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 will be presented on **Tuesday, February 3** from **15:30-17:30 PST**

Translational Research & Additional Late-Breaking Science Posters will be presented on **Wednesday, February 4** from **15:30-17:30 PST**

Clinical Applications Posters will be presented on **Thursday, February 5** from **15:30-17:30 PST**

*** **Contemporary Forum & Rapid-Fire 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 3th and Wednesday, February 4th.

ePosters will be available to all registered attendees via the WORLD*Symposium* mobile app beginning at 05:00 PST on Tuesday, February 3, 2025, and will remain accessible throughout the live meeting. On Demand registered attendees can access ePosters from February 11 to March 13, 2026.

Any poster numbers not listed will not be presented as the author is unable to attend the conference.

It is the policy of WORLD*Symposium* to publish all abstracts with the list of authors exactly as the abstract was submitted to WORLD*Symposium*. The first author of the submitted abstract will be listed as the presenting author on the Preliminary Program, Agenda, and Poster List.

Poster Session I: Basic Science — Tuesday, February 3, 15:30-17:30 PST

#	Author	Title	Kiosk
5	Noor Ul Ain	Gaucher disease therapy mitigates monoclonal gammopathy: Clinical outcomes and glucosylsphingosine trajectories	1-A
8	Katia Alileche	Investigating microglial heterogeneity in a mouse model of mucopolysaccharidosis type IIIB	1-B
17	Isidro Arevalo-Vargas	Validation of top-performing in silico predictors for VUS identified by NGS in lysosomal disorders	1-C
18	Imane Assiri	Urinary sphingomyelin measurement: A simple tool for diagnosis and monitoring in Niemann-Pick disease type A	1-D
19	Maria Athanasopoulos	Decoding NEU1 - a molecular switch linking sialidosis, hepatic receptor regulation, and atherosclerosis	2-A
21	Alireza Ayoubi	Urine podocyte (UPC) globotriaosylceramide volume [V(GL3/PC)] is a non-invasive biomarker of Fabry nephropathy and correlates with increased albuminuria and proteinuria and reduced GFR in patients with Fabry disease	2-B
39	Abdelaati Berrachid	Metabolomic profiling of rosa damascena mill. hydroethanolic extracts reveals complex chelating signatures relevant to copper overload disorders	2-C
44	Maya Bhattiprolu	The impact of a GlcCer transporter on GBA1-associated Parkinson disease pathology in a murine model	2-D
49	Elizabeth Braunlin	Creation and characterization of a large animal model of MPS IVA	3-A
52	Barbara Burton	Persistent clinical burden and unmet needs in Hunter syndrome (MPS II) in the United States: A retrospective cohort study	3-B
61	Giorgio Cazzaniga	Zebra bodies recognition by artificial intelligence: A computational tool for Fabry nephropathy	3-C
67	Yun-Ru Chen	Genetic modifiers of cardiac outcomes in Taiwanese Fabry disease patients carrying the GLA c.639+919G>A variant	3-D
69	Eric Choy	Modeling CLN6 Batten disease using rapidly differentiated IPSC neurons	4-A
70	Gianna Cimarrusti-Jewhurst	Characterization of ocular structural alterations in mucopolysaccharidoses mouse models by immunofluorescence	4-B
80	Julia Dao	Tubular biomarkers detect early renal decline in Fabry disease	4-C

81	Supta Das	Multi-omic analysis of iPSC-derived GBA1-associated Parkinson disease microglia	4-D
85	Jordi Diaz-Manera	Cartography of transcriptomic changes in muscle biopsies of patients with late-onset Pompe disease	5-A
89	Matthias Dierick	Engineering low immunogenic enzyme replacement therapies for Fabry disease	5-B
98	Jessica Doxey	The important role of CRIM analysis by western blot in the setting of novel GAA variants	5-C
99	Hatim Ebrahim	Lysosomal diseases highly specialised services: Five centers service evaluation and experiences of patients and clinicians in laboratory diagnosis in England, United Kingdom	5-D
101	Tuba Eminoglu	Is ultrasonography a reliable approach for the evaluation of carpal tunnel syndrome in patients with mucopolysaccharidosis?	6-A
102	Tuba Eminoglu	Quality of life and related factors in patients diagnosed with mucopolysaccharidosis and their caregivers	6-B
104	Takumi Era	Studying for pathogenetic mechanism and drug development of Tay-Sachs disease using patient-derived iPS cells	6-C
105	Hamid Esmaeili	GCase deficiency disrupts the protein LRRC15: Implications for neuroinflammation	6-D
106	Erin Falsey	Novel genotypes of MFSD8 presenting with adult onset macular dystrophy	7-A
107	Sadig Fatullaev	Analysis of relationships of serum markers of liver fibrosis with laboratory parameters in children with rare disease	7-B
109	Gorka Fernández-Eulate	Mechanisms of spinal motor neuron dysfunction in late-onset GM2 gangliosidosis	7-C
110	Vinicius Ferraz	Not all elevated Lyso-Gb1 is a Gaucher disease: Do not forget the SCARB2-related diseases	7-D
111	Vinicius Ferraz	Clinical, epidemiological, diagnostic and therapeutic profile of Fabry disease patients: A study based on the Brazilian national network for rare diseases	8-A
112	Vinicius Ferraz	First report of the c.869G>A (p.Arg290His) variant in homozygosity trisomy 21 patient: Dilemmas for follow up	8-B
113	Lucas Ferreira Teixeira	Should the HFE gene be sequenced in every Gaucher disease patient? An exploratory study	8-C
117	Naomi Fisher	Exploring electroencephalography markers of excitation-inhibition imbalance in type 3 Gaucher disease with myoclonic epilepsy	8-D
123	Jeffrey Gaynes	PS gene-editing system corrects Sandhoff disease with a brain-penetrant HEXO-APOE enzyme	9-A
130	Pilar Giraldo	Immunomodulatory and metabolic role of D vitamin in lysosomal diseases	9-B
139	Caio Gomes	Aberrant MAN2B1 transcripts revealed by long-range RT-PCR in a child with suspected alpha-mannosidosis	9-C
140	João Gonçalves	Comparative immunogenicity of agalsidase alfa and pegunigalsidase alfa in Fabry disease: Incidence, cross-reactivity, neutralization, and clinical correlates	9-D
144	Russell Gotschall	M021: Unlocking the future of Pompe ERT	10-A
145	Meghana Govindaraj	Transcriptomics study of Morquio syndrome type A mice aortas	10-B
149	Angela Gritti	Therapeutic benefits of hematopoietic stem cell gene therapy using optimized bicistronic lentiviral vectors in mouse models of GM2 gangliosidosis	10-C
150	Nathalie Guffon	KI-6 as a new biomarker of lung improvement in ASMD patients undergoing ERT: A single-center study	10-D
151	Nathalie Guffon	Clinical impact of the MAN2B1 c.2248C>T variant in patients with alpha-mannosidosis: Genotype-phenotype insights from the SPARKLE registry	11-A
156	Miloud Hammoud	Detection of Gb3 and lactosylceramide in non-Fabry urine samples: Insights into the biochemical landscape of lysosomal disorders	11-B
165	Wendy Heywood	Urinary glycerophosphoinositol is a potentially non-invasive clinically applicable biomarker for CLN3 disease	11-C

