POSTERS

01. Amino acid disorders	Submitter name		Abstract title
P-001	Belo	Anabela	Case Study: Methylmalonic Acidemia and Episodes of Pancreatitis Requiring Parenteral Nutrition
P-002	Bieneck Haglind	Charlotte	Health-related quality of life in children with phenylketonuria: a study using the DISABKIDS-37 module
P-003	Boudabous	Hela	Early Targeted Neonatal Screening and Treatment in Type I Tyrosinemia: Impact on Complication Prevention
P-004	Burgaç	Ezgi	Food Neophobia in Hyperphenylalaninemia: An Overlooked Aspect of Feeding Behavior
P-005	Candela	Egidio	Clinical Presentation and Treatment Strategies in Pediatric Alkaptonuria: A Systematic Review
P-006	Chalazan	Brandon	Large Single Center Respective Study - Asparaginase Induced Hyperammonemia
P-007	Cheon	Chong Kun	AAV-Based Gene Therapy Restores Metabolic Function in Liver Organoid Models of Maple Syrup Urine Disease
P-008	Choi	Shinjie	The diagnostic accuracy of amino acid ratios in neonatal intrahepatic cholestasis caused by citrin deficiency
P-009	Del'haye	Gigly	Generation of FAH and SLC7A11 knockouts to study metabolism in tyrosinemia type 1
P-010	Demirbaş Yülüklü	Göksu	Late-Onset Citrullinemia Type 1 Diagnosed During Evaluation of a Bleeding Disorder: A Case Report
P-011	Dios Fuentes	María Elena	Biochemical outcomes and clinical features of nitisinone therapy in alkaptonuria: experience from an adult cohort
P-012	Dolins	Karen	Disease burden and quality of life: findings of the MSUD patient registry
P-013	García Arenas	Dolores	Functional activity assessment and targeted nutrition workshop for adolescents and adults with amino acid disorders
P-014	Gülbahçe	Aliye	When Episodic Ataxia Is Metabolic: A Late Onset Presentation of HIBCH Deficiency
P-015	Ito	Tetsuya	Successful Preconception Use of Pegvaliase with Classical Phenylketonuria Followed by Dietary Management During Pregnancy
P-016	Köşeci	Burcu	The Diagnostic Importance of Adenosine Kinase Deficiency in a Case Presenting with Cholestasis and Hypoglycemia
P-017	Halak	Renata	Metabolic Profile Assessment in Patients with STXBP1-Related Encephalopathy
P-018	Kozich	Viktor	Genetic architecture of CBS deficiency: data from E-HOD registry
P-019	Niño-Traslaviña	Katherin	Age-related variation and cross-fluid relationships of amino acids in biological fluids using HPLC
P-020	Niño-Traslaviña	Katherin	Amino acid correlation networks in patients with diagnosed aminoacidopathies
P-021	Leipnitz	Guilhian	Nicotinamide riboside improves mitochondrial dynamics and reduces superoxide levels in fibroblasts with sulfur metabolism disorders
P-022	Mccarron	Eamon	Management of pregnancy in lysinuric protein intolerance complicated by immune dysregulation and severe thrombocytopenia
P-023	Messina	Martina	Clinical Spectrum, Long-Term Management, and Outcomes in Classic and Attenuated Isovaleric Acidaemia
P-024	Mohamed	Sarar	An ultra-rare condition revealed by WES in a child with brittle hair and hypotonia.
P-025	Murphy	Elaine	Pregnancy in maternal isovaleric acidaemia (IVA): service level experience of management and outcomes
P-026	Nardecchia	Francesca	A novel electroclinical phenotype resembling Lennox–Gastaut syndrome in dihydropteridine reductase deficiency: a case report
P-027	Ngoc Nguyen	Khanh	Long-term Outcomes of MSUD at a Vietnam Referral Center
P-028	Niinikoski	Harri	Clinical characteristics and treatment of Finnish patients with lysinuric protein intolerance.
P-029	O Shea	Darragh	Correlating neuroimaging, clinical findings and leucine levels in Irish patients with maple syrup urine disease
P-030	Özgen	Yasemin	Pre-symptomatic Propionic Acidemia Detected During Evaluation of DYNC2H1-Related Skeletal Dysplasia
P-031	Rallapalli	Anvitha	From Lens Dislocation to Stroke: Clinical, Neuroimaging and Genetic Spectrum of Classical Homocystinuria
P-032	Salazar	María	Leucine Tolerance, Metabolic Control and Neurodevelopmental Outcomes from Genetic Variants in a Chilean MSUD Cohort
P-033	Sharma	Reena	Remethylation defects and acquired vitamin B12 deficiency- diagnostic algorithm in one adult metabolic centre.
P-034	Stanescu	Sinziana	Neurocognitive Manifestations in Two Patients With Tyrosinemia Type I on Long-Term Nitisinone Treatment
P-035	Streck	Emilio	Minocycline prevents neurochemical alterations induced by the administration of branched-chain amino acids in Wistar rats
P-036	Streck	Emilio	Resveratrol improves neurochemical parameters in rats submitted to a Maple Syrup Urine Disease protocol
P-037	Sundaravel	Selvamanojkumar	Diverse clinical and molecular spectrum of MTHFR deficiency – Experience from a tertiary care centre
P-038	Tandogan	Zelal	Digital Management of Phenylketonuria: Impact of a Mobile Application on Metabolic Control and Dietary Adherence
P-039	Topuz	Hanim Seyma	Methionine-Restricted Diet in Sulphite Oxidase Deficiency Associated with a SUOX Variant: Clinical Improvement in a Paediatric Case
P-040	Utassy	Kata Rebeka	Comorbidities of Adult Alkaptonuria Patients Living in Hungary
P-041	Van Meijel	Rens	Comparison of creatinine- and cystatin C–based estimated glomerular filtration rate in adults with phenylketonuria
P-042	Verde	Alessandra	“String of Pearls” before diagnosis in MOCS2-Related Molybdenum Cofactor Deficiency
P-043	Wajner	Moacir	Redox-dependent mitochondrial dysfunction induced by the pathognomonic metabolite of tyrosinemia type I in rat kidney
P-044	Wajner	Moacir	Succinylacetone markedly disrupts hepatic bioenergetics: a potential mechanism underlying liver injury in tyrosinemia type I
P-045	Wajner	Moacir	L-carnitine prevents lipopolysaccharide and α-ketoisocaproic acid-induced neuroinflammation and mitophagy in neonatal rat cerebral cortex
P-046	White	Lucy	Case study: Low protein diet in siblings diagnosed with S-adenosylhomocysteine hydrolase deficiency; a successful treatment?
P-047	Wortmann	Saskia	Lysinuric Protein Intolerance: Allogeneic Peripheral Blood Stem Cell Transplantation for an Inborn Error of Metabolism&Immunity
P-048	Wyse	Angela TS	Homocysteine alters markers of blood-brain barrier and p-TAU217 in brain of rats
P-049	Wyse	Angela TS	Hyperhomocysteinemia impairs memory and causes NeuN loss in male rats: GFAP expression as sex-specific predictors

P-050	Wyse	Angela TS	Inflammatory and oxidative status alterations in heart of hyperhomocysteinemic rats: protection by folate and simvastatin
P-051	Yildiz	Yilmaz	Neurological and Psychiatric Phenotypes in Maple Syrup Urine Disease: Interim Cohort Findings
P-052	Zubarioglu	Tanyel	Evidence of Docosahexaenoic Acid Deficiency in Maple Syrup Urine Disease: Insights from Plasma LC-PUFAs Status
P-053	Zubarioglu	Tanyel	Early Laboratory Indicators of Acute Metabolic Decompensation During Emergency Presentations in Maple Syrup Urine Disease
02.Biophysics and chemical physics	Submitter name		Abstract title
P-054	Lundqvist	Thomas	Lorenzo's Oil and Reversible Cytopenias in X-Linked Adrenoleukodystrophy: A Membrane-Centered Case Study
03.Carbohydrate disorders, excluding CDG's	Submitter name		Abstract title
P-055	Aghakıshılı	Hanım	Empagliflozin Therapy in Fanconi-Bickel Syndrome: A Single-Center Case Series
P-056	Alirzayeva	Leman	A novel SLC16A2 variant in a male infant with severe hypotonia and thyroid dysregulation
P-057	Aras Çöl	Tuğçe	Clinical, Genetic And Long-Term Outcomes İn Galactosemia: A Single-Center Cohort Study
P-058	Araujo	Carolina	Nationwide Real-World Data on Galactosemia in Brazil: Insights from a Multicenter Rare Disease Network
P-059	Baran	Berat	Non-PTH related mild hypercalcemia as a potential biomarker of metabolic decompensation in adult GSD
P-060	Boschma	Tirza	Hepatocellular carcinoma in glycogen storage disease type I: a systematic literature review
P-061	Çiki	Kismet	Remarkable Clinical and Biochemical Response to Dietary Intervention in Generalized UDP-Galactose 4'- Epimerase Deficiency
P-062	Durdağ Öztürk	İrem	A Rare Diagnosis Behind Hypertriglyceridemia Detected in a School Screening: Glycerol Kinase Deficiency
P-063	Felipe	Ana	"Classic Galactosemia? Following the Diagnosis of a Leukodystrophy"
P-064	Doederlein Schwartz	Ida Vanessa	Glycogen storage disease type I in Brazil: genotype-phenotype correlations in 38 patients
P-065	Rubio-Gozalbo	M Estela	Clinical translation gaps and strategies to advance clinical trials in CG: a Galactosemia Network review
P-066	Gugelmo	Giorgia	Comparing Continuous Glucose Monitoring Alert Strategies, Accuracy, and Patient-Reported Outcomes in Adults with IMDs
P-067	Gülbahçe	Aliye	Beyond Uncertain Significance: PCK1 Variants and the Diagnostic Complexity of PEPCK Deficiency
P-068	Jalajas	Kärt	Prenatal Diagnosis and Early Management of Hereditary Fructose Intolerance: A Case Report
P-069	Karppinen	Sinikka	Use of empagliflozin in three children with Fanconi-Bickel syndrome
P-070	Kieu	Quynh	SLC2A1-Related GLUT1 Deficiency: A Fifteen-Year Diagnostic Odyssey
P-071	Konopka	Agnieszka	From hypoglycaemia to ketosis. A novel approach in Congenital Hyperinsulinism - case study.
