
119	Miles Greenberg	Heparan sulfate reduction in cerebrospinal fluid is associated with long-term cognitive outcomes in Hurler syndrome	10-B
121	Nathalie Guffon	Olipudase alfa in adult and pediatric ASMD patients: Interim results from French early access program	9-A
122	Nathalie Guffon	Long-term motor function and quality of life outcomes in patients with alpha-mannosidosis: Data from two velmanase alfa extension studies over 10 years	9-B
124	Punita Gupta	Infantile Tay-Sachs disease: Why do we continue to miss this diagnosis prenatally?	9-C
126	Karolina Stepien	The cognitive and mental health support of adults with lysosomal diseases: A review of the neuropsychology service in one tertiary metabolic centre	9-D

129	Caroline Hastings	Trappsol® Cyclo™ (hpβCd) for the long-term treatment of Niemann Pick type C1: Efficacy and safety data from 4 clinical studies and the ongoing expanded access program	10-C
130	Caroline Hastings	Trappsol®Cyclo™ and NPC: Efficacy shown across individual 5D domains and utilization of future assessment tools to demonstrate clinically relevant outcomes	10-D
132	Candice Herber	Age-dependent reference intervals for cerebrospinal fluid (CSF) and urine heparan sulfate (HS) and dermatan sulfate (DS) and CSF gangliosides	11-C
133	Juan Hernandez	Design of a gold nanoparticle based delivery system functionalized with recombinant hexosaminidase for Tay-Sachs disease treatment	11-D
135	Louise Himmelstrup	Perseverance is key for regulatory success in ultra-rare diseases - key learnings from arimoclomol's regulatory journey	12-C
136	Jennifer Hiros	Trial in progress: An open-label study (AT1001-025 RENEW) to evaluate the safety and pharmacokinetics of migalastat in patients with Fabry disease and amenable GLA variants and severe renal impairment or end-stage renal disease treated with dialysis	12-D
137	Niels Høeg Brandt-Jacobsen	Changes in patient demographics with the introduction of Fabry specific therapy in 2001: Data from the National Danish Fabry Cohort	13-C
138	Myrl Holida	Improved tolerability following enzyme replacement therapy switch to pegunigalsidase alfa: A case series from two centers of the expanded access program	13-D
140	Krista Casazza	A multi-disciplinary collaboration to investigate the potential contexts of using sleep-based DHTs into clinical research and practice	14-C
141	Robert Hopkin	Evaluating the relationship between infusion-related reactions and anti-drug antibody status: Results from 111 patients with Fabry disease treated with pegunigalsidase alfa	14-D
142	Robert Hopkin	Quality of life of migalastat-treated adolescents with Fabry disease: Results from the ASPIRE study and open-label extension	15-C
146	Derralynn Hughes	Isaralgagene civaparvovec (ST-920) gene therapy in adults with Fabry disease: Updated results from an ongoing phase 1/2 study (STAAR)	15-D
147	Derralynn Hughes	Clinical assessment of disease severity in patients with Fabry disease treated with pegunigalsidase alfa: An integrated analysis	16-C
149	Majdolen Joleen Istaiti	Three decades of cancer data in patients with Gaucher disease: Insights from the world's largest referral center	16-D
150	Majdolen Joleen Istaiti	Safety and efficacy study of taliglucerase alfa in Gaucher disease type 3: A 12-month multi-center trial	17-A
151	Majdolen Joleen Istaiti	High-dose ambroxol for Sidransky syndrome: The AGPI clinical trial	17-B
155	Monika Izdebski	Social determinants of health in the mucopolysaccharidoses	18-A
161	Karolina Stepien	The port-a-cath use in adult patients with lysosomal diseases: Indications, contraindications, and new challenges - a review of the clinical practice in one tertiary metabolic centre	18-B
163	Diana Jussila	The impact of a community: Engaging with the FDA and other stakeholders	19-A
164	Diana Jussila	Patient experiences in a clinical study for late-onset GM2: Insights before, during, and after a trial	19-B
165	Ilkka Kantola	Enzyme replacement therapy with renin-angiotensin system inhibition prevents kidney function decrease in most Finnish Fabry disease patients treated for 5 or 10 years	20-A
168	Thomas Kenny	Speaking the same language: The Fabry lexicon and the implications for how the healthcare community understands the impact of ERT	20-B
169	Thomas Kenny	Awareness of Fabry disease among non-Fabry specialists: Opportunities for education	21-A