166	Stephan Hold	Next-generation biomarker analysis: A 6-minute LC-MS/MS assay for α-mannosidosis oligosaccharide monitoring	11-D
169	Xinying Hong	Multiplexed endogenous non-reducing end glycosaminoglycan biomarkers and oligosaccharides panel: A clinical lab's experience in diagnosis and treatment monitoring	12-A
171	Mahin Hossain	Glial cell dysfunction and neurodegeneration in a novel knock-in mouse model of lysosomal free sialic acid storage disorder	12-B
172	Ellen Howard	Caregiver treatment preferences in late infantile and early juvenile metachromatic leukodystrophy: A best-worst scaling study	12-C
173	Shih-Chang Hsueh	Novel cyclodextrins enhance potency of neutralization and clearance of endogenous psychosine in cellular and in vivo models of Krabbe disease	12-D
180	Margarita Ivanova	Cytokine dysregulation in pediatric Gaucher disease: Implications for bone and immunity	13-A
181	Keerthana Iyer	Comparative proteomic analysis of saliva, urine, and serum in mucopolysaccharidosis type I patients	13-B
182	Maroua Jakani	Rational diagnostic approach to Tay-Sachs disease highlighting urinary ganglioside excretion	13-C
185	Julia Jamieson	Assessment of the gastrointestinal manifestations of mucopolysaccharidosis type IIIB in the NAGLU knockout mouse model	13-D
197	Martyna Kasprzyk	Production and characterisation of a functional recombinant human palmitoyl-protein thioesterase 1 in K. phaffiifor the treatment of CLN1	14-A
208	Engin Köse	Clinical manifestations and molecular genetics of seven patients with Niemann-Pick disease type C: A case series with a novel variant	14-B
209	Engin Köse	Long-term clinical evaluation of patients with alpha-mannosidosis - a multicenter study	14-C
214	Didier Lacombe	Description of the French cohort of ASMD type AB patients	14-D
220	Chris Lee	Quantification profiles of enzyme activity, secretion, and psychosine levels of Krabbe disease galactosylceramidase missense variants	15-A
221	Malte Lenders	A rapid method to reduce drug interferences for antibody measurements in pegunigalsidase alfa-treated patients with Fabry disease	15-B
222	Veronica Lentini	Inflammation and lysosomal dysfunction in Fabry macrophages	15-C
223	Livia Lenzini	Extracellular vesicles as new mediators of pathology in Anderson Fabry disease	15-D
228	Tina Loeffler	Neonatal ICV delivery and longitudinal NfL monitoring as tools for early CNS intervention in preclinical studies	16-A
229	Tina Loeffler	Cross-indication profiling of neurodegenerative markers in lysosomal disease models	16-B
233	Anna Ludlaim	IPSC-derived neural cells: A platform for biomarker and therapeutic development across the Gaucher disease phenotypic spectrum	16-C
236	Leslie Lynch	Effect of vosoritide on bone biomarkers in prepubertal children with mucopolysaccharidosis types IVA and VI	16-D
239	Francois Maillot	The slowly progressive form of MPS VI: A French-Brazilian case-series	17-A
242	Haylen Marin Gomez	Beyond substrate storage: Genotype-specific inflammatory signatures in Fabry disease	17-B
243	Haylen Marin Gomez	From storage to signaling: Immune activation as the first hit in Fabry disease	17-C
246	Tomomi Masuda	A transferrin receptor-targeted α-L-fucosidase, JR-471, reduced core-fucosylated glycoasparagine in the brain and preserved motor function in a murine model of fucosidosis	17-D
255	Ali Mohajer	Age-stratified clinical burden of post-allogeneic HSCT complications in Hurler syndrome: A real-world data approach	18-A
256	David Moreno Martínez	Neuronal dysfunction beyond lysosomes in Fabry disease: Evidence from iPSC-derived forebrain neurons	18-B
260	Shiny Nair	Spatially resolved mechanisms of liver carcinogenesis in Gaucher disease implicate lipid-driven immunosuppressive niches	18-C

261	Samira Najeh	Hereditary tyrosinemia in Morocco: Clinical spectrum, diagnostic challenges, and genetic insights	18-D
262	Leyla Namazova-Baranova	Ventriculomegaly in mucopolysaccharidosis	19-A
264	Katherin Niño-Traslaviña	Evaluation of metabolomic profile in a NAGLU-deficient cellular model for MPS IIIB	19-B
269	Petra Oliva	Integrating α-mannosidosis into the differential diagnostic algorithm for suspected MPS: A 24-month update	19-C
270	Petra Oliva	Breaking barriers in lysosomal disorder screening: A novel simultaneous LC-MS/MS approach for Tay-Sachs, Sandhoff, and GM1 gangliosidosis diseases	19-D
283	Tyler Picariello	DYNE-401 demonstrates potential to address Pompe disease with low and infrequent dosing	20-A
285	Camila Pinheiro	Impairment of mitochondrial bioenergetics and alterations in the crosstalk between mitochondria and lysosomes in cellular models of mucopolysaccharidoses types I and II	20-B
292	Karthikeyan Rajagopal	In vitro evaluation of therapeutic beta-glucuronidase enzyme uptake and penetration in articular cartilage	20-C
293	Uma Ramaswami	Raised amylase - is it a feature of inflammation in Fabry disease?	20-D
295	Uma Ramaswami	Fabry disease mosaicism: Genotype-phenotype correlation and the role of next generation sequencing	21-A
297	Shoshana Revel-Vilk	Expanding the cutaneous phenotypic landscape of type 1 Gaucher disease	21-B
298	Shoshana Revel-Vilk	Risk and prevalence of overweight and obesity among adults with Gaucher disease	21-C
300	Sonia Roca-Esteve	Characterization of fibrosis and net related biomarkers in patients and carriers of lysosomal acid lipase deficiency	21-D
301	Lina Rodríguez	Characterization of recombinant hexosaminidase A produced in a glyco-engineered komagataella phaffii strain	22-A
307	Anastasia Rykunova	Characteristic of the microstructural morphometric brain parameters in children with different types of MPS: New findings and correlations	22-B
308	Anastasia Rykunova	Characteristics of macrostructural morphometric brain parameters in children with different types of MPS: New findings and correlations	22-C
312	Pau Sarlé Vallés	Next-generation enzyme replacement strategies for Gaucher disease	22-D
316	Christoph Schwering	Brainstem auditory evoked potentials (BAEP) in CLN2 patients under treatment with cerliponase alfa	23-A
317	Irene Serrano Gonzalo	Identifying predictive markers of lysosomal acid lipase deficiency in a retrospective patient cohort	23-B
321	Ibrar Siddique	Blood-based biomarkers for mucopolysaccharidosis type I and III	23-C
322	David Smerkous	A novel hig+F2:F376hly sensitive machine learning model for automated measurement of kidney peritubular capillary endothelial cell globotriaosylceramide accumulation	23-D
322	David Smerkous	A novel highly sensitive machine learning model for automated measurement of kidney peritubular capillary endothelial cell globotriaosylceramide accumulation	24-A
325	Fernanda Sperb Ludwig	The diagnostic odyssey of a biochemically confirmed case of ML II: The first western patient with LYSET associated disease	24-B
328	Richard Steet	MPS I missense variants that cause a gain of glycosylation effect are amenable to treatment with novel glycosylation inhibitors	24-C
333	Karolina Stepien	Evidence of secondary mitochondrial dysfunction in alpha-mannosidosis	24-D
340	Diego Suarez	Understanding the molecular and metabolic changes on a CRISPR-Cas9 generated HEXAKO astrocytoma model for the study of Tay-Sachs disease	25-A
341	Ying Sun	Brain delivery of long-acting enzymes via SapC-DOPS nanocarrier for neuronopathic Gaucher disease	25-B