P-072	Matsui	Miki	A Case of Hereditary Fructose Intolerance Diagnosed in Infancy with Marked Hepatomegaly and Fatty Liver
P-073	Mccarron	Eamon	Cardiac involvement in adults with Glycogen Storage Disease Type IIIa: an observational cohort study
P-074	Nguyen	Thu Ha	Clinical and molecular features of fructose-1,6-bisphosphatase deficiency in Vietnamese children
P-075	Paci	Sabrina	Atypical Mild Presentation of Glycogen Storage Disease Ib in Two Adolescent Sisters: A Case Report
P-076	Peñafiel-Freire	Diego Mauricio	Triosephosphate isomerase deficiency: case report
P-077	Pretese	Roberta	Glycogen storage disease 3: low carbohydrate diet, biochemical parameters and left ventricular size
P-078	Doederlein Schwartz	Ida Vanessa	Global Prevalence of Galactosemia and the Role of Neonatal Screening: A Scoping Review
P-079	Sdogou	Triantafyllia	GALE Deficiency Detected Through the Greek National Newborn Screening Program: Molecular Heterogeneity and Genotype–Phenotype Correlation
P-080	Sdogou	Triantafyllia	Galactokinase(GALK) Deficiency Identified Through the Greek National Newborn Screening Program: Molecular Spectrum and Clinical Outcomes
P-081	Stojiljkovic	Maja	Comparative Analysis Before and During SGLT2 Inhibitor Therapy of 35 Glycogen Storage Disease 1b Patients
P-082	Veiga-da-Cunha	Maria	Loss of glucose-6-phosphate inhibition drives hexokinase 1 hyperactivity in NEDVIBA-associated HK1 variants
P-083	Wang	Raymond	Cognitive impact of mucopolysaccharidosis VII (MPS VII): Baseline results from the disease monitoring program (DMP)
04.Clinical studies, patient reported outcome measures, real world data	Submitter name		Abstract title
P-084	Adang	Laura	Phenotypic and neuroimaging description of individuals affected with SAMHD1-related Aicardi Goutières Syndrome
P-085	Akyüzlüer Güneş	Setenay Merve	Continuous Renal Replacement Therapy For Acute Metabolic Decompensation In Inborn Errors Of Metabolism: Single-Center Cohort
P-086	Al-humaidi	Suzan	Incidence of newborn screening disorders in Saudi Arabia: a national retrospective study (2013–2017)
P-087	Basan	Hacer	Retrospective Evaluation of Coexisting Second Disorders in Patients with Inherited Metabolic Diseases
P-088	Gürbüz	Berrak Bilginer	Adult Consultations to a Pediatric Metabolic Center: Clinical Spectrum and Diagnostic Outcomes
P-089	Bilgün	Ceren	Liver Transplantation in Inherited Metabolic Diseases: A Single-Center Follow-Up Experience
P-090	Candela	Egidio	Benefits of a Structured Transition Model in Inherited Metabolic Disorders: A Real-World Cohort Study
P-091	Chien	Yin-Hsiu	Alglucosidase alfa to avalglucosidase alfa switch improves biomarkers in infantile-onset Pompe disease: Pompe Registry analysis
P-092	De Castro Lopez	Maria Jose	Pegzilarginase Bridges to Liver Transplant by Dissociating Arginine Neurotoxicity from Glutamine-Driven Ureagenesis in Arginase1 Deficiency
P-093	De Giorgi	Agnese	Neurological-dominant Wilson's disease without significant hepatic involvement: evidence from a 40-year follow-up of two siblings
P-094	Demirdöken	Emine Didem	A rare cause of recurrent acute liver failure in childhood: NBAS and SCYL1 variants
P-095	Ezgü	Fatih S	Trial Update: Intravenous HPβCD TRANSPORT-NPC Pediatric Sub-Study
P-096	Fischinger Moura De Souza	Carolina	Casa dos Raros: an integrated care model for rare diseases in Brazil—first 1,000 patients

P-097	Freiburghaus	Katrin	ERNDIM quantitative external quality assessment improves performance in biochemical genetic testing
P-098	Gil González	Marta	Neurocognitive impairment in treatable inherited metabolic disorders: an integrated neuropsychological, brain volumetry, and lipidomic study
P-099	Güneş	Dilek	Unmet Needs and Experiences of Parents of Children Diagnosed with Mucopolysaccharidosis (MPS)
P-100	Harmeling	Simone	Real world use of arimoclomol in Niemann Pick type C: Findings from a German Compassionate Use Programme
P-101	Hoyle	Kirsty	Community-Driven Development of a National Patient Organisation for Inherited Metabolic Disorders in Sri Lanka
P-102	Cho	Sung Yoon	Intracerebroventricular idursulfase beta in neuronopathic mucopolysaccharidosis type II: First Korean Phase1 biomarker and neurodevelopmental outcomes
P-103	Kerti	Andrea	PHKA2 Variants in Glycogenosis Type IX: Genotype–Phenotype Correlations in a Hungarian pediatric Cohort
P-104	Kilavuz	Sebile	SGPL1 Deficiency and the Limits of S1P Receptor Modulation: First Report of Fingolimod Administration
P-105	Koç Yekedüz	Merve	Cutaneous Clues in Inherited Metabolic Disorders: Insights from a Single-Center Cohort
P-106	Koç Yekedüz	Merve	Retrospective Evaluation of Ocular Findings in Inherited Metabolic Diseases: A 13-Year Single-Center Experience
P-107	Mekdade	Tristan	Atypical Leigh-like presentation in low-excretor glutaric aciduria type I: a multicenter series
P-108	Mengel	Karl Eugen	LEAP2MONO Subgroup Analyses Demonstrate Neurologic Efficacy With Venglustat While Maintaining Systemic Control in GD3
P-109	Ortan Efe	Gülşah	Our experience in the management of pregnancy in cases diagnosed with ınherited metabolic disorders
P-110	Parke	Brittany	Comparative Analysis of Symptomatology and Diagnostic Latency in Three Patients with Homocystinuria Cobalamin G Deficiency.
P-111	Parke	Brittany	Characterizing Behavioral Manifestations in Classical Homocystinuria Using Patient-Reported Data
P-112	Peters	Heidi	Sepiapterin responsiveness and improved protein tolerance in a child with phenylketonuria previously unresponsive to sapropterin
P-113	Poh	Sherry	Late-Onset Molybdenum Cofactor Deficiency Type B Presenting with Stroke-like Episodes and Nystagmus
P-114	Porta	Francesco	Euphenic and non-euphenic inborn errors of metabolism: the role of environmental dependence
P-115	Sailer	Sabrina	Acylcarnitine ratios for monitoring ketogenic diet therapy: A retrospective study
P-116	Sass	Jörn Oliver	IEM Continuum: From Non-Disease to Disease
P-117	Seenivasan	Abinaya	Correlating the Genotype and Phenotypes of Patients with MLD in the UK
P-118	Sharma	Reena	A service evaluation of patient-initiated follow-up – the experience of a large adult IMD service
P-119	Tan	Chern Yan	Natural History and Comorbidities of Childhood Ketotic Hypoglycaemia: Experience from a Regional Specialist Centre
P-120	Seker Yilmaz	Berna	Rare Disease Awareness Among Young People: A Youth-Led Survey of Knowledge, Perceptions, and Educational Needs
P-121	Tosi	Martina	Functional Gastrointestinal Disorders in School-Aged Children with Hyperphenylalaninemia: An Italian Multicentre Survey
P-122	Trang	Nguyen Thi Ngoc	Severe Neonatal Inborn Errors of Metabolism in Southern Vietnam: Burden and Outcomes
P-123	Uzun Dincturk	Duygu	An alternative cause of a Leigh-like phenotype: VAC14-related disease
P-124	Watanabe	Yoriko	Early Detection of Hereditary Urolithiasis Through Urinary GC–MS: Insights from a Nationwide Japanese Cohort
P-125	Wolter	Minou	Expanding the genotypic spectrum of PYCR2-related hypomyelinating leukodystrophy: description of three new cases
P-126	Yoldaş Çelik	Merve	Clinical Assessment of Humoral Immunity and Vaccine Response in Pediatric Inherited Metabolic Disorders
05.Dietetics and nutrition	Submitter name		Abstract title
P-127	Akbulut Cakir	Selin	Long-Term Growth and Metabolic Control Outcomes of On-Demand Breastfeeding in Classical PKU
P-128	Akça Yeşil	Sedanur	Evaluation of Nutritional Status and Psychosocial Effects in Patients with Maple Syrup Urine Disease,Their Families
P-129	Al Nabhani	Ayda	Methionine Supplementation in the MARS1 gene: A case report
P-130	Belo	Anabela	How effective can one carbon be? – a Clinical Case
P-131	Benítez Ávila	Rosa María	Know your formula: a dietary tool to help PKU patients understand their daily protein requirements
P-132	Byers	Laura	Determinants of successful human milk feeding in Inherited Metabolic Disorders (IMD) on dietary treatment.
P-133	Byers	Laura	Association between post-illness low leucine levels and illness type in Maple Syrup Urine Disease (MSUD)
P-134	Candela	Egidio	Dietary management and clinical outcomes in glycogen storage disease type IX: a systematic review
P-135	Caro	Amanda Rocío	ACPEIM: An app for monitoring protein intake developed by a patient association.
P-136	Daly	Anne	Targeted Branched Chain Amino Acid Supplementation Corrects Persistent Deficiency in Urea Cycle Disorders: Five Year Longitudinal Analysis
P-137	Ellerton	Charlotte	Use of Glycomacropeptide (GMP) in Maternal PKU; a multicentre prospective observational study
P-138	Gaff	Lisa	Protein Substitutes: challenges applying International Dysphagia Diet Standardisation Initiative standards for an aging PKU community
P-139	Gillingham	Melanie	Validation of MyRareDiet™, a novel application tracking dietary intake of patients consuming protein restricted diets
P-140	Giorda	Sara	Structured Nutritional Counselling Improves Metabolic Control in Patients with Phenylketonuria
P-141	Koç Yekedüz	Merve	Evaluating the Scientific Validity and Suitability of ChatGPT in Methylmalonic Acidemia Dietary Management
P-142	MacDonald	Anita	Eating disorders and disordered eating in PKU: a systematic review
P-143	Muñoz Alonso	Ana	Morphofunctional assessment in children with inborn errors of protein metabolism
P-144	Pedro	Tope	Impact of ARCT-810 mRNA therapy on dietary intake and biochemical data in participants with OTCD
P-145	Poloni	Soraia	Nutritional Management of Inborn Errors of Metabolism within the Brazilian Health System: A National Perspective
P-146	Poloni	Soraia	Trends in BMI, Protein and Energy Intake During Sepiapterin Treatment in Brazilian Patients with PKU
P-147	Robertson	Louise	Galactosaemia and cheese: cheese analysis for lactose and galactose content in the UK, 2022–2026

P-148	Robertson	Louise	An Exercise and Dietary Induced Fat-Free Mass Increase in Two Patients with Phenylketonuria (PKU)
P-149	Salazar	María	Nutritional Management of Synovial Sarcoma in a Patient with Phenylketonuria: A Case Report
P-150	Strengell	Katrin	Protein intake, plasma amino acid concentrations and growth in ASLD infants
P-151	Tapley	Simon	Timeliness in UK Adult PKU Care: Auditing Dried Blood Spot Turnaround Time and Follow-up Intervals
P-152	Timmer	Corrie	MCT oil is suitable for a diet low in plant sterols.