170	Thomas Kenny	Maximising engagement through feedback: Insights from shared decision-making toolkit for Fabry disease patients	21-B
171	Lea Keufen	Clinical benefit on the skeletal system in a MPS IH patient after hematopoietic stem cell transplantation and long-term enzyme replacement therapy	22-A
172	Aneal Khan	Lentiviral gene therapy for Fabry disease - 5 year end of study analysis for the FACTS trial	22-B

173	Aneal Khan	Outcome of patients with Fabry disease: An independent analysis from a single clinic in Calgary	23-A
175	Priya Kishnani	Updated analyses of patients with Pompe disease identified through newborn screening in the United States: Data from the Pompe Registry	23-B
176	Masahisa Kobayashi	Clinical evaluation of migalastat in the correlation between plasma Lyso−Gb3 level and enzyme activity increase on in vitro assay in Fabry disease patients	24-A
177	Gerasimos Konidaris	Development of a novel patient-reported outcome (PRO) measure for acid sphingomyelinase deficiency according to the United States Food and Drug Administration (USFDA) guidance	24-B
185	Didier Lacombe	An observational cohort study evaluating outcomes in patients with Fabry disease treated with migalastat in a real-world setting in France (MIGA-Fab)	24-C
186	Florian Lagler	Building safe systems for patients with mucopolysaccharidoses (MPS) - results of an expert survey on patient safety	24-D
187	Florian Lagler	Simulation-facilitated training for parents of patients with mucopolysaccharidosis increases parental self-efficacy and reduces anxiety and stress: Results from a mixed-method psychological study	23-C
188	Daniël Lambregts	A systematic review on the clinical experiences of lentiviral gene therapy in treating lysosomal diseases	23-D
189	Christina Lampe	First real world experience: 4 adult patients with Fabry disease treated with 2mg/kg pegunigalsidase alfa every 4 weeks	22-C
191	Dawn Laney	Development and validation of an automated predictive scoring system to identify patients at increased risk for Fabry disease using Japanese electronic cardiac failure data	22-D
192	Dawn Laney	Improved growth in children with Fabry disease during treatment with agalsidase beta: A Fabry Registry analysis	21-C
193	Heather Lau	Treatment with UX111 gene therapy rapidly reduced heparan sulfate (HS) exposure in cerebrospinal fluid (CSF) and improved long-term cognitive function in children with mucopolysaccharidosis type IIIA (MPS IIIA)	21-D
199	Kaye LeMoine	Experience with shortened agalsidase-beta infusion in Fabry disease	20-C
205	Connor Lewis	Diffusion tensor imaging and correlational tractography findings in late-onset GM2 gangliosidosis differentiate Sandhoff and Tay-Sachs subtypes	20-D
206	Connor Lewis	Volumetric magnetic resonance imaging and diffusion tensor imaging metrics correlate with clinical outcomes following gene therapy in GM1 gangliosidosis patients	19-C
209	Juan Llerena	Analysis of juvenile vs. adult-onset Pompe disease: Insights from a multinational registry	19-D
213	Nicola Longo	Lower rate of infusion-related reactions in patients with Fabry disease after switching from agalsidase beta to pegunigalsidase alfa	18-C
216	Nicole Lyn	The lengthy and burdensome journey to diagnosis for female patients with Fabry disease	18-D
217	Tippi MacKenzie	Interim results from a first in human phase 1 clinical trial of in utero enzyme replacement therapy for lysosomal disorders	17-C
219	Benedicta Marshall-Andrew	Barriers and facilitators to clinical trial participation: Improving accessibility, logistics, and awareness	17-D
221	Ana Maria Martins	Patient-centered clinical outcomes - EQ-5D-3L and net promoter score in lysosomal diseases: Data from a participating center in the National Rare Diseases Network (RARAS)	32-A
222	Ana Maria Martins	Long-term impact of pabinafusp alfa on disease burden in Hunter syndrome: A 4-year follow-up of patient-reported outcomes	32-B
223	Ana Maria Martins	The effect of interruption of enzyme replacement therapy for acid sphingomyelinase deficiency - a case report of a single center in Brazil	31-A
224	Martí Mascaró Pol	Heart transplant in a male diagnosed with Gaucher disease type 3c	31-B
225	Paul McIntosh	POM-005: A global, prospective, observational registry of people living with Pompe disease	30-A
228	Eugen Mengel	Efficacy results from a 12-month double-blind randomised trial of arimoclomol for treatment of Niemann-Pick disease type C- presenting are scored 4-domain NPC clinical severity scale	30-B