342	Stephanie Tannous	Lipid-dependent uptake of pegunigalsidase alfa across cell types: Implications for multiorgan treatment	25-C
343	Mitra Tavakoli	Non-invasive assessments of sudomotor dysfunction and autonomic neuropathy in Fabry disease	25-D
348	Katarina Trebusak Podkrajsek	A non-coding signature in SHROOM3 is associated with kidney disease progression in Fabry disease	26-A
349	Giuseppe Uras	The lipidomic signature of Fabry disease: Beyond Gb3 accumulation	26-B
351	Alfredo Uribe-Ardila	Assessment of alpha-mannosidase and alpha-L-fucosidase in dried blood spots collected on filter paper. High-risk screening results in patients with Hurler-like phenotype in Colombia	26-C
352	Alfredo Uribe-Ardila	Leukocyte acid sphingomyelinase analysis in control population and patients with clinical suspicion of Niemann-Pick disease type A/B	26-D
353	Emilio Vaena	Characterization of bone and joint cell models of MPS II generated through CRISPR/Cas9 technology	27-A
355	Nato Vashakmadze	Interim results of a multicenter, open-label, multicohort study of the safety, pharmacokinetics, pharmacodynamics, and efficacy of verenafusp alpha in patients with mucopolysaccharidosis type II	27-B
378	Christopher Wingrove	Exploring the lived experience of the Fabry community in Czechia	27-C
385	Xiangli Zhao	A progranulin derivative blocks the C5a/C5aR1 signaling and mitigates pathology in Gaucher disease	27-D
386	Ewa Ziolkowska	Neuropathological alterations in TPP1-deficient cynomolgus macaques recapitulate key features of human CLN2 disease	28-A
LB-02	Hera Akmal	Volumetric MRI findings as differentiating biomarkers for juvenile-onset GM2 gangliosidoses: Tay-Sachs versus Sandhoff disease	28-B
LB-07	Maryam Banikazemi	AMT191 investigational gene therapy in adult males with classic Fabry disease; Initial safety and biomarker results of phase 1/2 study	28-C
LB-08	Morgan Barnes	Assessment of recombinant human palmitoyl-protein thioesterase 1 (PPT1) for ocular delivery	28-D
LB-10	Julia Becerra	Assessment of cardiac structure and function in mucopolysaccharidosis type IV patients utilizing echocardiography	29-A
LB-13	Elizabeth Berry-Kravis	Substantial survival benefit and slowing of disease progression with adrabetadex treatment in individuals with infantile-onset Niemann-Pick disease type C (NPC)	29-B
LB-14	Magnar Bjørås	Nizubaglustat reinstates pro-neuronal transcriptional programs in human CLN3 retinal organoids	29-C
LB-17	Charles Chmelik	Case report: Early infantile Pompe disease treated with avalglucosidase alfa	29-D
LB-18	Marie-Anne Colle	Deciphering the role of neuromuscular junctions in infantile Pompe disease pathogenesis	30-A
LB-19	Marie-Anne Colle	Intracellular membrane repair dysregulation and accumulation of mature myostatin protein are novel markers of muscle pathophysiology in Pompe disease	30-B
LB-21	Manuel Cueto	Migalastat therapy is associated with stabilization of quality of life in Fabry disease: A 12-month longitudinal analysis	30-C
LB-22	Jordi Diaz-Manera	Update on FORTIS: A phase 1/2 open-label clinical trial on AT845 gene replacement therapy for late-onset Pompe disease	30-D
LB-24	Jessica Gambardella	Entry of pegunigalsidase alfa in cardiomyocytes and impact on diastolic dysfunction in a mouse model of Fabry disease	31-A
LB-25	Jeannette Goh	Objective 3D surface mapping for longitudinal monitoring of dysmorphology in MPS VI: A feasibility study	31-B
LB-26	Russell Gotschall	M021 is more effective than all approved ERTs for glycogen clearance and reversal of cellular pathology in Pompe mice	31-C

LB-27	Nathan Grant	The needs of patients and families with mucopolysaccharidosis (MPS) during the transition from pediatric to adult health care	31-D
LB-28	Nathalie Guffon	Determinants of therapeutic adherence and disease acceptance in acid sphingomyelinase deficiency (ASMD): A mixed-methods study	32-A
LB-29	Kristen Hagarty-Waite	Plasma glial fibrillary acidic protein (GFAP) is a promising biomarker for central nervous system involvement in infantile-onset Pompe disease	32-B
LB-30	Christiane Hampe	Translational data from the first-in-human clinical trial of autologous human B cells engineered to express human iduronidase in subjects with MPS I: Support for pediatric studies	32-C
LB-31	Zackary (Ari) Herbst	A one-punch 10-plex method for lysosomal disorders, X-ALD, and CTX and an all-in-one quality control dried blood spot set for multiplexed enzyme activity and biomarker screening methods	32-D
LB-34	Niels Høeg Brandt-Jacobsen	Prescription patterns in Fabry disease prior to diagnosis; a retrospective, real world analysis from the national Fabry cohort in Denmark	33-A
LB-35	Dione Holder	Comprehensive characterization and translational implications of the *Galns*^{R384C} mouse model of mucopolysaccharidosis type IVA	33-B
LB-36	Robert Hopkin	Efficacy and safety of venglustat in patients with symptomatic Fabry disease: The PERIDOT study, a 12-month phase 3 randomized clinical trial	33-C
LB-37	Derralynn Hughes	Correlation between initial FIPI scores and MSSI evolution over time in Fabry disease: insights from a large retrospective cohort	33-D
LB-38	Ludmilla Kedenko	Clinical manifestation of the p.Arg356Pro *galactosidase alpha* variant of Fabry disease in an Austrian family	34-A
LB-40	Francyne Kubaski	Sulfatide analysis in cerebrospinal fluid of metachromatic leukodystrophy patients by liquid chromatography tandem mass spectrometry	34-B
LB-42	Emil Lenzing	Pulmonary involvement in Fabry disease: a retrospective, real-world analysis from the national Fabry cohort in Denmark	34-C
LB-46	Anika Menetrey	Genetic landscape in Niemann-Pick disease type C: insights from the INPDR registry	34-D
LB-47	Pramod Mistry	Safety and efficacy of venglustat versus imiglucerase in patients with Gaucher disease type 3 (LEAP2MONO): A phase 3, randomized, double-blind multicenter trial	35-A
LB-48	Marine Moutia	Organellar ion channels: Novel insights into lysosomal electrophysiology	35-B
LB-50	Simona Murko	Quantification of specific urinary oligosaccharide biomarkers for diagnosis and treatment monitoring of alpha-mannosidosis	35-C
LB-51	Krishan Sai Musini	Characterization of novel mucopolysaccharidosis type IVA GALNS hAAVS1 mouse model	35-D
LB-52	Dmitriy Niyazov	Coexistence of Niemann-Pick disease and Aicardi-Goutieres syndrome: Importance of comprehensive genetic testing in treatment and prognosis	36-A
LB-53	Peter Nordbeck	Isaralgagene civaparvovec (ST-920) shows stable cardiac function over one year in patients with Fabry disease: Results from the registrational phase 1/2 STAAR gene therapy study	36-B
LB-59	Marc Patterson	The Functional Independence Measure for Children (WeeFIM®) in Niemann-Pick disease type C	36-C
LB-60	Marc Patterson	Subgroup evaluation of adults and children with Niemann-Pick disease type C in the phase 3 (IB1001-301) extension phase (IB1001-301) assessing N-acetyl-L-leucine	36-D
LB-61	Karen Pignet Aiach	Five years follow-up update on AAVance, a phase 2/3 clinical trial of LYS-SAF302 gene therapy in children with Sanfilippo syndrome type A	37-A
LB-62	Kimia Rezaei	Validation of the University of California, Irvine corneal cystine crystal score (UCI CCCS): Correlations with clinical, imaging, and symptom outcomes in cystinosis	37-B
LB-63	Candela Romano	Large vessel strokes in patients with Fabry disease	37-C
LB-64	Yosef Scher	Comparison of the pre-ERT disease severity scoring system (DS3) and the Gaucher risk assessment for fracture (GRAF) score as predictors of future fracture risk	37-D