P-153	Timmer	Corrie	Effect of dietary treatment in adults with AMACR deficiency
P-154	Tosi	Martina	Heterogeneity in Sapropterin Administration Among Paediatric/Adult Patients with Phenylketonuria: Findings from a National Survey
P-155	Tosi	Martina	Short term use of a slow release Phenylalanine free microtablet protein substitute in children and adolescents with Phenylketonuria
P-156	Tosi	Martina	Dietetic Education and Training in Inherited Metabolic Disorders: Results from an SSIEM-Dietitians Group pilot survey
P-157	Tosi	Martina	Breast Milk Composition in Untreated Maternal PKU: Protein, Phenylalanine, and Fatty Acid Analysis
P-158	Tummolo	Albina	Legacy Effect of Diet-Therapy in PKU and T1DM: A Pilot Study
P-159	Uygur	Esma	Impact of Low Phenylalanine Free Diet on Compliance and Metabolic Control in Tyrosinemia Type 1
P-160	Uygur	Esma	On-Demand Breastfeeding in HMG-CoA Lyase Deficiency: Clinical and Metabolic Outcomes of Two Cases
P-161	Van Der Wiel	Adorée	Twin pregnancy in PKU: substantially increased protein and energy needs in a dichorionic gestation
P-162	White	Fiona	Infantile-onset lysosomal acid lipase deficiency – impact of fat intake on growth and duodenal histology.
P-163	Yildiz	Harun	Artificial Intelligence Meets Metabolic Nutrition: How Reliable Are ChatGPT's Dietary Recommendations?
06.Disorders of fatty acid oxidation and ketone body metabolism	Submitter name		Abstract title
P-164	Basan	Hacer	Functional evidence supporting the pathogenicity of a novel homozygous SCP2 variant associated with adult-onset leukoencephalopathy
P-165	Basu	Shakuntala	β-Hydroxybutyrate Formulation Performance Compared to Triheptanoin in Acadvl-/-/Acadl-/- Muscle-Double Knockout Mouse Model
P-166	Belo	Anabela	Use of Human Milk in a Ketogenic Diet in an Patient with Glut-1 Deficiency Syndrome
P-167	Çalışgan	Kağan	Clinical, genetic, and longitudinal dietary characterization of 12 Turkish patients with HMG-CoA lyase deficiency
P-168	Elizondo	Gabriela	Clinical and Molecular Insights into Sudden Cardiac Death in Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
P-169	Imbard	Apolline	CPT1 deficiency: an underdiagnosed condition?
P-170	İşler Soylu	Elif	Pteridine metabolites as biomarkers of inflammation and oxidative stress in fatty acid oxidation disorders
P-171	Ismail	Zona	Factors Affecting Long-Term Outcomes in a Cohort of Adults with LC-FAODs
P-172	Karunanidhi	Anuradha	Dicarboxylic- and Methylmalonyl-D-Dipeptides Outperform as Anaplerotic Treatment in Patients' Fibroblasts with Fatty Acid Oxidation Disorders
P-173	Kurkjian	Brooke	Pyruvate Carboxylase Deficiency: Combination Anaplerotic and Cofactor Therapies in Presumed Type B Presentation
P-174	Madeira	Catarina A.	FAD supplementation for the rescue of medium-chain acyl-CoA dehydrogenase p.K304E variant
P-175	Martin Hernandez	Elena	Fatty acid oxidation disorders detected by newborn screening: clinical, biochemical, outcomes in a single-center cohort
P-176	Mercimek-Andrews	Saadet	Development and validation of a clinical severity score for long-chain fatty acid oxidation disorders
P-177	Mercimek-Andrews	Saadet	Exploratory immunophenotyping in two patients with very long chain acyl-CoA dehydrogenase deficiency
P-178	Olsson	David	Incretin and gut peptide regulation of fasting in children with beta-oxidation disorders
P-179	Sasai	Hideo	Starvation induces a shared adaptive transcriptional response in fibroblasts from ketone body utilization disorders
P-180	Skeath	Rachel	Multiple acylCoA dehydrogenase deficiency (MADD): single-centre review of clinical, dietetic and biochemical disease severity spectrum
P-181	Suárez González	Marta	Anthropometric nutritional assessment and use of carbohydrates in pediatric patients with Medium-Chain Acyl-CoA Dehydrogenase Deficiency
P-182	Tan	Shyh-Yeu	Efficacy of Triheptanoin in Long-Chain Fatty Acid Oxidation Disorders: A Systematic Review and Meta-Analysis
P-183	Venegas Moreno	Eva María	Clinical and biochemical characterization of fatty acid β-oxidation disorders: an adult single-center cohort
P-184	Verde	Alessandra	Beyond genetics: unmasking Multiple Acyl-CoA Dehydrogenase Deficiency through functional analysis in an adult patient
P-185	Yüksel Yanbolu	Ayşe	Clinical Diversity of Multiple Acyl-CoA Dehydrogenase Deficiency: A Comprehensive Long-Term Follow-up of 16 Cases
07.Disorders of purines, pyrimidines, nucleic acids and porphyrias	Submitter name		Abstract title
P-186	Gregoric	Nadan	Regression of indeterminate liver lesions after givosiran therapy in acute intermittent porphyria: an unusual case
P-187	Hall	Patricia	Performance of Plasma Maximum Fluorescence Wavelength with Acidic Preparation for the Classification of Porphyrias
P-188	Nakajima	Yoko	Targeted LC–MS/MS Profiling of Blood Pyrimidine Related Metabolites in CAD Deficiency
P-189	Sucur	Nediljko	Kidney function improvement with empagliflozin in acute intermittent porphyria: a case report
P-190	Topuz	Hanim Seyma	Refractory Status Epilepticus in Adenylosuccinate Lyase Deficiency: Clinical Response to Dietary and Supportive Interventions
P-191	Tortorelli	Silvia	Complex porphyria diagnoses: expanding the clinical and biochemical phenotype of individuals with biallelic HMBS variants
08.Disorders of vitamins, cofactors and trace elements	Submitter name		Abstract title
P-192	Amartino	Hernan	Progressive retinopathy despite favorable systemic response to coenzyme Q10 in COQ2 deficiency
P-193	Benzoni	Chiara	Late-onset CblC deficiency: a single-center case series of a treatable condition

P-194	Bernardino Cuesta	Beatriz	Presymptomatic FLAD1 and MADD Detected by Newborn Screening: Three Cases with Favorable Outcomes
P-195	Brennerova	Katarina	Thiamine metabolism dysfunction syndrome 1 (TMDS1) manifested during Parvovirus B19 infection
P-196	Çiki	Kismet	Molybdenum cofactor deficiency : a single-center experience
P-197	Demirbaş Yülüklü	Göksu	High ALP Levels, Periosteal Reactions, and Seizures: The Hidden Face of Menkes Disease
P-198	Furlan	Francesca	Motor neuropathy presenting with pseudo-ataxia in biotinidase deficiency: a diagnosis not to miss
P-199	Tagliaferri	Francesco	Current management of biotinidase deficiency following newborn screening in Italy:evidence from a clinical nationwide survey
P-200	Furlan	Francesca	Vitamin-B12 status in a cohort of mother-infant dyad: an Italian single center experience
P-201	Imbard	Apolline	Severe Hyperhomocysteinemia in Adults: Expanding the Differential Beyond Inborn Errors of Metabolism
P-202	Kilic Yildirim	Gonca	Biotinidase Deficiency in Central Anatolia: Clinical and Molecular Spectrum of 161 Patients
P-203	Köşeci	Burcu	COASY Mutation Identified by Elevated C0 in Acylcarnitine Profile: A Case Report
P-204	Majid	Hafsa	Characterizing BTD Gene Mutations and Clinical Phenotypes in Pakistani Biotinidase Deficiency Patients
P-205	Mathur	Priyanshu	Missed Opportunities in Treatable Transcobalamin II Deficiency Diagnosed After Two Previously Affected Sibling Deaths
P-206	Matthys	Imke	Case Report: Late-Onset Cobalamin C Deficiency in a 16-Year-Old Patient
P-207	Mohamed	Sarar	PGAP3-Related Hyperphosphatasia Syndrome in a Child with Developmental Delay and Epilepsy
P-208	Mohamed	Sarar	Genetic Counseling Complexity in a Family With Hypomagnesemia, Joubert Syndrome, and Down Syndrome Diagnoses
P-209	Pálmay	Andrea	Infantile presentation of FLAD1-associated MADD (case report)
P-210	Parker	Madeline	Case report: treatment of GEMIN5-related neurodevelopmental disorder with coenzyme Q10 supplementation
P-211	Pavlidou	Despina Christina	Retrophenotyping: Unexpected diagnosis of molybdenum cofactor deficiency type A in an adult
P-212	Feillet	François	Occipital horn syndrome phenotype with normal ATP7A sequencing and a cosegregating pathogenic SLC2A10 variant
P-213	Stanovský	Samuel	When vitamin B12 cannot reach cells: a case report of transcobalamin deficiency in an infant
P-214	Tummolo	Albina	PKU Biochemical Markers: Interplay of Dietary Compliance, B-Vitamins, and Homocysteine levels in PKU children.
P-215	Kose	Engin	Untargeted Metabolomic and Lipidomic Profiling Reveals Distinct Biochemical Patterns in Treated Biotinidase Deficiency
P-216	Doederlein Schwartz	Ida Vanessa	Next generation analysis of ATP7B gene and Wilson Disease: a genotype-phenotype study
P-217	Vorster	Chris	Transient riboflavin transporter deficiency 1 in a Black African neonate: an Nngwe case report
09.Genetic therapies including RNA-based therapies	Submitter name		Abstract title
P-218	Boffa	Iolanda	Periportal OAT expression via AAV does not alter ureagenesis, supporting gene therapy safety for GACR
P-219	Cardamone	Maria Dafne	Stabilizing Glutamine Synthetase via hepatic A-to-I RNA editing reduces hyperammonemia in multiple Urea Cycle Disorders
P-220	Garanto	Alex	Towards an n-of-1 personalized antisense oligonucleotide treatment for Stargardt disease
P-221	Goker-Alpan	Ozlem	Disease Biology Drives Long-Term Immune Responses After AAV Gene Transfer Therapy
P-222	Kaufman	Christina	Preclinical development of an antisense oligonucleotide treatment for complex II deficiency
P-223	Older	Jonna	Genetic and metabolic correction of propionic acidaemia
P-224	Kreher	Nerissa	tRNA medicines for Stop Codon Disease: A platform enabling therapeutic development across rare genetic disorders
P-225	Vondran	Andrej	Treatment of rare metabolic disorders by homolog activation through gene editing
10.Glycosylation disorders/CDG, protein modification disorders	Submitter name		Abstract title
P-226	Akyüzlüer Güneş	Setenay Merve	First Report of a Successful Pregnancy in a Woman with PGM1-CDG Under Galactose Treatment
P-227	Amartino	Hernan	Prenatal diagnosis of SLC39A8-CDG with cortical malformation and manganese-related electrolyte disturbances
P-228	Taverna	Elena	Cohen syndrome organoids uncover a functional lysosome–cilia axis in human brain development
P-229	Bijarnia-Mahay	Sunita	Inherited disorders of GPI-anchor proteins – a clinico-molecular spectrum from India
P-230	Blanita	Daniela	NMR Spectroscopy as a Tool for the Differential Diagnosis of Suspected Congenital Disorders of Glycosylation
P-231	Holubová	Veronika	Mortality in Patients with Phosphomannomutase 2-congenital disorder of glycosylation (PMM2-CDG)
P-232	Kasapkara	Çiğdem Seher	Congenital Disorder of Deglycosylation 2: Report of a Novel MAN2C1 Pathogenic Variant
P-233	Kraoua	Ichraf	Phenotype and genotype Analysis of PMM2-CDG in a Tunisian Cohort
P-234	Laakso	Saila	SLC35A3-glycosylation disorder causes significant brain abnormalities in a newborn
P-235	McSweeney	Mel	Understanding ataxia and gross motor function in PMM2-CDG: Insights from patients, caregivers, and clinicians
P-236	Õunap	Katrin	A case series study: the treatment of five PMM2-CDG patients with epalrestat
P-237	Öztürk-Hişmi	Burcu	Clinical and Molecular Spectrum of NGLY1 Deficiency: A Multicenter Cohort
P-238	Peñafiel-Freire	Diego Mauricio	Cases of pediatric congenital disorders of glycosylation in Navarra (Spain) during 2020-2025
P-239	Sinha	Pallavi	DPAGT1-Related Disorders: The Expanded Phenotypic Spectrum
P-240	Tan	Chern Yan	Expanding the Phenotypic Spectrum of PIGB Deficiency: Genotype–Outcome Associations and Under-Recognised Respiratory Morbidity

11.Innovative and new therapies including repurposing of medicines and regenerative medicine but excluding genetic therapies	Submitter name		Abstract title
P-241	Altun	Akif	Virtual screening of Alpha-Aminoadipate Aminotransferase inhibitors from FDA approved drugs for Glutaric Acidemia Type I
P-242	Demirbaş Yülüklü	Göksu	Intravenous Alpha-Lipoic Acid in Rare Metabolic Neuropathies: A Three-Case Clinical Experience
P-243	Pontoizeau	Clément	Muscle-targeted and liver detargeted AAV gene therapy provides sustained correction of MSUD in neonatal mice
P-244	Lundqvist	Thomas	Low-dose metformin in malonyl-CoA decarboxylase deficiency: cardiomyopathy regression, enhanced fasting tolerance, and neurometabolic improvement
P-245	Lundqvist	Thomas	Metformin reinforces ketogenic metabolism in pyruvate dehydrogenase deficiency: two adolescent cases
P-246	Marinova	Cecilia	Can we optimize the diagnostic pathway for Allan–Herndon–Dudley Syndrome (AHDS) based on specialist views?
