Scientific Program

Sunday | 31 August 2025

International Network for Fatty Acid Oxidation Research and Management, INFORM
15:00-21:00, Kyoto Brighton Hotel (Registration at INFORM)

Monday | 1 September 2025

International Network for Fatty Acid Oxidation Research and Management, INFORM
08:00-18:00, Kyoto Brighton Hotel (Registration at INFORM)

Cerebrotendinous Xanthomatosis (CTX) International Workshop Series: Ongoing International Collaboration for a CTX Patient Registry, Clinical Severity Score and CTX Heterozygotes Research & Findings
08:15-15:45, The Prince Kyoto Takaragaike (Registration at CTX)

The 6th International Symposium on Urea Cycle Disorders (UCD): Advancements in Understanding: Global Perspectives and Innovations in Urea Cycle Disorders
17:00-20:00, Hotel Okura Kyoto (Registration at UCD)

Tuesday | 2 September 2025

The 6th International Symposium on Urea Cycle Disorders (UCD): Advancements in Understanding: Global Perspectives and Innovations in Urea Cycle Disorders
07:00-17:00, Hotel Okura Kyoto (Registration at UCD)

Challenges in adult metabolic care
08:30-12:00, Room 6 (Sakura) (Open to all participants)

CDG Satellite Symposium
09:00-12:40, Room 4 (Room B-2) (Open to all participants)

Chairs: Eva Morava, USA
Taroh Kinoshita, Japan

09:00 Incidence and Prevalence of Phosphomannomutase 2-Congenital Disorder of Glycosylation: Past, Present, and Future
Peter McWilliams, Glycomine, Inc., USA

09:20 Pathogenic mutations define new, ultra-rare Congenital Disorders of Glycosylation
Hudson Freeze, Sanford Burnham Prebys Medical Discovery Institute, USA.

09:40 Deciphering the Neurological Puzzle in ALG13-CDG Through Cortical Organoid Modeling.
Tamas Kozicz, Icahn School of Medicine at Mount Sinai, USA

10:00 Mitochondrial dysfunction is a driver of cardiac complications in PGM1-CDG with implications for therapy
Silvia Radenkovic, UMC Utrecht, Netherlands

10:20 Break

Chairs: Hudson Freeze, USA
Yoshiko Murakami, Japan

10:40 NAD precursors as a potential supportive treatment in DHDDS-CDG
Eva Morava, Icahn School of Medicine at Mount Sinai, USA

11:00 Development of Therapeutic Strategies for Fukuyama Congenital Muscular Dystrophy
Mariko Taniguchi-Ikeda, Kochi Medical School, Kochi University, JAPAN

11:20 Toward Finding a Cure for NGLY1 Deficiency
Tadashi Suzuki, RIKEN Pioneering Research Institute (PRI), Japan

11:40 Therapeutic Potential of Gene Therapy for Inherited GPI Deficiencies
Yoshiko Murakami, Research Institute for microbial diseases (RIMD), The University of Osaka, Japan

Chairs: Eva Morava, USA
Yoshiko Murakami, Japan
Junpei Tanigawa, Japan

12:00 Round Table Discussion (Topic: CDG Diagnosis & Treatment)
Taroh Kinoshita, Center for Infectious Disease Education and Research (CiDER), The University of Osaka, Japan
Yoshinao Wada, Osaka Women's and Children's Hospital, Japan
Nobuhiko Okamoto, Osaka Women's and Children's Hospital, Japan
Junpei Tanigawa, Graduate School of Medicine, The University of Osaka, Japan

JSIMD Board Meeting
13:00-15:00, Meeting Room M2 (Room C-2) (Closed)

Metabolic Dietitians Group Meeting
13:00-16:30, Room 2 (Room A) (Open to all participants)

Registration
15:00-18:00, Event Hall

JIMD Editorial Board Meeting
15:00-19:00, Meeting Room M1 (Room C-1) (Closed)

ACIMD Board Meeting
15:30-16:30, Meeting Room M2 (Room C-2) (Closed)