LB-69	Benjamin Solar	Shortened elosulfase alfa infusion: Lessons from a public health rare disease program in Chile	38-A
LB-70	Karolina Stepien	Psychosis and seizures as a long-term complication of hematopoietic stem cell transplantation in MPS I (Hurler syndrome): One centre experience	38-B
LB-71	Thanyachai Sura	The unparalleled benefits for health and the burden of life activities: a study of Fabry disease patients on ERT in different age groups in Thailand	38-C
LB-72	John Taggart	ASMD perspective index: Miss the angle, miss the diagnosis	38-D
LB-73	Sophie Thomas	Beyond the patient: Longitudinal insights into psychological wellbeing and caregiver burden in lysosomal diseases	39-A
LB-74	Cynthia Tifft	Adeno-associated virus type 9 gene therapy for GM1 gangliosidosis type 2: A phase 1/2 trial	39-B
LB-75	Assel Tulebayeva	Anesthetic management during dental treatment in patient with mucopolysaccharidosis type II	39-C
LB-77	Dana Velasquez Rivas	Stability assessment of lysosomal sphingolipid biomarkers (Lyso-Gb1, Lyso-Gb3, Lyso-SM) in dried blood spots over a nine-month storage period	39-D

Poster Session II: Translational Research

Wednesday, February 4, 15:30-17:30 PST

#	Author	Title	Kiosk
1	Sumiko Abe	Serial brain MRI volumetrics and DTI analysis in atypical CLN2 patients treated with ICV cerliponase alfa	1-A
2	Nicolas Abreu	Feasibility study of video-based quantitative gait analysis in late-onset Tay-Sachs disease	1-B
11	Jose Alvarez Gonzalez	Advances in the administration of ERT (oral pathway) improving the biodistribution of enzymes	1-C
12	Jose Alvarez Gonzalez	Advances in the early diagnosis of mucolipidosis type II	1-D
20	Christiane Auray-Blais	What about newborn urine screening possibilities using samples collected on filter paper by parents?	2-A
24	Rafael Badell-Grau	Treating mucopolysaccharidosis type IIIC with small molecule therapy that stabilizes lysosomal function and reduces inflammation	2-B
25	Rafael Badell-Grau	Hematopoietic stem cell gene therapy for mucopolysaccharidosis type IIIC	2-C
26	Tomas Baldwin	Development of an ultra-sensitive LC-MS/MS method for lyso-Gb1 and Gb1 quantitation in mouse CSF: Enabling pre-clinical and clinical applications for monitoring Gaucher and Parkinson disease	2-D
30	Miguel Barajas	Integrating genomics and phenotypes: Early insights into GLA and GAA variants in the All of Us Research Program	3-A
31	Kaitlin Batley	Implementation of newborn screening for Pompe disease and a pilot for pre-symptomatic monitoring	3-B
38	Donna Lee Bernstein	Rapidly progressive, infantile lysosomal acid lipase deficiency prevalence in the Mizrahi Jewish population	3-C
47	Shaun Bolton	A global unique identifier (GUID) tool for Niemann-Pick diseases	3-D
48	Natasha Bonhomme	Talk the talk: Using parent feedback and technology to improve result communication	4-A
50	Alice Brinckmann Oliveira Netto	Expanding Brazilian neonatal screening for lysosomal diseases: An evidence-based molecular genetics approach	4-B
53	Elizabeth Caller	A virtual psycho-education group for adults with Fabry disease: A pilot study	4-C
56	Krista Casazza	Characterizing neurological and psychiatric manifestations of Niemann-Pick disease type C: Differences across age and reporting perspectives	4-D
57	Krista Casazza	Difference in disease progression across the lifespan- findings from the NPC patient survey	5-A
58	Krista Casazza	Investigating the real-world experience of patients with Niemann-Pick disease type C (NPC) and their carers: Preliminary results	5-B
59	Laura Case	Early enzyme replacement therapy in late onset Pompe disease diagnosed by newborn screening	5-C
62	Betul Celik	Co-transplantation of hematopoietic stem cells and highly purified rapidly expanding clones (REC) of human mesenchymal stem cells rescued the bone pathology of MPS IVA mice	5-D
66	Hui-An Chen	Follow-up of Taiwanese males with high risk of classic Fabry disease via newborn screening	6-A
68	Alessandra Chiarot	Evaluating an exercise training approach to enhance muscle function and mitigate pathology in Pompe disease patients on enzyme replacement therapy	6-B
71	Sarah Clifford	Understanding symptoms and impacts in lysosomal diseases: A qualitative literature review and conceptual model	6-C
74	Matthieu Colpaert	GAA-based therapeutic strategies for neurological glycogen storage diseases	6-D
83	Ankit Desai	Improved outcomes in infantile-onset Pompe disease through early enzyme replacement therapy enabled by newborn screening: Insights from a multisite US cohort	7-A
97	Gabriela Dostalova	Rethinking Fabry disease genetic screening when exons are not enough	7-B

103	Tuba Eminoglu	Determination of cut-off values using the MOM approach in lysosomal diseases in Turkey: Findings from a newborn frequency study	7-C
108	Allan Feng	Therapeutic efficacy of a novel glucocerebrosidase variant in a new preclinical model of neuronopathic Gaucher disease	7-D
114	Grace Ferrell	Healthcare providers' attitudes toward newborn screening for Niemann-Pick disease type C	8-A
120	Jillian Gallagher	Testing a dual AAV gene therapy vector construct to treat sialidosis and galactosialidosis using small and large animal models	8-B
121	Courtney Garrett	Dual site administration of AAV gene therapy for treatment of feline GM1 gangliosidosis	8-C
122	Kathryn Gasperian	Reflecting on the past seven years: A single-center's experience managing newborn screen-identified patients with late-onset Pompe disease	8-D
124	Michael Gelb	Biochemical newborn screening for all treatable lysosomal diseases	9-A
129	Aidan Gill	Participation in clinical trials for neurological lysosomal disease: Patient and family perspectives	9-B
138	Jennifer Goldstein	The clinical genome resource's collaborative approaches towards understanding the genetics of lysosomal diseases	9-C
142	Shelly Goomber	Functional profiling-based evaluation of GAA VUS in Pompe disease using a transient expression system upgraded for capacity and robustness	9-D
147	Melissa Greco	Infantile Krabbe disease presenting with intermediate psychosine (2-10 nmol/L) values in dried bloodspots	10-A
148	Giuseppina Grillo	Fabry disease patients treated with egunigalsidase alfa: One year patient reported experience of starting a new therapy at a single UK centre	10-B
154	Leanne Hagen	Dungeons and drug approval	10-C
155	Patricia Hall	Performance of endogenous MPS biomarkers as second-tier tests for MPS I and MPS II newborn screening	10-D
157	Tae Un Han	Development of a novel systemic AAV gene therapy for neuronopathic Gaucher disease	11-A
161	Nadene Henderson	Sudden hearing loss and literature review within the Fabry disease population	11-B
164	Wendy Heywood	A transfusion related adverse event attributed to IgA2 driven complement activation is observed in two Fabry disease patients given α-galactosidase based enzyme replacement therapy	11-C
170	Justin Hopkin	Case report: Treatment of the severe ASMD phenotype with olipudase alfa and JZP-150	11-D
174	Alex Huang	Self-amplifying mRNA enhances transamniotic fetal mRNA delivery	12-A
177	Derralynn Hughes	A scalable approach to detecting undiagnosed Fabry disease from electronic health records	12-B
184	Elizabeth Jalazo	Impact of NBS for early identification and management of MPS I in siblings with W402X/ L238Q genotype	12-C
186	Xuntian Jiang	Diagnostic and therapeutic applications of the glycan biomarker H3N2b in GM1 gangliosidosis	12-D
194	Diana Jussila	The patient experience: Effecting change in ultra-rare disease trial design	13-A
195	Staci Kallish	Evaluation for A-gal syndrome leading to diagnoses of late-onset Fabry disease	13-B
198	Tory Kaye	When follow-up falls short: A case series on infants lost in newborn screening follow-up for mucopolysaccharidosis type I	13-C
201	Grace Kick	Efficacy of AAV-mediated gene therapy in a sheep model of CLN1 disease	13-D
203	Laura Kirkpatrick	Pediatrician involvement in communicating positive newborn screening results for Krabbe disease: Barriers, facilitators, and ideas for interventions	14-A
210	Francyne Kubaski	Dried blood spot arylsulfatase A enzyme analysis for second-tier newborn screening and diagnosis of metachromatic leukodystrophy	14-B