P-247	Radzikowski	Michał	Antioxidant and antiglycation potential of α-tocopherol and β-tocopherol: in silico and in vitro analyses
P-248	van Karnebeek	Clara	Partnering with our Patients to Repurpose Drugs: Glycerol Phenylbutyrate Effective in Children with SLC6A1-Neurodevelopmental Disorder
12.Lysosomal disorders	Submitter name		Abstract title
P-249	Abilkassem	Rachid	Clinical and genetic characteristics of 60 Moroccan children's patients with Lysosomal storage diseases.
P-250	Adang	Laura	Characterization of gross regression in Metachromatic leukodystrophy (MLD) across the lifespan
P-251	Akiyama	Takeyuki	A patient group survey on patient-reported outcomes and treatment practices in MPS
P-252	Akyüzlüer Güneş	Setenay Merve	Hematopoietic Stem Cell Transplantation In Inherited Metabolic Diseases A Single-Center Experience
P-253	Al Jasmi	Fatma	Beyond binary: frequency-based symptoms for Fabry disease detection using machine learning algorithms
P-254	Vlad	Raluca Maria	Treat early and Hunter Syndrome will stop being a debilitating disease
P-255	Basan	Hacer	Diverse Clinical Spectrum of Niemann-Pick C: Insights from a Single Center
P-256	Basan	Hacer	Orofacial, Chewing, and Swallowing Dysfunction in Lysosomal Storage Diseases: Clinical Assessment and Feeding Management
P-257	Basan	Hacer	Variable Clinical Expression of Acid Sphingomyelinase Deficiency in Patients with the Same Genotype
P-258	Kraoua	Ichraf	Phenotypic Variability and Genotypic Spectrum of Neuronal Ceroid Lipofuscinosis
P-259	Benitez Provedo	Cristina	Clinical experience in three CLN2 patients: enzyme replacement effects and strategies after treatment discontinuation
P-260	Böttcher	Tobias	Higher diagnostic yield in suspected Lysosomal storage disorders by multiomic approach – clinical examples
P-261	Fischer	Steffen	Is an age- and sex-specific cut-off required for diagnosing Gaucher disease using lyso-Gb1 blood levels?
P-262	Burgaç	Ezgi	Unusual Infantile Presentation of MPS IIIA with Seizures: Early Diagnosis and a Novel Variant
P-263	Chapman	Kimberly	A Single-Year, Single-Institution Experience of Newborn Screening for MPS II
P-265	Chen	Yun-Ru	Adenine Base Editing Corrects GLA IVS4+919G>A in Fabry Disease Models
P-266	Doederlein Schwartz	Ida Vanessa	Clinical and Diagnostic Features of Type 1 Gaucher Disease in the Elderly: A Systematic Review
P-267	Doederlein Schwartz	Ida Vanessa	Safety and Efficacy of Migalastat in Fabry Disease: A Systematic Review and Meta-Analysis
P-268	Vlad	Raluca Maria	The way they walk - neurological involvement in acid sphingomyelinase deficiency
P-269	Ergin	Filiz Basak	Marked Allelic Heterogeneity in Sandhoff and Tay Sachs Disease: A Unique Variant Spectrum in a 16-Patient Cohort
P-270	Espitia Segura	Oscar Mauricio	Neurodegeneration biomarkers in CLN2: CSF neurofilament light chain levels in patients treated with cerliponase alfa
P-271	Eyskens	Francois	Population-based reference interval determination for GlcSph, Lyso-Gb3 and Lyso-SM on dried blood spots.
P-272	Ezgü	Fatih S	Phase 3 study design: evaluation of efficacy and safety of DNL126 in mucopolysaccharidosis IIIA
P-273	Faraguna	Martha Caterina	Comparing plasma and leukocyte alpha-glucosidase activity after alglucosidase and avalglucosidase infusion in classic-infantile Pompe disease
P-274	Faraguna	Martha Caterina	Central Nervous System Histopathology in Classic Infantile Pompe Disease: A Systematic Review with Clinical Relevance
P-275	Felipe	Ana	From Childhood to Adulthood: Descriptive Analysis of Clinical Features in Patients with Acid Sphingomyelinase Deficiency
P-276	Fiori	Laura	Olipudase Alfa for Acid Sphingomyelinase Deficiency: evidence of efficacy from the Italian Compassionate Use Program
P-277	Fujita	Yoshimi	Reduced cerebrovascular events after switching to agalsidase beta in Fabry disease: two case reports
P-278	Gambardella	Jessica	Pegunigalsidase alfa improves exercise tolerance and skeletal muscle bioenergetics in a Fabry disease mouse model
P-279	Girard	Aline	Human iPSC-derived models to unravel the role of glypicans in synaptic alterations in Sanfilippo syndrome
P-280	Giugliani	Roberto	Real-world safety and effectiveness of idursulfase treatment in patients with mucopolysaccharidosis II aged <16 months
P-281	Giugliani	Roberto	Updated Report on the Relative Frequency of Mucopolysaccharidoses in Brazil (2004-2025)
P-282	Goker-Alpan	Ozlem	Long-term safety and efficacy of pegunigalsidase alfa in treatment-naive Fabry disease patients in F60/BRILLIANCE trial
P-283	Gragnaniello	Vincenza	Early inflammatory and autophagic changes in presymptomatic Fabry disease: insights from newborn screening
P-284	Guffon-Fouilhoux	Nathalie	Detection of alpha-mannosidosis: a phenotypic similarity approach powered by natural language processing
P-285	Guffon-Fouilhoux	Nathalie	Neurological and functional assessment of 10 French MPS IIIC patients from the natural history study
P-286	Guler	Elif	Beyond the classic phenotype: Clinical and genetic insights from eight cases of Infantile Sandhoff Disease
P-287	Guler	Elif	The Complex Anatomy of GM1 Gangliosidosis: Natural Progression, Lyso-GM1, and Neurocytolysis Indicators
P-288	Hang	Alex	The new frontiers: New South Wales' experiences in the diagnosis of the mucopolysaccharidoses

P-289	Hastings	Caroline	Importance of Mode of Administration and Therapeutic Index on Intravenous HPBCD in Patients with NPC
P-290	Hastings	Caroline	Subgroup analyses of US EAP participants with Niemann-Pick disease type C treated with arimoclomol
P-291	Hastings	Caroline	Trial Update: Integrated Safety Summary from TRANSPORT-NPC, Evaluating Intravenous HPβCD in patients with NPC Disease
P-292	Hennermann	Julia B.	Effectiveness of idursulfase therapy in young children with mucopolysaccharidosis II: an international retrospective chart review
P-293	Hennermann	Julia B.	A predictive algorithm for the detection of Fabry disease: retrospective real-world database analysis from Germany
P-294	Jalazo	Elizabeth	Preliminary results from a Phase 1/2, first-in-human, open-label study of DNL126 in children with MPSIIIA
P-295	Ko	Jung Min	Early versus Late Enzyme Replacement Therapy for Korean Siblings with Morquio A: a Longitudinal Study
P-296	Kobayashi	Masahisa	In vitro Amenability to Migalastat does not always correlate with Clinical Efficacy in Fabry Disease
P-297	Kor	Deniz	From Infantile to Late-Onset Forms: Is Enzyme Therapy Rewriting the Natural Course of Pompe Disease?
P-298	Korkmaz Katilmiş	Billur	Clinical Evaluation of Hematopoietic Stem Cell Transplantation in Mucopolysaccharidosis: A Single-Center Experience
P-299	Laney	Dawn	The reality of untreated Fabry disease: Patient-reported challenges from a 238-participant cross-sectional survey
P-300	Laney	Dawn	Responsiveness of Early Signs and Symptoms of Fabry Disease in Agalsidase Beta-Treated US Pediatric Patients
P-301	Lazaridis	Christos	Sleep-study severity as a predictor of intraoperative airway difficulty in Mucopolysaccharidosis patients: a single-centre experience.