Welcome to Kyoto
16:30-18:00, Swan

Wednesday | 3 September 2025

Registration
08:00-20:00, Event Hall

2029 ICIEM Scientific Organizing Committee meeting
08:00-09:00, Meeting Room M2 (Room C-2) (Closed)

Opening Ceremony
09:30-10:00, Room 1 (Main Hall)

Posters
09:30-19:00, Annex Hall / Event Hall

Keynote Lecture 1
Chair: Yoshikatsu Eto, Japan
10:00-10:45, Room 1 (Main Hall)

10:00 KL1 **My Life in the Mitochondria**
John Walker, Nobel Laureate in Chemistry (1997), University of Cambridge, UK

Symposium 1: Cutting-Edge Basic Research
Chairs: Yair Anikster, Israel
Ljubica Caldovic, USA
11:00-12:30, Room 1 (Main Hall)

11:00 SP1-1 **Congenital Disorders of Glycosylation (CDG): Expanding Horizons and Subterranean Currents**
Hudson H. Freeze, Sanford Burnham Prebys Medical Discovery Institute, USA

11:20 SP1-2 **Programmable RNA/RNP Switches for Cell-Specific Gene Control and Next-Generation Therapeutics**
Hirohide Saito, RNP Synthetic Biology and Biotechnology Institute for Quantitative Biosciences, The University of Tokyo, JAPAN / Center for iPS Cell Research and Application (CiRA), Kyoto University, JAPAN

11:40 SP1-3 **The utility of iPSCs and gene editing to study inborn errors of metabolism and genetic adaptation.**
Knut Woltjen, Center for iPS Cell Research and Application (CiRA), Kyoto University, Japan

12:00 SP1-4 **Bidirectional Control of mRNA Translation via RNA G-quadruplex Induction Using Staple Oligomers**
Yousuke Katsuda, Faculty of Advanced Science and Technology, Kumamoto University, StapleBio Inc., Japan

Symposium 2: Gene therapy from Japan, the Land of Tradition
Chairs: Kimihiko Oishi, Japan
Gerard Berry, USA
11:00-12:30, Room 2 (Room A)

11:00 SP2-1 Current status and challenges of gene therapy for pediatric genetic diseases
Masafumi Onodera, Graduate School of Engineering, The University of Osaka, Japan

11:20 SP2-2 Recent Advances and Challenges in Gene Therapy for Metabolic, and Neurological Disorders in Japan
Kazuhiro Muramatsu, Jichi Medical University, Department of Pediatrics and Center for Gene Therapy Research, Japan

11:40 SP2-3 Research and Development of Gene Therapy for Pompe disease
Hiroshi Kobayashi, Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine, Japan

12:00 SP2-4 Gene therapy for inborn errors of metabolism: A regulatory perspective
Akihiro Kume, Jichi Medical University, Japan

Oral 1: Lysosomal Disorders 1
Chairs: Giancarlo la Marca, Italy
Marc Patterson, USA
11:00-12:30, Room 3 (Room B-1)

11:00 OP1-1 A Meta-analysis to Unveil the Diagnostic Gaps in Anderson-Fabry Disease in Women
Livia Lenzini, Department of Medicine, University of Padova,Italy

11:15 OP1-2 Reduced incidence of stroke in patients with Fabry disease treated with agalsidase beta: a matched analysis from the Fabry registry
Alessandro Burlina, Dept. of Medicine, Neurology Unit, St. Bassiano Hospital, Italy

11:30 OP1-3 Plasma glial fibrillary acidic protein is a promising biomarker for central nervous system involvement in infantile-onset Pompe disease
Neha Regmi, Duke University School of Medicine, USA

11:45 OP1-4 From Infancy to Adulthood: 23 Years of Single-Center Follow-Up in the Dutch Classic Infantile Pompe Disease Cohort
A once-lethal disease transformed by enzyme replacement therapy
Johanna M.P. van den Hout, Center for Lysosomal and Metabolic Diseases, Department of Pediatrics, Erasmus MC Medical Center, Netherlands

12:00 OP1-5 Improved Outcomes in Very Early Treated Infantile-Onset Pompe Disease after Two Years Switching to Avalglucosidase Alfa: Real-world Data from Patients Diagnosed through Taiwan Newborn Screening
Chia-Feng Yang, Taipei Veterans General Hospital, Taiwan