211	KeriAnn Kuperman	Rewriting the natural history of late onset Pompe disease: A case series comparing patients diagnosed before and after newborn screening	14-C
225	Daniel Lewi	Enhancing study participation through targeted patient empowerment	14-D
227	Mona Lindschau	Five-year follow-up after gene therapy with ABO-101 in five children with Sanfilippo syndrome type B	15-A
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234	Troy Lund	GAG endogenous non-reducing ends as a new biomarker in CSF and plasma for Hurler syndrome	15-C
240	Natasha Malgarezi	Detection of mucopolysaccharidosis in high-risk patients: Comparison among different screening strategies	15-D
249	Bryce Mendelsohn	Parental carrier screening is the first line approach to lysosomal disorders	16-A
252	Kathy Meyer	A combined fertility, embryofetal development, AAV integration and germline transmission risk study in mice with isaralgagene civaparvovec (ST-920) for Fabry disease	16-B
265	Dau-Ming Niu	Whole genome sequencing: Real-time analysis from birth to lifelong insights	16-C
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272	Cara O'Neill	Leveraging clinical metadata in Sanfilippo syndrome: A scoping review of study characteristics, data elements, outcome measures and methodologies	17-B
274	Saida Ortolano	Methods to detect cross-reactivity to pegunigalsidase of anti-alpha-galactosidase A IgG antibodies produced against other enzyme replacement therapy in Fabry disease patient serum	17-C
276	Marc Patterson	Glycoproteinoses: The International Society for Mannosidosis and Related Disorders (ISMRD): Forum on scientific progress, unmet patient needs, and the path to therapies	17-D
284	Françoise Piguet	Novel intravenous AAV gene therapy for mucopolysaccharidosis type IIIA and IIIB in mouse and canine model of the pathology - towards clinical translation	18-A
286	Hanna Polari	Screening performance of a new Revvity NeoLSD™ 7plex LC-MSMS kit	18-B
287	Hanna Polari	Multiplexed LC-MS/MS measurement of MLD, CTX, NP-C and X-ALD markers from a single dried blood spot sample	18-C
289	Maria Praggastis	Preclinical evaluation of a durable, tissue-targeted gene therapy for Pompe disease demonstrates superior efficacy over standard ERT	18-D
290	Rashi Priya	Enhanced correction of skeletal muscle and brain pathology in a Pompe mouse model using transferrin receptor-mediated delivery of GAA	19-A
291	Sarah Quinn	Eating and drinking outcomes for children with CLN2 receiving intraventricular enzyme replacement therapy	19-B
296	Salvatore Recupero	Gallbladder abnormalities in metachromatic leukodystrophy: Preliminary analysis in patients treated with atidarsagene autotemcel (autologous hematopoietic stem cell gene therapy) and untreated patients	19-C
304	Courtney Rouse	Procession to IND of a capsid mutated AAV8 codon optimized NAGLU vector for treatment of Sanfilippo syndrome type B	19-D
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309	Sampurna Saikia	The complementary strength of the AAV9 gene therapy in hematopoietic stem cells transplanted into MPS IVA mice	20-B
311	Ryunosuke Sanada	Development of gene editing technologies to correct a mutation in GNPTAB of mucolipidosis type II/III patients	20-C
319	Raghu Sharma	Hepcidin as a potential biomarker for monitoring inflammation in Gaucher disease	20-D

320	Surendra Sharma	Rapid transient antibody depletion using genetically engineered IgG-degrading enzyme allows efficient rAAV9 gene delivery in an α-AAV9-Ab⁺ rabbit model	21-A
338	Emily Stuebing	Impact of newborn screening on the clinical spectrum of neurodegenerative lysosomal disorders: 20 years of a single-center experience	21-B
344	Sophie Thomas	Co-producing UK clinical guidelines for lysosomal disorders: A patient professional partnership	21-C
345	Sophie Thompson	Facilitating early diagnosis of Fabry disease through cascade screening	21-D
346	Sophie Thompson	Enhancing Fabry cascade screening through direct contact of relatives	22-A
347	Hiroaki To	Treatment strategy for hematopoietic stem cell transplantation as a curative therapy for mucolipidosis type III and development of gene therapy as a novel treatment approach	22-B
354	Rachelen Varghese	Early manifestations, treatment outcome data, family impact, and emerging therapies necessitates newborn screening for Fabry disease	22-C
357	Daniel Virga	Anti-transferrin receptor 1-targeted AAV9 therapy prevents CNS and visceral pathologies in acid sphingomyelinase deficiency	22-D
358	Kristin Voorhees	Caregivers' perspectives of the MPS IIIA (Sanfilippo sydrome type A) diagnostic journey and ways to improve for future families	23-A
359	Sandra Vranic	Graphene flakes as versatile carriers for lysosomal enzymes: Activity, delivery kinetics, and bio-persistence in patient-derived fibroblasts	23-B
360	Christine Waggoner	Global characterization of GM1 gangliosidosis: Insights from a patient-powered registry	23-C
364	Udayanga Wanninayake	Enhanced CIMPR binding and cellular uptake of HP-GALNS (M161): A next-generation enzyme replacement therapy for Morquio syndrome type A	23-D
365	WM Subadra Wanninayake	Single centre experience on the disease modifying treatment for lysosomal disorder	24-A
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372	Chester Whitley	Long-term outcome of Hurler syndrome following bone marrow transplantation	24-C
373	Anna-Maria Wiesinger	Unraveling the gut puzzle: Lessons from irritable bowel syndrome in managing gastrointestinal symptoms in pediatric Fabry disease	24-D
374	Anna-Maria Wiesinger	Global insights into parental perception of treatment options for mucopolysaccharidosis: A collaborative international survey for advancing clinical trials	25-A
375	Anna-Maria Wiesinger	A decision analysis framework for individualized immunomodulatory therapy in MPS: Early clinical insights	25-B
376	Anna-Maria Wiesinger	Prepared and empowered for Pompe emergencies - PE2	25-C
383	Chia-Feng Yang	Long-term outcomes of very early treated infantile-onset Pompe disease with the improvement after 24-month switching to avalglucosidase alfa: Real-world experiences based on Taiwan nationwide newborn screening program	25-D
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LB-04	Christiane Auray-Blais	Metabolomic and lipidomic profilings for mucopolysaccharidosis type II patients	26-C
LB-05	Christiane Auray-Blais	High-risk screening for lysosphingolipidoses in urine dried on filter paper using tandem mass spectrometry	26-D
LB-06	Swathi Balaji	Comparison of enzyme and molecular based newborn screening for Pompe, Gaucher, and acid sphingomyelinase deficiency: A 10-year review of cases at Ann & Robert H. Lurie Children's Hospital of Chicago	27-A
LB-09	Kimberly Beasley	Newborn screening for Pompe disease and MPS I: The Iowa experience (2023-25)	27-B