P-302	Lenzini	Livia	Circulating Extracellular Vesicles Contribute to Endothelial and Cardiac Dysfunction in Anderson–Fabry Disease
P-303	Lourenço	Charles	Double the trouble": Dual genetic diagnosis in Fabry disease may not be so uncommon
P-304	Lu	Yung-Hsiu	Newborn Screening–Enabled Therapeutic Window in Late-Onset Fabry Disease: Long-Term Divergent Remodeling Trajectories With Early-Stage Treatment
P-305	Makrygianni	Maria	Sertraline treatment can mimic Niemann-Pick type C biomarker profile:a diagnostic pitfall with potential clinical consequences
P-306	Giugliani	Roberto	Improved outcome with early introduction of ERT in MPS VI: a sibling study in Brazil
P-307	Mengel	Karl Eugen	Easily overlooked: Niemann–Pick disease type C2 with attenuated phenotype in patients homozygous for c.358C>T
P-308	Minea	Carmen	Bisphosphonates in Managing Bone Involvement in Gaucher Disease: Real-World Evidence Across Different Multiple Skeletal Sites
P-309	Mitchell	John	Phase 1/2 study of intravenous tividenofusp alfa for mucopolysaccharidosis type II: an analysis by genotype
P-310	Monastiri	Kamel	Niemann Pick Type C in Newborns: Four Case Reports
P-311	Muenzer	Joseph	Phase 1/2 study of intravenous tividenofusp alfa for MPS II: an analysis by ERT status
P-312	Murko	Simona	Alpha-Mannosidosis: Biomarker-based Treatment Monitoring
P-313	Murphy	Elaine	Impact on biochemical markers following reduced dose olipudase alfa in adults with acid sphingomyelinase deficiency
P-314	Muschol	Nicole	Clinical impact of MAN2B1 c.2248C>T variant in patients with alpha-mannosidosis: genotype–phenotype insights from SPARKLE registry
P-315	Nakamura	Katsuya	A Pilot Study of Quantitative Sudomotor Function Testing in Patients with Fabry Disease
P-316	Namdar	Mehdi	Cardiac Arrhythmias and Conduction Blocks in Fabry Disease: Early Manifestations That Precede Left Ventricular Hypertrophy
P-317	Newman	Stephanie	AAV9 ARSA Therapy Reverses CNS and PNS Deficits in Post Symptomatic MLD
P-318	Nguyen	Thi Thanh Huong	Genotype-phenotype correlation and CRIM status in Vietnamese children with Pompe disease: a single-center experience
P-319	Nordbeck	Peter	followME Fabry Pathfinders registry: 5-year cardiac, renal and multisystem effectiveness in patients on migalastat
P-320	Ochoa-Ferraro	Antonio	Enhancing Enzyme Replacement Therapy in Lysosomal Storage Disorders: Faster Infusion Rates Across the UK
P-321	Onenli Mungan	H Neslihan	Neurological Manifestations in Infantile Pompe Disease Other Than Muscle Involvement
P-322	Pajares Garcia	Sonia	Juvenile and adult onset of GM2 Gangliosidosis AB variant: two novel cases reported in Spain
P-323	Park	Sanghyun	10-Year Clinical Course of a Patient with Fabry Cardiomyopathy and Severe Left Ventricular Hypertrophy
P-324	Patsora	Maryna	Mild clinical presentation and discordant biochemical findings in siblings with MPS II
P-325	Perretta	Fernando	Switching to migalastat in Fabry disease: clinical evidence and Argentine real-world experience
P-326	Pettazzoni	Magali	Improving lysosomal storage disorder diagnosis using multiplex DBS-MS/MS: 7-Year French reference center experience
P-327	Pettazzoni	Magali	Clinical and Biochemical Characterization of the GLA p.I198T Variant in Seven Families: Fabry or Not?
P-328	Plaiasu	Vasilica	Unmasking alpha-mannosidosis: key clinical lessons from two unrelated families
P-329	Ramaswami	Uma	Subdomain Analysis of the NPC Clinical Severity Scale Evaluating Long-Term Efficacy of Levacetylleucine in NPC
P-330	Ramaswami	Uma	Long-Term Efficacy and Safety of Levacetylleucine in Adults and Children with Niemann-Pick Disease Type C
P-331	Ramaswami	Uma	Measuring Functional Abilities and Assistance Level in People with Niemann-Pick Disease Type C Receiving Levacetylleucine
P-332	Braun	Fabian	Investigation of organ immune interactions in Fabry disease through CITE-Seq and PBMC-organoid-coculture
P-333	Scherer	Thomas	Baseline disease burden and real-world monitoring practices: global insights from the followME Pompe Journey registry
P-334	Schlotawa	Lars	Purvalanol A identified by in silico screening as treatment option for Multiple Sulfatase Deficiency
P-335	Sechi	Annalisa	Management of skeletal complications in adults with Gaucher Disease: results of an Italian multicentre survey.
P-336	Seker Yilmaz	Berna	Diagnostic Delay in Metachromatic Leukodystrophy: Real-World Evidence Supporting Newborn Screening in the Gene Therapy Era
P-337	Seo	Jiwon	Changes in Cardiopulmonary Exercise Performance During Enzyme Replacement Therapy in Fabry Disease.
P-338	Şeren	Nazlıcan	Evidence of Mitophagy Activation in MPS III: Increased LC-3 and PINK-1 in Leukocytes
P-339	Rodrigues	Esmeralda	Infantile onset pompe disease - still a challenge
P-340	Shimada	Yohta	Therapeutic potential of ex vivo T-cell gene therapy using modified enzyme in GM1 gangliosidosis

P-341	Sredzinska	Malgorzata	Schindler disease coused by rare variant in the NAGA gene
P-342	Streubel	Berthold	Integrating α-mannosidosis into the differential diagnostic algorithm for suspected MPS: A 30-month update
P-343	Sudo	Yuta	Newborn screening for Fabry disease detects milder phenotypes of GLA dysfunction: lessons from Aichi, Japan
P-344	Sürücü Kara	İlknur	Urinary System Involvement and Voiding Dysfunction in Patients with Mucopolysaccharidosis
P-345	Sürücü Kara	İlknur	Clinical Characteristics and Diagnostic Journey of Patients with Alpha-Mannosidosis
P-346	Tagi	Veronica Maria	Fat Mass deficiency evidence in an ASMD patient: implications for nutritional management and follow-up
P-347	Tebani	Abdellah	Exploring metabolic dynamics in Gaucher disease type 1 through multi-omics lenses
P-348	To	Hiroaki	Umbilical cord blood transplantation for mucolipidosis III: clinical stabilization and urinary heparan sulfate reduction
P-349	Tonin	Rodolfo	Secondary GM1 ganglioside accumulation across lysosomal storage and neurological disorders: a comparative human cell-based study
P-350	Tüzel Gündüz	Nazmiye	Galactosialidosis: Clinical Variability Even in the Same Family with Three Patients
P-351	Ugena Garcia	Alicia	Real-world multicenter experience with four-week pegunigalsidase-alfa dosing in Fabry disease
P-352	Serrano Gonzalo	Irene	Genotype-specific skeletal phenotypes in Gaucher disease type 1: insights from the Spanish registry
P-353	Vijay	Suresh	Enzyme replacement therapy and stem cell transplantation in 9 patients with lysosomal acid lipase deficiency
P-354	Vukić	Vana	Three-year experience with olipudase alfa therapy in acid sphingomyelinase deficiency in pediatric patients in Croatia
P-355	Wijnen	Mark	Experience with teriparatide in two patients with mucopolysaccharidosis type IVB
P-356	Yazici	Havva	Beyond Premedication: Desensitisation and Omalizumab for Severe ERT Hypersensitivity in Pediatric LSDs
P-357	Yildirim Sozmen	Eser	Leukocytes from mucopolysaccharidosis patients exhibit signs of mitochondrial dysfunction.
P-358	Yoldaş Çelik	Merve	Diagnostic Challenges in Dual Monogenic Renal Disease: Cystinosis Masked by Congenital Nephrotic Syndrome
P-359	Zakharova	Ekaterina	Routine biochemical diagnostic index for Lysosomal acid lipase deficiency and Niemann–Pick disease type A/B, C
P-360	Zarante Bahamon	Ana Maria	Wolman Disease with Homozygous LIPA p.Arg218Ter: One-Year Follow-Up After Early Sebelipase Alfa
P-361	Žigman	Tamara	Clinical stabilisation in two patients with infantile-onset Pompe disease treated with cipaglucosidase alfa and miglustat
P-361A	Hopkin	Robert	Tolerability of 1 Hour Infusions of Pegunigalsidase Alfa in Fabry Disease: Data from the F60/BRILLIANCE Trial
P-361B	Hopkin	Robert	Long-term safety and efficacy of pegunigalsidase alfa in patients who switched from agalsidase alfa
13.Metabolic liver diseases	Submitter name		Abstract title
P-362	Karalar Pekuz	Özge Kamer	Assessment of hepatic fibrosis in patients with Gaucher and Niemann-Pick Disease
P-363	Kilavuz	Sebile	A Novel Hepatic Phenotype in CYP4V2 Deficiency: Bietti Crystalline Dystrophy as an Inherited Metabolic Disorder
P-364	Rombaut	Matthias	Unraveling the transcriptomic and metabolic signature of alkaptonuria through an in vitro liver disease model
P-365	Şahin Uyar	Sibel Burçak	Rare Metabolic Diseases Diagnosed with a Cholestasis Presentation in the Neonatal and Early Infantile Period
14.Metabolic myopathies	Submitter name		Abstract title
P-366	Grafakou	Olga	A case of GGPS1 associated congenital muscular dystrophy with an unusual phenotype
P-367	Selamioğlu	Arzu	When exercise intolerance meets hemolysis: three cases of glycogen storage disease type VII
15.Metabolomics and other omics	Submitter name		Abstract title
P-368	Bianco	Sabrina	Membrane remodeling drives metabolic dysregulation in methylmalonic acidemia
P-369	Cicalini	Ilaria	Uncovering novel biomarkers of gaucher disease through untargeted metabolomics applied to dried blood spots
P-370	Faneca	Joana	Quality evaluation of untargeted urine metabolomics as a first-line test for inherited metabolic disorders
P-371	Furuta	Yutaka	Genome sequencing enables detection of rare non-coding variants in inborn errors of metabolism
P-372	Griffiths	William	Lipidomics analysis- a novel approach to identify patients with Lysosomal Storage Disease- preliminary results.
P-373	Körver-Keularts	Irene	Metabolomics in blood cells for improved diagnostics and biochemical follow-up of inherited metabolic disorders (MELAS,PDE,galactosemia,GNE)
P-374	Laguna Moreno	Javier	SWATH acquisition enhances the identification of metabolites associated with inherited metabolic disorders by UHPLC-QTOF-MS
P-375	Radenkovic	Silvia	Improving diagnosis of HMGCS2 deficiency through untargeted plasma metabolomics
P-376	Stocker	Kim Lucien	Leveraging control cohorts in metabolomics to overcome sample size limitations in detecting inherited metabolic disorders
P-377	Tiivoja	Elis	Implementation of an untargeted metabolomics workflow into clinical practice in Estonia
P-378	Vieira Neto	Eduardo	Dissecting mitochondrial trifunctional protein functions: dissociation between cardiolipin remodeling and mitochondrial bioenergetics
16.Mitochondrial disorders	Submitter name		Abstract title
P-379	Ardissone	Anna	Mitochondrial leukodystrophies riboflavin responsive: two new cases and a review of the literature
P-380	Aghakıshılı	Hanım	Brain MRI phenotypes in mitochondrial diseases: A genotype-neuroimaging correlation study
P-381	Aihara	Hisato	Renal Involvement in RRM2B-related Mitochondrial DNA Depletion Syndrome: Review in Japan
P-382	Alirzayeva	Leman	TEFM Gene Mutation Mimicking Neurotransmitter Disorders: A Challenging Case of Mitochondrial Dysfunction with Complex Neurological Presentation
P-383	Aljaberi	Rana	Combined oxidative phosphorylation deficiency 30 (COXPD30): a case series expanding the clinical spectrum
P-384	Alpdogan	Sedef	Mitochondrial Neurogastrointestinal Evaluation of Endocrine Functions in Encephalomyopathy: A Single-Centre Cohort Study
P-385	Alpdogan	Sedef	Early-onset ataxia , optic atrophy ,peripheral neuropathy Homozygous SLC25A46 Variant: Case Report
P-386	Amartino	Hernan	AIFM1-related disorder mimicking atypical spinal muscular atrophy with skeletal dysplasia: expanding the phenotypic spectrum

P-387	Amartino	Hernan	Neonatal fatal TRMU deficiency mimicking pyruvate dehydrogenase deficiency with prenatal brain anomalies
P-388	Boudabous	Hela	Clinical and Genetic Characterisation of Eight Paediatric Cases with Dihydrolipoamide Dehydrogenase Deficiency in Tunisia
P-389	Demirbaş Yülüklü	Göksu	Two Rare Cases: Mitochondrial DNA Depletion Syndrome in Siblings with a Novel Pathogenic SLC25A4 Variant
P-390	Dwinanda	Novitria	Mitochondrial Disease as a Master of Disguise: Diagnostic Pitfalls in Pyruvate Dehydrogenase E1-Alpha Deficiency
P-391	Gahlot	Arushi	Expanding the Spectrum of ECHS1 Deficiency with movement disorder, Novel and Recurrent c.476A>G Hotspot Variants
P-392	German	Hannah	MDH1 facilitates transsulfuration and affects cellular methylation capacity via redox modulation
P-393	Gort	Laura	Identifying new diseases by using MitoCarta3.0 to analyse data of patients with suspected mitochondrial disease
P-394	Gülbahçe	Aliye	Pyruvate Dehydrogenase Complex Deficiency: Clinical and Biochemical Insights
P-395	Saada (Reisch)	Ann	Mitochondrial Derived Vesicles- (MDVs), capsules of information and energy
P-396	Heath	Oliver	Reverse phenotyping after exome sequencing in mitochondrial disease diagnosis – are tissue biopsies still needed?
