ePOSTERS

01. Amino acid disorders	Submitter name		Abstract title
EP-001	Abilkassem	Rachid	Disorders of Amino Acid Metabolism : About 50 Cases
EP-002	Köşeci	Burcu	A Hypotonic Neonate with Elevated C5: A Rare Case of ACADSB Deficiency
EP-003	Mohamed	Sarar	Novel variant in Nonketotic hyperglycinemia.
EP-004	Prochazková	Dagmar	Alkaptonuria in Moravia (Czech Republic)
EP-005	Ostapyshena	Yuliia	Does Massive Amino Acid Wasting Mimic Hypotonia-Cystinuria Syndrome in Heterozygous 2p21 Deletion?
EP-063	Novosiadla	Zoriana	Uncovering novel diet-disease interactions using untargeted nutrigenomics in Drosophila
03.Carbohydrate disorders, excluding CDG's	Submitter name		Abstract title
EP-006	Belo	Anabela	Extended Release Cornstarch Improves Metabolic Control in a Child With Glycogen Storage Disease Type Ib
EP-007	Mohamed	Sarar	Coexistence of two genetic diseases in an infant: The burden of consanguinity
04.Clinical studies, patient reported outcome measures, real world data	Submitter name		Abstract title
EP-008	Ateş	Zülfükar Ege	Knowledge, awareness, and clinical self-efficacy regarding rare diseases among medical trainees in Türkiye
EP-009	Ben Omran	Tawfeg	Clinical studies, patient reported outcome measures, real world data
EP-010	Bilen	Merve	PLA2G6-associated neurodegeneration (PLAN): a case report with claval hypertrophy as a distinctive neuroimaging finding
EP-011	Teke Kısa	Pelin	β-Ketothiolase Deficiency Presenting with Acute Encephalopathy: A Case Report and Clinical Management
06.Disorders of fatty acid oxidation and ketone body metabolism	Submitter name		Abstract title
EP-012	AlSharfa	Shirin	Atypical CPT1A Deficiency Presenting with Hepatic Encephalopathy Without Hypoglycemia or Hyperammonemia: A Novel Variant
EP-013	Basan	Hacer	Mitochondrial HMG-CoA Synthase Deficiency Due to a Novel Homozygous Frameshift Variant in HMGCS2
EP-014	Bin Hadyan	Maryam	Triheptanoin treatment in severe neonatal-onset CPT II deficiency with a novel mutation: A case report
EP-015	Mohamed	Sarar	When Genetics Doesn't Follow the Rules: VLCAD Deficiency with Uncertain Inheritance
EP-016	Trang	Nguyen Thi Ngoc	Severe ketoacidosis revealing SCOT deficiency in a Vietnamese infant: novel OXCT1 variants
07.Disorders of purines, pyrimidines, nucleic acids and porphyrias	Submitter name		Abstract title
EP-017	Ramón-Gómez	Jorge Luis	Unmasking cerebral palsy mimics: two cases of Lesch Nyhan Syndrome
08.Disorders of vitamins, cofactors and trace elements	Submitter name		Abstract title
EP-018	Alarifi	Rakan	Transcobalamin II Deficiency Presenting as Pancytopenia and Pancreatic Insufficiency in Early Infancy
EP-020	Mohamed	Sarar	Two siblings with mild Menke disease: To treat or not to treat with Copper Histidine
10.Glycosylation disorders/CDG, protein modification disorders	Submitter name		Abstract title
EP-021	Monastiri	Kamel	GMPPA-Congenital Disorder of Glycosylation with Alacrima and Achalasia About a Family
EP-022	Yoldaş Çelik	Merve	SLC39A8-Related CDG: Diagnostic Challenges in an Infant with Overlapping Biochemical Findings
12.Lysosomal disorders	Submitter name		Abstract title
EP-023	Kara	Esra	Two distinct lysosomal storage diseases in siblings: acid sphingomyelinase deficiency and mucopolysaccharidosis type IIIA
EP-024	Arslan	Nur	Early juvenile metachromatic leukodystrophy: from initial symptoms to diagnosis
EP-025	Leoni	Simona	Incidental hyperferritinemia and progressive pulmonary alterations uncovering a lysosomal storage disorder
EP-026	Monastiri	Kamel	Infantile Pompe disease revealed by bronchiolitis at a six months old baby girl in Tunisia
EP-027	Quelhas	Dulce	Unmasking Hunter Syndrome in a PMM2-CDG Patient: When other CDG patients become the key controls
EP-028	Tulebayeva	Assel	Sleep related breathing disorder in patient with mucopolysaccharidosis II (clinical case)
16.Mitochondrial disorders	Submitter name		Abstract title
EP-029	Aydoğan	Ayça	Beware of Unexplained Hyperammonemia: Megdhel Syndrome
EP-030	Boudabous	Hela	Phenotypic Heterogeneity of DGUOK Gene Mutations: Report of Two Tunisian Paediatric Cases
EP-031	de Boer	Lonneke	Parenteral nutrition as supporting treatment in a girl with Pearson syndrome, case report