LB-11	Samantha Behunin	Characterizing the natural history and clinical spectrum of β-mannosidosis through the CORDS registry	27-C
LB-12	Lisa Berry	From newborn screen to grandfather clock: A wide spectrum of attenuated Hunter syndrome cases	27-D
LB-15	Elizabeth Braunlin	Effect of early hematopoietic cell transplantation (HCT) on cardiac valves in mucopolysaccharidosis type I (MPS I)	28-A
LB-16	Charlotte Chanson	The MLD Alliance: Facilitating advancement of a candidate condition to screen-ready status	28-B
LB-20	Jonathan Cooper	S1S3 phosphotransferase mediated hyperphosphorylation on N-glycans improves the efficacy of gene therapy for CLN2 disease	28-C
LB-23	Sean Ekins	Safety, pharmacokinetics and central nervous system distribution of intracerebroventricular delivered rhPPT1 in cynomolgus monkey	28-D
LB-32	Rachel Hickey	Alternative dosing strategy for enzyme replacement therapy with cerliponase alfa	29-A
LB-33	Michaël Hocquemiller	Intracisternal vs intravenous rAAV9 gene therapy for acid ceramidase deficiency related disorders: Safer CNS targeting in NHPs	29-B
LB-39	Sujeong Kim	Intracerebroventricular administration of recombinant human heparan-N-sulfatase (GC1130A) reduces brain heparan sulfate levels and restores cognitive behavioral function in MPS IIIA mice	29-C
LB-41	Ashley Lahr	Two case examples of inconclusive newborn screens for mucopolysaccharidosis type II	29-D
LB-43	Hsiang-Yu Lin	A simplified quantitative assay for catabolic mannose-containing N-Glycans enables accurate diagnosis of alpha-mannosidosis	30-A
LB-44	Mackenzie Mcneely	Economic based proposal for increasing diagnosis for lysosomal disorders (LD)	30-B
LB-45	Mackenzie Mcneely	An innovative data tool for summarizing lysosomal disorder (LD) newborn screening implementation across the United States	30-C
LB-49	Jasper Mullenders	PLA2G15: a novel therapeutic target for lysosomal diseases	30-D
LB-54	Andreas Øberg	Nationwide newborn screening for metachromatic leukodystrophy (MLD) in Norway: First-year implementation and performance	31-A
LB-55	Virginia Kimonis	Genotype-phenotype correlations and clinical reports of new patients with β-mannosidosis	31-B
LB-56	Cara O'Neill	Psychosocial burden in parents of individuals with Sanfilippo syndromes enrolled in a parent support program: Preliminary findings	31-C
LB-57	Yan Ouyang	Phase 2 study of AL01211 in treatment-naïve classic male Fabry patients: Results from the primary 6-month stage and extension	31-D
LB-58	Cristobal Passalacqua	Preclinical and clinical safety and activity of AB-1009: A new AAV gene therapy for Pompe disease	32-A
LB-65	Boris Sevarika	Nanotechnology-enabled enzyme replacement therapy: Delivery enhancement and immunogenicity elimination through encapsulation	32-B
LB-66	Volkan Seyrantepe	AAVrh10-mHexa administration slightly improves neuropathological outcomes in a mouse model of Tay-Sachs disease by reducing neuronal loss and reactive astrogliosis	32-C
LB-67	Alyaa Shmara	Fused MANBA enzyme CNS delivery and therapeutic response in a mouse model of beta mannosisdosis	32-D
LB-68	Joseph Skeate	Engineered T cell micropharmacies as a platform for in vivo enzyme replacement therapy in lysosomal disorders	33-A
LB-76	Raquel van Gool	Noninvasive markers of central and musculoskeletal symptoms in infantile- and late-onset Pompe disease	33-B
LB-78	Shrijay Vijayan	Gemfibrozil attenuates disease progression in mouse model of Krabbe disease: Basis for the initiation of SOTERIA, a phase 2 clinical trial of PLX-200 (newly formulated gemfibrozil) in lysosomal disorders	33-C
LB-79	Hongling Zhu	Liver-directed AAV gene therapy for CNS delivery of beta-hexosaminidase A in a mouse model of GM2 gangliosidosis	33-D

Poster Session III: Clinical Applications

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4	Patricio Aguiar	Evaluating the relationship between antidrug antibodies and infusion-related reactions/safety outcomes in patients with Fabry disease receiving enzyme replacement therapy (ERT): A systematic literature review	1-B
6	Hera Akmal	Expanded access use of intravenous gene transfer with AAV9 vector galactosidase in child with type II GM1 gangliosidosis	1-C
7	Laura Akroyd	Pompe physiotherapy service improvement - facilitating monitoring in a specialist metabolic centre	1-D
9	Krishan Almeida	Joint arthroplasty is an effective treatment option in managing hip pain in Gaucher disease	2-A
13	Hülya Apaydin	Supporting shared decision-making in Pompe disease: Making complex treatment evidence accessible for patients	2-B
15	Carolina Araujo	Real world data of patients with lysosomal acid lipase deficiency under enzyme replacement therapy experience from a reference center in São Paulo, Brazil	2-C
16	Carmen Arellano	Induction of immune tolerance in a patient with type 3 Gaucher disease	2-D
22	Olga Azevedo	followME Fabry Pathfinders real-world registry in Spain and Portugal: Cardiac and renal outcomes with migalastat in patients with Fabry disease	3-A
23	Abdelghani Bachir Cherif	Benefits of measurement of left atrial size and function in the early diagnosis of Fabry disease	3-B
27	Maryam Banikazemi	Cardiac scoring in Fabry disease: From missed opportunities to improved outcomes	3-C
28	Maryam Banikazemi	Neutralizing antibodies in Fabry disease CRIM-like stratification, immunomodulation, and substrate reduction therapy (SRT)	3-D
29	Maryam Banikazemi	Project searchlight: Implementation and evaluation of a rare disease algorithm to identify persons at risk of Gaucher disease using electronic health records in the United States	4-A
32	Karen Bean	Healthcare resource use for mucopolysaccharidosis type III (MPS III) patients in the United States based on analysis of claims data	4-B
33	Michal Becker-Cohen	Longitudinal follow-up of GBA1-carriers for prodromal features of Parkinson disease, Sidransky syndrome	4-C
34	Amy Berger	A phase I, multi-center, open-label study design to evaluate the safety, tolerability, pharmacokinetics, and pharmacodynamics of DNL952 in adult participants with late-onset Pompe disease	4-D
35	Maria Ester Bernardo	Sustained supraphysiological alpha-L-iduronidase (IDUA) activity, reduction of glycosaminoglycans (GAGs), and clinical benefits at 5 years post-treatment with OTL-203, an autologous hematopoietic stem cell gene therapy (HSC-GT), in patients with mucopolysaccharidosis type I (MPS IH) Hurler syndrome	5-A
36	John Bernat	Isaralgagene civaparvovec (ST-920) shows positive mean annualized eGFR slope in adults with Fabry disease: Topline results from the registrational phase 1/2 STAAR gene therapy study and long-term follow-up study	5-B
37	John Bernat	Long-term safety and efficacy of pegunigalsidase alfa in patients with Fabry disease: Results from the phase III BRILLIANCE extension study	5-C
40	Lisa Berry	Real-world utilization and adherence patterns of enzyme replacement therapy in Fabry disease: Survey results from 238 patients in the USA and Canada	5-D
41	Caroline Hastings	Real-world safety and effectiveness of arimoclomol in patients with NPC: Outcomes from the US early access program (EAP) over a 4-year period	6-A