P-398	Keshavan	Nandaki	Prospective UK-wide Natural History of Primary Pyruvate Dehydrogenase Deficiency
P-399	Kondo	Hidehito	Neonatal Mitochondrial Disease with Pontocerebellar Hypoplasia and Hypertrophic Cardiomyopathy from Combined ATAD3A/B and 1p36 Deletions
P-400	Kraoua	Ichraf	Neuromuscular involvement in POLG-related MNGIE syndrome: a descriptive study of a Tunisian paediatric series
P-401	Basan	Hacer	Aberrant Splicing from SLC25A3 c.158-9A>G Variant Reveals Tissue-Specific Effects in Mitochondrial Phosphate Carrier Deficiency
P-402	Malekkou	Anna	Expanding the phenotypic spectrum of MRPS2-related disease: Mild clinical presentation and a novel variant
P-403	van der Meijden	Chris	Clinical, molecular, and cardiac characterization of four rare mtDNA tRNA variants in pediatric mitochondrial disease
P-404	Mohamed	Sarar	ISCA2-Related Multiple Mitochondrial Dysfunction Syndromes Type 4 With Atypical Features: Expanding The Phenotype
P-405	Nevalainen	Pasi	A case of multisystem neurological disease associated with a variant in the AFG3L2 ATPase domain
P-406	Noyan	Bilge	C12ORF65-Related Mitochondrial Disease in Dizygotic Twins Presenting with Optic Atrophy and Neuropathy
P-407	Ptackova	Hana	Severe, Non-HUPRA Neurological Phenotype as a Novel Manifestation of SARS2 Deficiency
P-408	Saligova	Jana	Intrafamilial Variability in EARS2 Deficiency: A Case Study of Two Sisters
P-409	Schiff	Manuel	National cohort of patients with TTC19 mutation: Clinical, genetic, and outcome characteristics: a neuroradiological signature?
P-410	Sofou	Kalliopi	Stroke-like episodes in mitochondrial diseases: genetic causes and outcomes
P-411	Tan	Chern Yan	Divergent Respiratory Trajectories in Paediatric Mitochondrial Disease: A Case Series of SCO2 and IBA57 Mutations
P-412	Torregrosa	Ruben	Energetic compensation at a cost: hypermetabolism in human motor neurons with ATP synthase deficiency
P-413	Tsiakas	Konstantinos	De-novo MORC2-variants in two patients with severe neurologic disorder: pleiotropic effects of an epigenetic signature
P-414	van Alkemade	Gijs	Developmental delay, hearing loss, and parieto-occipital white matter lesions due to a heterozygous LONP1 variant
P-415	Lam	Amanda	Implementing Targeted Proteomics for VUS evaluation in the UK Healthcare system for Primary Mitochondrial Disease.
P-416	Yano	Shoji	Elevated cerebrospinal fluid 2-Hydroxybutyric acid in two siblings with aspartate-glutamate carrier 1 deficiency
P-417	Yüksel Yanbolu	Ayşe	Expanding the Phenotypic Spectrum of COX10-Related Mitochondrial Disease: A Comprehensive Multi-Systemic Evaluation of Two Siblings
P-418	Zweers	Heidi	The effect of low-carbohydrate high-fat diet on the metabolome of adult patients with m.3243A>G mutation
P-419	Zweers	Heidi	The influence of a Low-carbohydrate high-fat diet on symptoms of adult m.3243A>G patients
P-420	Zweers	Heidi	Gut microbiota dysbiosis in people with mitochondrial disease caused by the m.3243A>G mutation
17.Neurotransmitter and creatine related disorders	Submitter name		Abstract title
P-421	Alhashem	Amal	Expanding the Phenotype of GCH1-Related Disorders:A Founder Synonymous Variant with Developmental Regression and Levodopa Response
P-422	Ateş	Zülfükar Ege	Expanding the phenotypic spectrum of GLYT1 encephalopathy: A case report and literature review
P-423	Centofanti	Cristiana	Slc6a8Y389C knock-in rat model of creatine transporter deficiency shows cardiac phenotype changes and motor dysfunction
P-424	Márquez Mesa	Elena	GAMT Deficiency: A Rare but Treatable Cause of Refractory Epilepsy
P-425	Pisani	Francesco	Gene therapy in an adult patient with AADC deficiency
P-426	Mercimek-Andrews	Saadet	Outcomes of betaine therapy in two males with creatine transporter deficiency
P-427	De Giorgi	Agnese	Transient neurodevelopmental alteration and spastic paraplegia in TH deficiency
P-428	Pope	Simon	Measurement of kynurenine metabolites in cerebrospinal fluid (CSF) and correlation with monoamine and pterin metabolites
P-429	Ricciardi	Giacomina	Primary Disturbances of the Synthesis of Monoamine Neurotransmitters: Impact on the Noradrenergic System
P-430	Bhattacharya	Kaustuv	Careful Consideration of the Use of Dextrothomorphan in Attenuated Non-Ketotic Hyperglycinemia
P-431	Williams	Monique	GAMT Deficiency: Sodium Benzoate Treatment Withdrawal in Two Paediatric Cases
P-432	Yousefimonfared	Anita	Dopa-Responsive Dystonia Associated with Heterozygous AADC Variant, Cys438Tyr: Recombinant Variants and In vitro DDC-knockout Cell Models
18.New Diseases	Submitter name		Abstract title
P-433	Genc	Hamide Sevinc	Uncovering a Rare Metabolic Culprit: Early-Onset Gout Associated with LDHD Mutation
P-434	Koç Yekedüz	Merve	Multimodal Non-Invasive Biomarker Characterization of Structural and Functional Alterations in ADSS1 Myopathy
P-435	Özgen	Yasemin	Beyond the classic ARC triad: atypical VIPAS39-related disease with multisystem involvement
19.New research methods in IEM	Submitter name		Abstract title

P-436	Drell	Philip	Genetically encoded biosensors method to study inborn errors of redox metabolism
P-437	Léal	Pauline	Ultra-High-Field MRI of Brain Organoids: A New Tool to Understand Metabolic Disorders Affecting Neurodevelopment
P-438	Niu	Dau-Ming	Real-Time AI-Integrated Whole Genome Sequencing for Lifelong Precision Health
P-439	Paci	Sabrina	“Unlocking the Unknown”: Artificial Intelligence in Rare Diseases-The Example of Hereditary Fructose Intolerance-
P-440	Saldaña	Diego Aeljandro	Genome-scale metabolic modeling for biomarker discovery in glycogen storage disease Ia via flux-based gluconeogenic failure
P-441	Singh	Rani	Evaluation of Large Language Models for Optimizing Evidence Extraction in Inherited Metabolic Disorders Nutrition Guideline
20.Newborn screening	Submitter name		Abstract title
P-442	Anadón Ruiz	Aránzazu	Expanding Newborn Screening Beyond Blood: High-Throughput Detection of Urinary Metabolites Using Tandem Mass Spectrometry
P-443	Babalola	Ayoade Desmond	Hidden burden of glucose-6-phosphate dehydrogenase deficiency in Brazil: population-based and systems-informed analysis of neonatal mortality
P-444	Zakharova	Ekaterina	Aromatic L-Amino Acid Decarboxylase Deficiency: Detection by Tandem Mass Spectrometry and Implications for Newborn Screening
P-445	Sanchez Pintos	Paula	Improving diagnostic accuracy in mucopolysaccharidosis I NBS combining multiple lysosomal enzymatic assays with urine GAG
P-446	Colon Mejeras	Cristobal	Untargeted proteomics in dried blood spots identifies candidate biomarkers for cystic fibrosis among IRT-positive newborns
P-447	Cortès-Saladelafont	Elisenda	Low Carnitine in Paediatric Practice: a 10-Year Single-Centre Cohort from Newborn Screening and Paediatric Care
P-448	De Giorgi	Agnese	Hereditary spherocytosis mimicking cobalamin metabolic disorder at newborn screening
P-449	de Sain van der Velden	Monique	CLIR covariate-adjusted intervals and markers significantly improve newborn screening accuracy and reduce false positives
P-450	de Sain van der Velden	Monique	Policy and operational considerations regarding the (non-) adoption of CLIR across European newborn screening programmes
P-451	Dufour	Céline	Advances in hyperhomocysteinemia newborn screening in French-Speaking Belgium
P-452	Fatima	Tabeer	Expanding newborn screening for inborn errors of metabolism: insights from 155 treatable conditions
P-453	Fiori	Laura	Diagnostic Challenges of Abnormal Newborn Screening in Cholestatic Neonates: A Case Series with Congenital Hyperinsulinism
P-454	Funghini	Silvia	Design, Implementation, and Outcomes of Simmesn External Quality Assurance Program in Newborn Screening
P-455	Tagliaferri	Francesco	More Than Carriers? Why Biochemistry Still Matters in the Era of Newborn Genetic Screening
P-456	Glab-Jablonska	Ewa	C5DC/C8 and C5DC/(C8+C10) ratios as discriminative predictors for glutaric acidemia type 1 in newborn screening
P-457	Khneisser	Issam	N-acetyl-tyrosine in DBS: free add on decision tool for IEM newborn screening of AA.
P-458	Kinoshita	Yuya	Newborn Screening for Adenosine Deaminase Deficiency Identifies a Case Missed by SCID Screening
P-459	Lepage	Nathalie	Implementation of newborn screening for X-linked adrenoleukodystrophy in one province of Canada (Ontario).
P-460	Lorrai	Daniele	Immunodeficiency Newborn Screening Program: the First Year of Experience in the Emilia Romagna Region
P-461	Lundbäck	Veroniqa	Implementation of genetic 2nd tier testing in Newborn Screening for Carnitine Uptake Deficiency in Sweden
P-462	Matern	Dietrich	Multiple Sulfatase Deficiency: A seconary target of newborn screening for MPS II and/or MLD?