229	Eugen Mengel	Longer-term efficacy and safety evaluation of arimoclomol treatment in patients with Niemann-Pick disease type C- data from 48 months open label trial	29-A
230	Eugen Mengel	Survival and causes of death in patients with Gaucher disease type 3: A multinational cross-sectional study	29-B
231	Pramod Mistry	Long-term outcomes of imiglucerase treatment in children with Gaucher disease type 1 or type 3 starting therapy before the age of 2 years	28-A
232	John Mitchell	Long-term outcomes of MPS IVA patients treated with elosulfase alfa: Findings from the Morquio A Registry Study (MARS) after 10 years	28-B
233	Mark Moran	Dysarthria-related intelligibility scores in a cohort of late-onset GM2 gangliosidosis patients	27-A
234	Elizabeth Morris	National peer-review of the transition process in lysosomal disease centers in England	27-B
235	Simon Moser	Anesthesia in mucopolysaccharidoses - retrospective analysis and insights of a large cohort	26-A
236	Tahseen Mozaffar	AT845 gene replacement therapy for late-onset Pompe disease: An update on safety and preliminary efficacy data from FORTIS, a phase 1/2 open-label clinical study	26-B
238	Joseph Muenzer	Skeletal findings in patients with attenuated MPS I receiving laronidase enzyme replacement therapy: Descriptive data from the MPS I registry	25-A
239	Lindsey Murray	Using clinical outcome assessments for rare disease clinical trial efficacy endpoints: Regulatory considerations and case studies	25-B
240	Nicole Muschol	Clinical profiles of 134 patients with alpha-mannosidosis from the velmanase alfa clinical program and SPARKLE registry	25-C
241	Shiny Nair	Molecular cell atlas of the brain in neuronopathic Gaucher disease	25-D
243	Eunwoo Nam	Clinical significance of complex intronic haplotype in Fabry disease screening cohort: A prospective cohort study in Korean patients	26-C
248	Sofia Nordin	Pompe disease in Sweden: A real-world evidence study investigating disease burden, treatment patterns for enzyme replacement therapy and concomitant medications	26-D
250	Antonio Ochoa-Ferraro	Differences in initiation of enzyme replacement therapy across the United Kingdom	27-C
252	Andrew Oldham	Single centre experience of starting/ switching to one of two new enzyme replacement therapies in LOPD	27-D
253	Karolina Stepien	Hydrotherapy as a therapeutic modality for treating pain and improving mobility in adults with mucopolysaccharidosis type II	28-C
258	Karyn O'Neil	Safety, tolerability, and activity of ABX1100, a CD71 Centyrin siRNA conjugate targeting GYS1 in a phase 1 study in normal healthy volunteers	28-D
268	Tyler Picariello	The FORCE™ platform delivers acid alpha-glucosidase to muscle as well as central nervous system and resolves pathology in Pompe disease mice	29-C
270	Nishitha Pillai	Retrospective cohort study evaluating the disease burden of patients with Pompe disease treated with enzyme replacement therapy in the United States	29-D
272	Guillem Pintos Morell	Individual and health-care system socioeconomic impact of lysosomal disorders in Spain	30-C
273	Frances Platt	Insights into the mechanism of action of a acetyl-leucine as a therapeutic for lysosomal diseases	30-D
275	Melissa Pritchard	Communication and eating & drinking skills in five children with CLN2 at the time of diagnosis	31-C
276	Melissa Pritchard	Speech and language therapy assessment of communication and eating & drinking skills in children with CLN2 presented alongside Hamburg LINCL Scale Language Scores & Weill-Cornell LINCL feeding scores	31-D
279	Allegra Quadri	Retrospective chart review of Gaucher disease type 1 patients homozygous for the p.N409S variant	32-C
283	Uma Ramaswami	Long-term safety and efficacy of migalastat in adolescent patients with Fabry disease: Results from the ASPIRE study and open-label extension	32-D
287	Shoshana Revel- Vilk	MyGauch™ mobile technology platform to enhance Gaucher disease patients' care	33-A

288	Mark Roberts	Post-baseline outcomes of the UK Early Access to Medicines Scheme registry for cipaglucosidase alfa plus miglustat in late-onset Pompe disease	33-B
297	Martin Magner	Outcome of hematopoietic stem cell transplantation in 19 patients with alpha-mannosidosis	34-A
298	Martin Magner	Insights from the Czech MPS IVA patient cohort, effect of therapy	34-B
300	Maurizio Scarpa	Validation of a machine learned diagnostic algorithm for improving early diagnosis of acid sphingomyelinase deficiency using US health records	35-A