EP-032	Kasapkara	Çiğdem Seher	3-Hydroxyisobutyryl-CoA Hydrolase (HIBCH) Deficiency: Report of Two cases
EP-033	Sucur	Nediljko	Homozygous COQ2 p.Asn228Ser: retinal worsening during CoQ10 supplementation in primary deficiency

17.Neurotransmitter and creatine related disorders	Submitter name		Abstract title
EP-034	Köşeci	Burcu	Delayed Diagnosis, Dramatic Response: Segawa Syndrome Associated with TH Gene Mutation
EP-035	Doederlein Schwartz	Ida Vanessa	Beyond nonketotic hyperglycinemia: diagnostic pitfalls and the role of genetic analysis of neonatal encephalopathies
18.New Diseases	Submitter name		Abstract title
EP-036	Monastiri	Kamel	Pelizaeus–Merzbacher-Like Disease Type 2 : A Rare Disease with a Possible Founding Mutation in Tunisia
20.Newborn screening	Submitter name		Abstract title
EP-037	Belo	Anabela	Challenges in the Long-Term Management of Glutaric Aciduria Type I Despite Newborn Screening
EP-038	Perko	Dasa	Newborn Screening in Southeastern and Central Europe: Rapid Expansion but Persistent Heterogeneity
EP-039	Salimbayeva	Damilya	The use of a biobank of dried blood spots for scientific research in Kazakhstan
23.Organic acidurias	Submitter name		Abstract title
EP-040	Al Kindy	Farah	Further insights into the clinical spectrum of cobalamin J deficiency
EP-041	Alpdogan	Sedef	Evaluation of Cases with Cobalamin Metabolism Disorders:Single Center Experience
EP-042	Aras Çöl	Tuğçe	Heterogeneous Cardiac Involvement In Propionic Acidemia: From Reversible Dysfunction To Fatal Cardiomyopathy
EP-043	Aukes	Ryan	Elevated C5-OH in inborn errors of metabolism: systematic review of associated disorders and diagnostic value
EP-044	Niño-Traslaviña	Katherin	Organic acid profiling in Colombian children (0–96 months): age-related metabolic characterization
EP-045	Mohamed	Sarar	Genetic and metabolic characteristics of 194 patients with autism: A potentially novel organic acid finding
EP-046	Paponja Mihanović	Kristina	Delayed treatment of alkaptonuria in a patient with advanced ochronotic complications
EP-047	Satekge	Tumelo	Case Report: Unexpected Metabolites in an Unusual Case of Propionic Acidemia with Missed Opportunities
EP-048	Shin	Young-Lim	The effectiveness of long-term use of high-dose carglumic acid in propionic acidemia: a case report
24.Peroxisomal, sterol, bile acid, lipid and lipoprotein metabolism	Submitter name		Abstract title
EP-049	Arslan Gülten	Zümrüt	A Rare Cause of Neonatal Seizures and Hypotonia: A Case of D-Bifunctional Protein (D-BP) Deficiency
EP-050	Kasapkara	Çiğdem Seher	Chylomicronemia due to GPIHBP1 gene mutation as an Unusual Cause of False-Positive NewbornScreening
EP-051	Terner-Rosenthal	Jolan	Adjunctive Cholic Acid Therapy in Peroxisomal Biogenesis Disorder–Zellweger Spectrum Disorders: Real-World Clinical Insights
25.Phenylketonuria	Submitter name		Abstract title
EP-052	Hamazaki	Takashi	Efficacy of sepiapterin in a patient with phenylketonuria carrying the PAH p.Ser70del variant
EP-053	Mohamed	Sarar	Clinical and Molecular Spectrum of Phenylketonuria at Tertiary Center in Saudi Arabia:A Retrospective Cohort Study
EP-054	Mohamed	Sarar	DNAJC12-Related Hyperphenylalaninemia: A Rare Disorder with Challenges in Treatment Initiation
EP-055	Morotti	Hadley	Improved global outcomes in two individuals with PAH deficiency following pegvaliase treatment.
EP-056	Nicola Orejas	Gabriela Noemi	Phenylketonuria (PKU) Population in the Balearic Islands Attended at the Reference Hospital in 2025
EP-057	Usurelu	Dan-Cristian	Personalizing dietary management in phenylketonuria patients using 1H-NMR spectroscopy-based metabolomics
27.Urea cycle disorders	Submitter name		Abstract title
EP-058	Aghakıshılı	Hanım	The Theory–Practice Gap in Adult Hyperammonemia Management
EP-059	Basan	Hacer	A Case of Late-Onset Ornithine Transcarbamylase Deficiency Presenting with Status Epilepticus and Encephalopathy
EP-060	Chin	Hui-lin	Phenotypic variability in Ornithine Transcarbamylase deficiency with OTC p.Arg277Trp: case report and systematic literature review
EP-061	Mohamed	Sarar	Newborn Screening for Arginase Deficiency: Challenges and Opportunities
EP-062	Yüksel Yanbolu	Ayşe	How Much Do We Learn About Hyperammonemia During Medical School and Pediatric Residency?