P-463	Messina	Martina	Impact of Newborn Screening on Diagnosis and Early Management of Isovaleric Acidaemia- single centre study
P-464	Noda	Yusuke	Newborn screening for hypophosphatasia: measuring tissue nonspecific alkaline phosphataseactivity in dried blood spots
P-465	Akyüz	Ayşe	Expanded Pilot Newborn Screening for Lysosomal Storage Diseases: Initial Results of First Pilot Screening from Turkey
P-466	Park	Hyun Kyung	Positive Rates and Confirmatory Outcomes of Lysosomal Storage Disease Newborn Screening in Korea
P-467	Perrone Donnorso	Michela	Behind C5-Acylcarnitine signal in Newborn Screening: False Positive Induced by Clubfoot Soft Cast
P-468	Pons Rodriguez	Montserrat	Maternal vitamin B12 deficiency in early pregnancy and its impact on newborn screening
P-469	Quin	Rebecca	Should familial hypercholesterolaemia be included in genomic newborn screening? Perspectives from affected families
P-470	Rosenberg	Aviva	Gaucher Disease: A Case Study in Geographic Disparities of Newborn Screening in the US
P-471	Ruoppolo	Margherita	Implementation of First-Tier Genomic Screening within the Italian Neonatal Program: Pilot Study of 1021 Newborns
P-472	Sanchez Pintos	Paula	Clinical and biochemical evaluation of the first neonatal screening program for Pompe disease in Spain
P-473	Sawada	Takaaki	Treatment course of three male patients with Fabry disease identified by newborn screening
P-474	Sohn	YoungBae	A registry study for nationwide newborn screening of six lysosomal storage disorders in South Korea
P-475	Tagi	Veronica Maria	Maternal Cobalamin deficiency: dietary patterns, intake awareness, and community-driven prevention. An Italian center analysis
P-476	Tummolo	Albina	Integrating NGS and Tandem-Mass Spectrometry as Co-First Tier Newborn Screening: The Experience of Apulia Region
P-477	Veldman	Abigail	Next-generation newborn screening: feasibility of combined genetic and biochemical testing for 95 inherited metabolic disorders
P-478	Vieira Neto	Eduardo	Newborn screening for phenylketonuria employing phenylalanine and tyrosine quantification by tandem mass spectrometry in Brazil
P-479	Wierenga	Klaas	Evaluating propionylcarnitine (C3) in newborn screening: a statewide analysis from Florida between 2020 and 2025
P-480	Wittenauer	Angela	Lessons Learned from Missed Newborn Screening Diagnoses in Georgia Since 2007
P-481	Zhyvytsia	Yuliia	Biochemical Overlap in Inherited Cobalamin Disorders: Implications for Presymptomatic Interpretation of Newborn Screening Results
21.Novel diagnostic/laboratory methods excluding omics	Submitter name		Abstract title
P-482	de Sain van der Velden	Monique	Dried urine spot (DUS) is demonstrated to be a useful matrix for carnitine excretion
P-483	Dumin	Elena	Implementing In-House Dried Blood Spot 3-O-Methyldopa Quantification for Early Aromatic L-Amino Acid Decarboxylase Deficiency Detection
P-484	Eminoğlu	Tuba	High Diagnostic Yield of Whole-Exome-Sequencing in Pediatric-Onset Neurodevelopmental Disorders Using a Weighted Clinical Selection Model

P-485	Hulshof	Tim	ChatRD: An AI-empowered clinical decision support tool for the identification and diagnosis of rare diseases
P-486	Kahraman	Ayça Burcu	Deep Intronic NPC1 Variants in Niemann–Pick Disease TypeC: A Pediatric Case Report and Systematic Review
P-487	Nguyen	Thi Thanh Huong	Revealing Diagnostic Gaps in Neonatal MSUD: Novel BCKDHB Variants and Limitations of Exome Sequencing
P-488	Ezgü	Fatih S	Rapid screening of lysosomal storage diseases via lysosphingolipid panel: A large at-risk cohort in Türkiye.
P-489	Oliva	Clara	Targeted LC-MS/M panel for the simultaneous quantification of organic acids, amino acids, and acylcarnitines.
P-490	Rodriguez Fraga	Olaia	Implementation of a blood-based diagnostic assay for glucose transporter type1 deficiency syndrome: Early clinical experience
P-491	Rossi	Claudia	Differential diagnosis of sphingolipidoses: a novel multiplex strategy for biomarker quantification in lysosomal storage diseases
P-492	Smith	Kathryn	Dilute, Shoot, and Screen: A Multiplex LC-QTOF-MS Urine Test for Inborn Errors of Metabolism
P-493	Vaz	Frederic	A targeted LC–MS/MS assay for alkylphosphocholine(23:0) enables biochemical diagnosis of Sjögren–Larsson syndrome
22.Nursing and allied healthcare studies in IEM	Submitter name		Abstract title
P-494	Alshehri	Monira	Expanding the Recessive Spectrum of DYNC1H1: A Lethal Prenatal Phenotype with Short-Rib Thoracic Dysplasia
P-495	Amartino	Hernan	Early integration of palliative care in a neurometabolic clinic: a Latin American experience
P-496	Bandeira	Anabela	Palliative care in children with inherited metabolic diseases followed by a Pediatric Palliative Care Team
P-497	Daly	Anne	Community Supported Home Management of MSUD Illness Outperforms Hospital Based Care
P-498	Maillot	François	Geriatric Metabolic Medicine: Expanding the Horizons of Inherited Metabolic Disorders
P-499	Morris	Elizabeth	A single centre experience of unplanned pregnancies in Inherited Metabolic Disorders (IMD) over 10 years
P-500	Sirrs	Sandra	Physician staffing needs for clinics for adults with inherited metabolic diseases
P-501	Sirrs	Sandra	Allied health infrastructure needs for clinics for adults with inherited metabolic diseases
P-502	Spolador	Gustavo	14.6% Coverage: Underrecognized Palliative Needs in Pediatric Metabolic Disorders
23.Organic acidurias	Submitter name		Abstract title
P-503	Alsaleh	Norah	Navigating Complexity:Liver Transplant in a Child with Methylmalonic Acidemia and Two other Coexisting Inherited Disorders
P-504	Altun	Ayşe Nur	3-Methylcrotonyl-CoA Carboxylase Deficiency Associated with MCCC2: A Case Series Contributing to Phenotypic Characterization
P-505	Bianchin Marcuzzo	Manuela	XJB-5-131 improves redox and bioenergetic homeostasis in a 3-hydroxy-3-methylglutaric acidemia rat model and patient fibroblasts
P-506	Caro	Amanda Rocío	When Parents Speak: Linking Clinical Features with Care in Colombian Patients with Propionic/Methylmalonic Acidemia
P-507	Cawtherley	Sarah	At-Risk Sibling Screening in Isovaleric Acidaemia (IVA): Prospective Management and Outcomes
P-508	Engelke	Udo	Novel biomarkers in glutaric aciduria type 1
P-509	Gumusoglu	Irem Ilgin	In vitro modelling and genetic correction of methylmalonic acidaemia
P-510	Güra	Miriam A.	Variant- and cell type-dependent decrease in mitochondrial energy production and dysfunction in methylmalonic aciduria
P-511	Janiec	Agnieszka	Clinical and biochemical outcomes of simultaneous liver-kidney transplantation in pediatric patients with methylmalonic acidemia.
P-512	Leipnitz	Guilhian	3-Methylglutaric acid disrupts redox and bioenergetic homeostasis and impairs neurodevelopment in neonatal rats
P-513	Mytsyk	Nataliia	Elevated C5OH in expanded newborn screening: secondary findings and interpretation challenges
P-514	Selvanathan	Arthavan	The trajectory of renal injury in patients with isolated methylmalonic acidaemia: a retrospective review
P-515	Traversi	Florian	Dissecting the role of gut microbiota in methylmalonic aciduria.
P-516	Verde	Alessandra	Medical management and long-term outcome in classical ethylmalonic encephalopathy: insights from a prolonged follow-up
P-517	Wajner	Moacir	L-carnitine mitigates lipopolysaccharide-induced neuroinflammatory response and neurodegeneration in cerebral cortex of neonatal Gcdh$^{-/-}$ mice
P-518	Wajner	Moacir	Protective role of L-carnitine on neurotoxicity induced by glutaric and quinolinic acids in GCDH-deficient mice
P-519	Yap	Sufin	Carglumic Acid (CGA) supports long-term management of Organic Acidurias: An 18-month analysis of PROTECT study.