12:15 OP1-6 PROPEL Australia subpopulation: efficacy and safety of cipaglucosidase alfa plus miglustat versus alglucosidase alfa in patients with late-onset Pompe disease
Michel Tchan, Department of Genetic Medicine, Westmead Hospital, Australia

Oral 2: Artificial Intelligence in Research
Chairs: Dau-Ming Niu, Taiwan
Ryuichi Mashima, Japan
11:00-12:30, Room 4 (Room B-2)

11:00 OP2-1 **Explainable Predictive Modeling for Mucopolysaccharidoses Early Diagnosis**
Ruba Fadul, Department of Biomedical Engineering and Biotechnology, Khalifa University, UAE

11:15 OP2-2 **Geo-temporal Analysis of Social Media Conversations on Mucopolysaccharidosis: Insights from Dynamic Topic Modeling**
Natnael Tumzghi Tsegai, Khalifa University, UAE

11:30 OP2-3 **Harnessing computational and experimental structural biology into an AI pipeline to create an atlas of disease-causing mutations**
Mihaela Atanasova, Centre for Medicines Discovery, Nuffield Department of Medicine, University of Oxford, UK

11:45 OP2-4 **Comparative validation of "in silico" predictors for VUS in lysosomal storage disorders**
Isidro Arevalo-Vargas, Fundación para el Estudio y la Terapéutica de la Enfermedad de Gaucher y Otras Lisosomales (FEETEG), Spain

12:00 OP2-5 **Elucidating a potential role of the infant gut microbiome on the bioavailibity of tyrosine in PKU**
Ines Thiele, University of Galway, Ireland

12:15 OP2-6 **ChatIMD: A Clinical Decision Support Tool for Inherited Metabolic and Rare Diseases**
Tim Hulshof, School of Medicine, University of Galway, Ireland / Digital Metabolic Twin Centre, University of Galway, Ireland / Ryan Institute, University of Galway, Ireland

Oral 3: Phenylketonuria 1
Chairs: Francjan J van Spronsen, Netherlands
David J Coman, Australia
11:00-12:30, Room 5 (Room D)

11:00 OP3-1 **Effect of enzyme substitution therapy on brain magnetic resonance imaging and cognition in adults with classical phenylketonuria**
Alberto B Burlina, Inherited Metabolic Diseases Unit,University Hospital of Padua, Italy

11:15 OP3-2 **Balancing phenylalanine, tyrosine and tryptophan to modulate neurotransmitter concentrations in early-treated phenylketonuria: An FDPD^{+}-databank study**
Ellis Marleen van Steenis, University Medical Center Groningen, Netherlands

11:30 OP3-3 **Patient Reported Outcomes in Phenylketonuria: PHEFREE- The US National Institutes of Health Rare Disease Consortium for Hyperphenylalaninemia**
Georgianne Lee Arnold, University of Pittsburgh Medical Center, USA

11:45 OP3-4 **Neuropsychiatric Comorbidities in Adults with PKU in Sweden**
Andreas Kindmark, Department of Medical Sciences, Uppsala University Hospital, Sweden

12:00 OP3-5 **Longitudinal study of blood metabolome alterations and in vivo brain energy metabolism in adulthood in a mouse model of PKU.**
Yann Dos Santos, Université de Tours, INSERM, Imaging Brain & Neuropsychiatry iBraiN U1253, France

12:15 OP3-6 **Performance of the Egoo Test for Phenylalanine Measurement in Females with Phenylketonuria**
Rani H Singh, Department of Human Genetics, Emory University School of Medicine, USA

Oral 4: Dietetics and Nutrition
Chairs: Yoko Nakajima, Japan
Alice Dianin, Italy
11:00-12:30, Room 6 (Sakura)

11:00 OP4-1 **How good is blood Phe control in Maternal PKU in Europe: results from 102 pregnancies**
Alex Pinto, European longitudinal project on blood Phe Control in PKU, UK

11:15 OP4-2 **Global use of casein glycomacropeptide in phenylketonuria: health professional perspectives**
Sharon Evans, Birmingham Children's Hospital, UK