42	Elizabeth Berry-Kravis	Adrabetadex treatment in individuals with Niemann-Pick disease type C1 re-establishes cholesterol trafficking, resulting in decreased markers of neuronal damage and cell death	6-B
43	Elizabeth Berry-Kravis	Substantial survival benefit with adrabetadex treatment in individuals with infantile-onset Niemann-Pick disease type C (NPC)	6-C
45	Daniel Bichet	Pharmacological chaperone therapy for Fabry disease in the Canadian Fabry Disease Initiative (CFDI) registry	6-D
46	Brian Bigger	Sustained biochemical correction and improved neurological outcomes at 36-months post hematopoietic stem cell gene therapy for Sanfilippo syndrome	7-A
51	Olivia Brown	Adherence and persistence in patients with lysosomal diseases at an academic health system specialty, home infusion, and ambulatory infusion pharmacies: A retrospective review	7-B
54	Philippe Campeau	Improvements in quality of life outcomes among individuals with MPS IVA treated with elosulfase alfa: Results from the Morquio A Registry Study (MARS)	7-C
55	Maria Camprodon-Gomez	From childhood to adulthood: A descriptive analysis of clinical features in patients with acid sphingomyelinase deficiency (ASMD)	7-D
60	Daniela Castillo Garcia	Long-term cardiac outcomes in attenuated MPS I patients under enzyme replacement therapy: A retrospective study from paediatric to adult metabolic centres	8-A
63	Magdalena Ceron-Rodriguez	MPS IV, Morquio disease: Treatment with elosulfase, vitamin d, and zoledronic acid. A strategy that impacts growth and bone health in patients	8-B
64	Irene Chang	Divergent neurocognitive outcomes in mucopolysaccharidosis (MPS) type IIIB siblings: Tralesinidase alfa versus supportive management	8-C
65	Irene Chang	Tividenofusp alfa treatment in a male sibling pair with non-neuronopathic mucopolysaccharidosis type II (MPS II)	8-D
72	Tanya Collin-Histed	Empowering sustainable Gaucher communities	9-A
73	Tanya Collin-Histed	Global reach and equity in lysosomal diseases: The role of the Global Lysosomal Storage Disease Collaborative in access, research, and regulation	9-B
75	Kathleen Coolidge	Bridging the gap in lysosomal disorders drug development: A call for endpoint innovation and economic foresight	9-C
76	Brittany Cooper	Expanded carrier screening leading to an unexpected diagnosis of a lysosomal disorder	9-D
77	Sandra Cowie	The NPC Suspicion Index: Re-establishing a valuable tool for earlier detection of Niemann-Pick disease type C	10-A
82	Patrick Deegan	Delayed diagnosis and missed opportunities: Results from an analysis of the diagnostic journey for LOPD in the UK	10-B
84	Jordi Diaz-Manera	The Spanish Pompe Registry: Real-world insights into clinical characteristics and therapeutic transitions in late onset Pompe disease	10-C
86	Jordi Diaz-Manera	Quantification of muscle glycogen using 13C-MRS as a potential biomarker in late-onset Pompe disease	10-D
87	Jordi Diaz-Manera	Evaluating Glc4 as a longitudinal marker of functional decline in late-onset Pompe disease	11-A
88	Jordi Diaz-Manera	Update on FORTIS: A phase 1/2 open-label clinical trial on AT845 gene replacement therapy for late-onset Pompe disease	11-B
90	Imke Ditters	Home infusion with recombinant human α-glucosidase in classic infantile and other children with Pompe disease is safe: The Dutch experience over 20 years with enzyme replacement therapy	11-C
91	Laurence Djatche	Patient journey and burden of type 3 Gaucher disease in France: A retrospective chart review study	11-D
92	Aimee Donald	The neuroimaging phenotype of neuronopathic Gaucher disease: Evidence of vascular and inflammatory components of neuronopathic disease?	12-A

93	Aimee Donald	Lentiviral stem cell gene therapy of neuronopathic Gaucher disease (GD3) achieves prolonged enzyme delivery, substrate reduction and stabilisation of neurologic and somatic disease manifestations	12-B
94	Aimee Donald	Inflammatory bowel disease in Niemann Pick disease type C: Observations & proposal for surveillance	12-C
95	Aimee Donald	Safety of AAV gene therapy programs for neurological indications - learning from programs in non-lysosomal disorders	12-D
96	Aimee Donald	Interim results from the PROVIDE Clinical Trial - A phase I/II Study of LY3884961(PR001) an AAV9-based gene therapy for type 2 Gaucher disease	13-A
115	Can Ficicioglu	Effect of clemidsogene lanparvovec (RGX-121), an investigational gene therapy, on neurodevelopmental outcomes in patients with Hunter syndrome	13-B
116	Viani Figueroa	Early multicenter experience with standardized approaches to seizure outcome measurement in pediatric epilepsy trials	13-C
118	Emma Flordal Thelander	Clinical characterization and healthcare usage in Fabry disease using Swedish national registers	13-D
119	Ellen Fung	Early efficacy of vosoritide for the treatment of growth deficits in MPS IVA and VI	14-A
125	Dominique Germain	Lucerastat, an investigational oral substrate reduction therapy in Fabry disease: Kidney biopsy results from the MODIFY open-label extension study	14-B
126	Dominique Germain	Hidden double hits in Fabry disease: A critical confounder in clinical trials	14-C
127	Vera Gielen	A structured methodology for evaluating patient-reported outcomes across a treatment program: A comparative application in migalastat	14-D
128	Vera Gielen	Matching-adjusted indirect comparisons (MAICs) and network meta-analyses (NMAs) of the oral small-molecule chaperone migalastat versus intravenous enzyme replacement therapies (ERTs) for clinical measures in Fabry disease	15-A
131	Roberto Giugliani	International Niemann-Pick Disease Registry (INPDR): Enrollment characteristics of patients enrolled in the largest database of patients with ASMD	15-B
132	Roberto Giugliani	A prospective natural history study for GM1 and GM2: 24-month data from the PRONTO study	15-C
133	Roberto Giugliani	Sustained cognitive and adaptive behavior outcomes of long−term treatment with pabinafusp alfa in patients with severe or attenuated mucopolysaccharidosis type II	15-D
134	Roberto Giugliani	Long-term data from a phase II study with oral nizubaglustat for late-infantile/juvenile GM2 and NPC diseases (RAINBOW)	16-A
135	Ozlem Goker-Alpan	A prospective dose escalation study with ambroxol in patients with Sanflippo syndrome	16-B
136	Ozlem Goker-Alpan	Individual participant outcomes after FLT201 AAV gene therapy for type 1 Gaucher disease: Two-year biomarker and clinical data	16-C
137	Ozlem Goker-Alpan	Safety, tolerability and biological activity of ABX1100, a CD71 centyrin siRNA conjugate targeting GYS1 in late-onset Pompe disease patients	16-D
141	Domingo Gonzalez-Lamuno Leguina	Therapeutic exercise in Pompe disease: Evidence from 2012-2025 and future directions for metabolic impact assessment	17-A
146	Christina Grant	Infantile onset Pompe disease patients switching from weekly alglucosidase to avalglucosidase show improved biomarkers and gross motor skills: A case series	17-B
152	Giorgia Gugelmo	Physical fitness and physical function in patients with Fabry disease: A cross-sectional multicenter study	17-C
153	Punita Gupta	Dual diagnosis of Hunter syndrome and Coffin Siris syndrome	17-D
158	Paul Harmatz	The effect of the investigational gene therapy clemidsogene lanparvovec (RGX-121) on long-term levels of D2S6 in cerebrospinal fluid in patients with Hunter syndrome	18-A
159	Caroline Hastings	Intravenous hydroxypropyl-beta-cyclodextrin (HPBCD): Biomarkers, efficacy, and impact of dosing and administration route on therapeutic index	18-B