24.Peroxisomal, sterol, bile acid, lipid and lipoprotein metabolism	Submitter name		Abstract title
P-520	Adang	Laura	Beta-Propellor Protein-Associated Neurodegeneration (BPAN), a genetic cause of autism spectrum disorder: behavioral and developmental profile
P-521	Altun	Ayşe Nur	The Power of Family Screening in Cerebrotendinous Xanthomatosis: A Case Series Revealing a Founder Effect
P-522	Basan	Hacer	From Classic to Mild: Expanding the Phenotypic Spectrum of OCRL-Related Lowe Syndrome
P-523	Benitez Provedo	Cristina	Intrathecal mesenchymal stem cells in cALD: early potential efficacy signals in patients without therapeutic alternatives
P-524	Kose	Engin	Residual Lipidomic Risk After Atorvastatin Therapy in Children with Heterozygous Familial Hypercholesterolaemia:Prospective Longitudinal Lipidomics Study
P-525	Bulut	Fatma Derya	Subclinical Adrenal Insufficiency in a Patient with Peroxisomal Acyl-CoA Oxidase-1 Deficiency
P-526	Terner-Rosenthal	Jolan	Chenodeoxycholic Acid (CDCA) Improves Biochemical Parameters in Patients with Cerebrotendinous Xanthomatosis (CTX) with Normal Cholestanol
P-527	Dingeo	Giulia	Rethinking CTX diagnosis: a multi-biomarker approach
P-528	Dwinanda	Novitria	Algorithmic Approach to Chronic Encephalopathy: Contrasting Alexander Disease and X-Linked Adrenoleukodystrophy in Limited-Resource Settings
P-529	Haas	Dorothea	Impact of Smith-Lemli-Opitz syndrome on health and daily life: results from a MetabERN family survey
P-530	Hama	Kotaro	LPLAT10-mediated phosphatidylcholine remodeling underlies C26:0-lysophosphatidylcholine production in X-linked adrenoleukodystrophy
P-531	Hart	Claire	Two siblings with Lathosterolosis: Diagnosis, monitoring and unexpected raised cholestanol in one individual

P-532	Kasapkara	Çiğdem Seher	A Pediatric Case of Tangier Disease with Ocular Manifestations
P-533	Kırmızıtaş	Melis	Growth outcomes in children with cerebrotendinous xanthomatosis: a 3-year longitudinal evaluation
P-534	Körbeyli	Hüseyin Kutay	Lipoprotein distribution in severe pediatric hypertriglyceridemia: insights from LDL/non-HDL ratio
P-535	Krol-Lagowska	Anna	Biochemical profiling of lysophosphatidylcholines (LPC) and their ratios in X-ALD Patients and ABCD1 mutation carriers
P-536	Lakshminarayana	Bindhu	Phenotype-dependent oxidative stress dysregulation and lipid peroxidation in X-linked adrenoleukodystrophy
P-537	Mintaş	Nuriye Ece	Primary Hypolipidemias in the Aegean Region of Türkiye: A Multi-Center Series
P-538	Morales Romero	Blai	Prenatal diagnosis of desmosterolosis by amniotic fluid sterol profiling: contribution to genetic variant interpretation
P-539	Myrseth	Marte	Stability study of C26:0-lysophosphatidylcholine in plasma
P-540	Radenkovic	Silvia	Itraconazole induced lanosterol accumulation mimicking genetic disorders of sterol metabolism
P-541	Santer	René	Combining biochemical and genetic screening reveals a high prevalence of familial hypercholesterolemia in South-German children
P-542	Sharma	Yashu	From Blood to Brain: Decoding X-linked adrenoleukodystrophy through Genetics, Biomarkers, and Patient-Derived Brain Organoids
P-543	Wang	Yuqin	SPG5A: Defining Variants of Uncertain Significance
P-544	Yoldaş Çelik	Merve	BAAT-Associated Bile Acid Conjugation Defect Type 1: Clinical and Genetic Findings in Three Patients
P-545	Yoldaş Çelik	Merve	Caregiver Attitudes and Perceived Barriers in Treatment Adherence in Pediatric Familial Hypercholesterolemia
P-546	Yoldaş Çelik	Merve	Sitosterolemia Presenting with Cytopenia and Splenomegaly: A Diagnosis Uncovered by CNV Reanalysis
25.Phenylketonuria	Submitter name		Abstract title
P-547	Ahring	Kirsten	LNAA as treatment for late-diagnosed PKU patients
P-548	AlSharfa	Shirin	The Chaperonopathies: Early Intervention in DNAJC12-Related Hyperphenylalaninemia
P-549	Arnold	Georgianne	Maternal PKU in the Post-MPKUCS Era: Reproductive Outcomes from the PHEFREE Rare-Disease Consortium for Phenylketonuria
P-550	Arnoux	Jean-Baptiste	Real world metabolic control in phenylketonuria in France: results from the national multicentre CLARIPHY FR study
P-551	Bélanger-Quintana	Amaya	Sepiapterin in Phenylketonuria: a Spanish Multicenter Case Series
P-552	Biancalana	Edoardo	Clinical outcomes in an adult with classic phenylketonuria treated with sepiapterin: first Italian real-life experience
P-553	Burlina	Alberto B.	Clinical outcomes of the first patients with phenylketonuria treated with sepiapterin in Italy
P-554	Burlina	Alberto B.	Long-term evaluation of quality of life in patients with Phenylketonuria treated with Pegvaliase
P-555	Campistol Plana	Jaume	Insufficient metabolic control and unmet therapeutic need in patients with phenylketonuria (PKU) in Spain (CLARIPHY-SP)
P-556	Christ	Shawn	Humanistic burden and unmet needs of patients with phenylketonuria (PKU): a US physician-reported survey perspective
P-557	Christ	Shawn	Neurocognitive outcomes in adults with phenylketonuria (PKU): Evidence from the PHEFREE Consortium Longitudinal Study
P-558	Doederlein Schwartz	Ida Vanessa	Barriers related to treatment and quality of life in Brazilian patients with phenylketonuria.
P-559	Doederlein Schwartz	Ida Vanessa	A deep analysis of the 14 day sepiapterin responsiveness test in PKU
P-560	Gregoric	Nadan	Classical phenylketonuria and partial lipodystrophy: an extremely rare and therapeutically challenging co occurrence
P-561	Gyenes	Artúr Szilárd	Peripheral neuropathy in adult patients with phenylketonuria
P-562	Höller	Alexander	Improving patient-provider communication in phenylketonuria: technical feasibility and first results from the GenKom-PKU Study
P-563	Jendo	Sidra	The invisible burden of PKU: a qualitative exploration of the impact of phenylketonuria across borders
P-564	Kania	Michał	Obesity in adults with hyperphenylalaninemia is not associated with genotypic-phenotype value – a single-center report
P-565	Kania	Michał	What distinguishes obesity in adults with phenylketonuria? – a single-centre report
P-566	Kania	Michał	Cardiovascular disease risk factors in adults with hyperphenylalaninemia – a single-center report from Poland
P-567	Kania	Michał	Factors associated with abundant amorphous phosphates in urine in adults with hyperphenylalaninaemia – a single-center-report
P-568	Kania	Michał	Calcium-phosphate metabolism and urinalysis in adults with hyperphenylalaninemia – a single-center report
P-569	Khan	Naureen	Clinical audit to look at compliance with HSE Sapropterin prescribing guidelines in neonates at NCIMD.
P-570	Kopesky	Jessica	Mixing studies with sepiapterin to provide Phenylketonuria (PKU) patients with additional administration options
P-571	MacDonald	Anita	Two-Year Sapropterin Outcomes in Phenylketonuria: Longitudinal Study of Metabolic, Dietary, Psychosocial Effects
P-572	MacDonald	Anita	Protein and Micronutrient Intake After Two Years of Sapropterin Treatment in PKU
P-573	Muntau	Ania C	Efficacy and safety of sepiapterin in individuals with phenylketonuria and baseline phenylalanine levels ≥900 µmol/L
P-574	R. Vargas	Paula	Challenges in PKU Journey: A Survey with Patients and Their Caregivers
P-575	Peters	Heidi	EPIPHENY study design: Phase 3b study of long-term neurocognitive outcomes in sepiapterin-treated children with phenylketonuria
P-576	Peters	Heidi	Dietary liberalization in patients with phenylketonuria and controlled blood phenylalanine levels: APHENITY Extension Study findings
P-577	Pinto	Alex	Gastrointestinal Symptoms Are Common in PKU But Under Recognised in Routine Care
P-578	Pinto	Alex	Point-of-Care Testing in PKU: A New ERA of Blood Phenylalanine Monitoring
P-579	Pinto	Alex	Seven Year Trends in Medication Use Among Individuals with PKU: European Retrospective Audit
P-580	Poloni	Soraia	How is consumption of ultra-processed foods among PKU patients? Data from Brazil and France
P-581	Poloni	Soraia	Decrease in lean body mass in young adults with early-treated PKU: 5 years of follow-up
P-582	Rodrigues	Catarina	Postprandial metabolic responses to a glycomacropeptide-based protein substitute in different meal contexts: Implications for Phenylketonuria

P-583	Sanchez-Valle	Amarilis	Efficacy and safety of pegvaliase in adolescents with phenylketonuria: interim results from PEGASUS part 2
P-584	Singh	Rani	Phenylalanine Reduction, LNAA Profiles, and Fat-Free Mass in Adults with PKU on Pegvaliase
P-585	Ishige	Mika	Management of Anaphylaxis during Pegvaliase Therapy in Adults with Phenylketonuria
P-586	Tummolo	Albina	Safety and acceptability of slow-release-large-neutral amino acids (LNAAs) in PKU patients not adhering to dietary
P-587	Tummolo	Albina	Geogenetic Stratification Correlates with Anthropometric Outcomes in a Mediterranean Cohort with PAH Deficiency
P-588	Tummolo	Albina	VAI and Nutritional Ultrasound to identify early sarcopenic obesity in PKU patients
P-589	Vant Westeinde	Annelies	Cognition and adaptive functioning in PKU – a national cohort study
P-590	Verde	Alessandra	Hidden renal risk in adult PKU: clinical insights from a single Center experience
P-591	Vilaseca-Capel	Adrià	Oxidative and mitochondrial status in long-term PKU patients: age and treatment-dependent profiles
P-592	Vockley	Jerry	Phase 1 SAD/MAD study for AG-181 in healthy subjects and phase 1b design in phenylketonuria
27.Urea cycle disorders	Submitter name		Abstract title
P-593	Arranz-Amo	Jose Antonio	Analytical verification of Nor-NOHA collection tubes for plasma arginine measurement: comparison with conventional K3EDTA tubes
P-594	Asami	Yurika	Patient organisations as co-creators for research: citrin deficiency as a case study
P-595	Bowron	Ann	Ambient ammonia analysis: Does it affect results from hyperammonaemic patients? What is the clinical impact?
P-596	Chan	Lap Kwan	Chronic liver injury as a consequence of citrin deficiency
P-597	Chu	Thi Hong Phuong	Bridging the "energy gap" in NICCD management: The role of MCT in reversing clinical course
P-598	Eisa	Osama	Severe hyperammonaemic crises in adults with urea cycle disorders: an international survey and literature review
P-599	Gropman	Andrea	Balancing Risk and Response: ACTH Therapy for Infantile Spasms in Citrullinemia Type I
P-600	Honto	Miki	Leaky pseudo-exon activation by a deep intronic variant in late-onset carbamoyl phosphate synthetase 1 deficiency
P-601	Iwasaki	Roman	Regulatory and Scientific Requirements for Gene Therapies in Urea Cycle Disorders
P-602	Kara	Esra	Neonatal-Onset Carbamoyl Phosphate Synthetase 1 Deficiency: Favorable Short-Term Outcome Following Early Aggressive Treatment
P-603	Kido	Jun	A Practical qPCR-Based Approach for Newborn Screening of Citrin Deficiency Targeting Common SLC25A13 Variants
P-604	Kido	Jun	Long-term Outcomes of Adult Patients with Citrin Deficiency: A Nationwide Study in Japan
P-605	Kim	Yoo-Mi	Growth and pubertal outcomes in pediatric patients with urea cycle disorders: a single-center experience
P-606	La Rosa	Alessandro	Longitudinal Assessment of Protein Tolerance in Mild Urea Cycle Disorders
P-607	Laemmle	Alexander	Generating Patient-Derived Induced Pluripotent Stem Cells to Study the Pathomechanisms of the c.77G>A OTC Variant
P-608	Leong	Huey Yin	A novel ASS1 deep intronic variant driving late-onset Citrullinemia type 1 in indigenous Iban Malaysians
P-609	Manolikos	Catherine	Heterogeneity in adult ornithine transcarbamylase deficiency: a genotype – phenotype study of 23 patients.
P-610	Mccarron	Eamon	Transition in urea cycle disorders from childhood to adulthood: a UK multi-centre mixed-methods service evaluation
P-611	Nurse	James	Headaches that kill? When to consider hyperammonemia in patients with headache and/or migraine
P-612	Rodrigues Nunes	Marcela	Detecting the silent: newborn screening biomarkers reveal oligosymptomatic urea cycle disorders with uncertain therapeutic implications
P-613	Roubos	George	Hyperammonaemia after bariatric surgery is the result of bacterial overgrowth and acquired urea cycle dysfunction.
P-614	Viau	Krista	Evidence- and consensus-based nutrition management guidelines for urea cycle disorders: recommendations for the reproductive period
P-615	Wagenmakers	Margreet	Clinical outcome in adult patients with argininosuccinic aciduria
P-616	Woodhead	Greg	Slimming Safely: GLP-1 Therapy in a Urea Cycle Disorder
P-617	Yoldaş Çelik	Merve	Arginase Deficiency Presenting with Gait Disturbance: Diagnostic Challenge and Early Outcomes After Liver Transplantation
P-618	Yu	Barbara	Contrasting patient experience with clinical understanding in citrin deficiency