301	Maurizio Scarpa	Children with chronic acid sphingomyelinase deficiency treated with olipudase alfa for 4+ years show improvements or normalization in multiple disease manifestations: Final results of the ASCEND-Peds trial	35-B
302	Fabiana Schneider	Against all odds: Very early enzyme replacement therapy on a severe form of Wolman disease	36-A
303	Benedikt Schoser	Clinically important improvements in 6-minute walk distance (6MWD) and forced vital capacity (FVC) in adults with late-onset Pompe disease (LOPD) switching from alglucosidase alfa (alg) to cipaglucosidase alfa plus miglustat (cipa+mig) in the PROPEL study	36-B
305	Angela Schulz	Seizures and movement disorders in patients with CLN2 disease treated with cerliponase alfa in the real-world setting	37-A
306	Angela Schulz	Prospective longitudinal analysis of cardiac function in patients with CLN2 disease under enzyme replacement therapy (ICV-ERT) with cerliponase alfa	37-B
309	Brooke Sebastian	Predictors of functional gait from initial assessment in a cohort of attenuated Morquio syndrome patients at a specialist tertiary centre	38-A
310	Jiwon Seo	Assessment of enzyme replacement therapy effect on cardiac imaging and clinical outcomes in Fabry disease cardiomyopathy	38-B
318	Reena Sharma	Results from GALILEO1, a first in human clinical trial of FLT201 AAV-gene therapy in adult patients with Gaucher disease type 1	39-A
322	Sofia Shrestha	Early clinical experience in initiating pegunigalsidase alfa in Fabry disease: Trends in safety, efficacy, and tolerability	39-B
327	Barbara Soberon	Evaluation of the correlation coefficient to determine the severity of bone marrow infiltration between bone marrow burden, fat fraction and Lyso GL1 in adult patients with Gaucher disease. A Study from the Argentine group for the diagnosis and treatment of GD	40-A
328	Barbara Soberon	Home infusion experience with enzyme replacement therapy for lysosomal diseases	40-B
330	Orna Staretz Chacham	Trappsol® Cyclo™: open label treatment in the transportnpc™ sub-study in patients under the age of 3 diagnosed with Niemann -Pick disease type c1	41-A
331	Robert Steiner	Retrospective cohort study evaluating the economic burden of patients with Pompe disease treated with enzyme replacement therapy in the United States	41-B
332	Karolina Stepien	Transition of care from pediatric to adult services for patients with mucopolysaccharidosis type II: An international observational study	41-C
338	Mark Thomas	Phase 1/2 clinical trial evaluating 4D-310 in adults with Fabry disease cardiomyopathy: Interim analysis of cardiac and safety outcomes in patients with 21-42 months of follow up	41-D
344	Suresh Vijay	Survival achieved in infants with rapidly progressive LAL-D via sebelipase alfa ERT: Results from the International LAL-D Registry	40-C
345	Jesus Villarrubia	Design of PROMs and PREMs questionnaire for ASMD care	40-D
346	Daniel Virga	Development of a durable gene therapy for targeting CNS and visceral pathologies in acid sphingomyelinase deficiency	39-C
347	Eric Wallace	Indirect treatment comparisons of pegunigalsidase alfa vs other therapies for left ventricular mass index in Fabry disease	39-D
349	Raymond Wang	Cardiovascular structure and function in MPS VII subjects	38-C
351	Slawomir Wantuch	Correction of glycogen accumulation in muscle, heart and CNS in a pre-clinical model of hematopoietic stem cell gene therapy for Pompe disease	38-D

352	Melissa Wasserstein	Final results of the ASCEND trial: Continued or sustained improvements in hepatosplenomegaly, respiratory outcomes, and lipid profile after 4 years of olipudase alfa enzyme replacement therapy in adults with acid sphingomyelinase deficiency	37-C
355	Anna-Maria Wiesinger	An evidence-based individual treatment trial with adalimumab in a patient with Morquio syndrome	37-D
362	Hiroyuki Yamakawa	Standardization of home enzyme replacement therapy for lysosomal disease in Japan	36-C
363	Chia-Feng Yang	Long-term outcomes of very early treated infantile-onset Pompe disease with short-term steroid premedication and the improvement of switching to avalglucosidase alfa: Real-world experiences from a nationwide newborn screening programme	36-D
364	Jaco Botha	Bone mineral density improvements in velaglucerase alfa-treated patients with Gaucher disease: Real-world data from the Gaucher Outcome Survey (GOS)	35-C