160	Caroline Hastings	Addressing a drug delivery controversy: Evaluation of the data for IT versus IV administration of hydroxypropyl-beta-cyclodextrin for the treatment of Niemann Pick disease type C patients	18-C
162	Christian Hendriksz	Enabling multinational data collaboration through scalable language translation in rare disease registries: A pilot study from Taiwan	18-D
163	Julia Hennermann	Effectiveness of idursulfase in patients with mucopolysaccharidosis type II: An international (non-US), retrospective chart review of treatment initiation in very young patients	19-A
167	Julia Holdorp	Assessment of cognitive functioning using the Vineland Adaptive Behavior Scale in patients with mucopolysaccharidosis type II	19-B
168	Gea-Ru Hong	Long-term safety and effectiveness of migalastat in patients with Fabry disease: Results from the Korean post-marketing surveillance	19-C
175	Derralynn Hughes	Evaluating the relationship between anti-drug antibodies and clinical efficacy in patients with Fabry disease receiving enzyme replacement therapy: A systematic literature review	19-D
176	Derralynn Hughes	When should patients be referred for whole genome sequencing for suspected lysosomal diseases: An international consensus initiative	20-A
178	Derralynn Hughes	Impact of mobility-aid use on late-onset Pompe disease (LOPD) patient experience: Insights from patient interviews	20-B
179	Derralynn Hughes	Long-term effectiveness and safety of agalsidase alfa in patients with Fabry disease treated for at least 19 years: The Fabry Outcome Survey	20-C
183	Elizabeth Jalazo	Preliminary results from phase I/II, first-in-human, open-label study of DNL126 in children with mucopolysaccharidosis type IIIA (MPS IIIA)	20-D
187	Franklin Johnson	Adjusted migalastat dose regimens in patients with Fabry disease and amenable GLA variants with severe renal impairment, or with end-stage renal disease and receiving hemodialysis/hemodiafiltration (HD/HDF): Pharmacokinetic (PK) and safety results from a protocol-specified interim analysis of the RENEW study	21-A
188	Mairead Jones	A case review of a man with late onset Pompe disease who has switched enzyme replacement therapies: Clinical and physiotherapy outcomes	21-B
189	Mairead Jones	A virtual physiotherapy led exercise class for late onset Pompe disease in a tertiary adult metabolic centre	21-C
190	Mairead Jones	Cause of death and health inequalities in late onset Pompe disease: A single centre experience	21-D
191	Simon Jones	Ex-vivo modification of autologous CD34+ HSPCs using a CD11b-directed lentiviral vector encoding ApoEII-tagged human IDS leads to supraphysiological enzyme activity and biochemical correction of neuronopathic MPS II patients	22-A
193	Kristina Julich	Multi-year subgroup analyses of Niemann-Pick disease type C participants treated with arimoclomol in the US early access program	22-B
196	Ilkka Kantola	Do we need lysoGb3 measurement in monitoring Fabry disease?	22-C
199	Thomas Kenny	True faces of rare: Authenticity over aesthetics shaping a new visual language for Fabry and other rare disease communications	22-D
200	Terry Kho	Baseline characteristics and medical history for participants enrolled in the MPS VII disease monitoring program (DMP)	23-A
202	Anhye Kim	A phase I first-in-human, single- and multiple- ascending dose study of glucosylceramide synthase (GCS) inhibitor YH35995 in healthy adult male participants	23-B
204	Priya Kishnani	Symptom onset, disease biomarkers, and treatment status in US Pompe disease patients identified by newborn screening	23-C
205	Priya Kishnani	208-week outcomes of cipaglucosidase alfa plus miglustat in patients with late-onset Pompe disease treated from PROPEL baseline: Pulmonary function	23-D
206	Priya Kishnani	90-month pulmonary function outcomes with cipaglucosidase alfa plus miglustat (cipa+mig) in adults with Pompe disease in ATB200-02, an open-label phase I/II study	24-A

212	Sandra Kyosen	National insights into the diagnostic odyssey of Fabry disease patients in Brazil: Data from the RARAS network	24-B
213	Sandra Kyosen	National insights into value-based care for Gaucher disease: Findings from the Brazilian JAV-RARAS project	24-C
215	Dawn Laney	Early disease manifestations and treatment outcomes in pediatric Fabry disease patients treated with agalsidase beta in the United States	24-D
216	Dawn Laney	Exploring the distinct challenges and unmet needs of female patients with Fabry disease: Findings from a cross-sectional survey of 238 participants	25-A
217	Dawn Laney	Changing natural history: A case series of causes of death in individuals with classic Fabry disease who began primary therapy later in disease progression	25-B
218	Heather Lau	Treatment with UX111 reduced cerebrospinal fluid (CSF) heparan sulfate (HS) exposure and stabilized or improved functioning across dose, age, and stage of MPS IIIA	25-C
219	Victoria Lawson	Characterizing peripheral pain and organ involvement at diagnosis in patients with Fabry Disease: Insights from the Fabry Registry	25-D
224	Austin Letcher	Cystinosis: The emerging adult phenotype	26-A
226	Alaena Lim	Barriers and carriers: A chart review investigating the ethnoracial disparities of the diagnosis, testing, and treatment for Fabry disease	26-B
230	Laura López de Frutos	A phase III study to evaluate the efficacy of nizubaglustat on GM1, GM2, and NPC diseases (NAVIGATE)	26-C
232	Yanmei Lu	Isaralgagene civaparvovec (ST-920) gene therapy for adults with Fabry disease: Pharmacology and immunogenicity outcomes from the phase 1/2 STAAR study and ongoing long-term follow-up	26-D
235	Helen Lycett	Insights from patients, caregivers, and healthcare professionals on the challenges of lysosomal disorders	27-A
237	Barbara MacFee	Outcomes of patients with rapidly progressive lysosomal acid lipase deficiency treated with sebelipase alfa before hematopoietic stem cell transplantation	27-B
241	Shauna Mangum	Understanding the burden of type 1 Gaucher disease: Pain, fatigue, and age and their effects on health-related quality of life	27-C
244	Thorsten Marquardt	ROSSELLA: An ongoing open-label, multicenter, global trial to study next-generation treatment of infantile-onset Pompe disease combining enzyme replacement with a stabilizing iminosugar	27-D
245	Ana Maria Martins	Long-term somatic efficacy of pabinafusp alfa across a broad spectrum of age groups and phenotypes in patients with mucopolysaccharidosis type II	28-A
247	Paul McIntosh	From frustration to function: Reimagining registry value with individual monitoring dashboards to visualize disease progression in Pompe disease	28-B
248	Kristin McKay	Quality of life (QoL), unmet needs, and treatment experience of people living with mucopolysaccharidosis type II (MPS II) and their caregivers: A community survey	28-C
250	Eugen Mengel	Efficacy of arimoclomol combined with miglustat at months 3, 6, 9 and 12 of the double-blind, randomized, placebo-controlled NPC002 trial	28-D
251	Jonathan Meyer	Single-centre experience with pegunigalsidase alfa-iwxj in treatment-naïve and switched Fabry patients: Safety, and immunogenicity	29-A
253	Carmen Minea	Vascular calcification in cholesteryl ester storage disease	29-B
254	Pramod Mistry	Beyond ERT/SRT: A neuro-pulmonary-lymphatic spatial-omics atlas redefines pathology and guides immunotherapy in neuronopathic Gaucher disease	29-C
257	Tahseen Mozaffar	208-week outcomes of cipaglucosidase alfa plus miglustat in patients with late-onset Pompe disease treated from PROPEL baseline: Muscle function and biomarkers	29-D
258	Joseph Muenzer	Phase I/II study of intravenous tividenofusp alfa for mucopolysaccharidosis type II	30-A

259	Nicole Muschol	Long-term administration of tralesinidase alfa enzyme replacement therapy (TA-ERT) results in profound and durable reduction of heparan sulfate (HS) and stabilization of cognitive function and cortical gray matter volume (CGMV) in patients with Sanfilippo syndrome type B (MPS IIIB)	30-B
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