11:30 OP4-3 **The micronutrient content of protein substitutes for Phenylketonuria diet: time to pay attention**
Albina Tummolo, Department of Metabolic Diseases and Clinical Genetics, Giovanni XXIII Children Hospital, Italy

11:45 OP4-4 **Macro and micronutrient intake in galactosaemia patients**
Alex Pinto, Birmingham Children's Hospital, UK

12:00 OP4-5 **DETECTION OF COBALAMIN DEFICIENCY THROUGH NEWBORN SCREENING: NEONATAL METABOLIC FINDINGS AND MATERNAL NUTRITIONAL STATUS IN AN ITALIAN COHORT**
Elvira Verduci, Metabolic Diseases Unit, Department of Pediatrics, Vittore Buzzi Children's Hospital; Department of Health Sciences, University of Milan, Italy

12:15 OP4-6 **Comparison of the benefits of Glycosade and Maizena in the pediatric treatment of GSD : the experience of Emilia Romagna**
Egidio Candela, Pediatric Unit, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Italy

Oral 5: Novel Diagnostic/Laboratory Methods Including Omics
Chairs: Hideo Sasai, Japan
Frank Rutsch, Germany
11:00-12:30, Room 7 (Room E)

11:00 OP5-1 **Deep Learning-Driven Detection of Cryptic Splice-Altering Variants in Non-Coding Regions Predicting Metabolomic Shifts in Unsolved Inborn Errors of Metabolism**
Sahnaz Vivinda Putri, Health Management Laboratory, Indonesia Open University, Indonesia

11:15 OP5-2 **Blood RNA-seq in Rare Disease diagnostics: A Comparative Study of Cases With and Without Candidate Variants**
Xiaomei Luo, Clinical Genetics Center, Shanghai Institute for Pediatrics, Xinhua Hospital affiliated to Shanghai Jiao Tong University School of Medicine, China

11:30 OP5-3 **Untargeted proteomics enables ultra-rapid variant prioritization in mitochondrial and other rare diseases and establishes XRN1 as a novel disease gene**
David Thorburn, Murdoch Children's Research Institute, Australia

11:45 OP5-4 **A Population Genomic Framework for a Variant Interpreting Atlas of IDS Missense Variants in Mucopolysaccharidosis Type II**
Shuhei Sako, The Jikei University School of Medicine, Japan / National Hospital Organization Nishisaitama-Chuo Hospital, Japan

12:00 OP5-5 **Newborn screening for X-ALD: Untargeted Lipidomics Identifies Novel Lipid Biomarkers**
Won-Tae Kim, New South Wales Newborn Screening Program, Australia

12:15 OP5-6 **Age-Specific Phytosterol Cutoffs Differentiate Sitosterolemia from Family Hypercholesterolemia in Children: A three-year prospective study**
Mengyuan Wu, Center for molecular medicine, Children's Hospital of Fudan University, China

SIMD Board Meeting
11:30-13:30, Meeting Room M1 (Room C-1) (Closed/ only for SIMD Board Members)

Industry-Sponsored Symposium
12:45-13:45, Room 2 (Room A)

Industry-Sponsored Symposium
12:45-13:45, Room 3 (Room B-1)

Industry-Sponsored Symposium
12:45-13:45, Room 4 (Room B-2)

Industry-Sponsored Symposium
12:45-13:45, Room 5 (Room D)

Industry-Sponsored Symposium
12:45-13:45, Room 6 (Sakura)

Lunch Service
13:00-14:00, Event Hall / Swan

Symposium 3: Global Trends in Gene Therapy
Chairs: Mireia Del Toro, Spain
Gerry Lipshutz, USA
14:00-15:30, Room 1 (Main Hall)

14:00 SP3-1 Leveraging Gene Insertion and Gene Editing Therapies for Congenital Disorders of Glycosylation
Stacy E Croteau, Boston Children's Hospital, USA

14:20 SP3-2 Oligonucleotide Therapies for Inherited Metabolic Disorders.
Haiyan Zhou, University College London, UK

14:40 SP3-3 Progress in the Long Road for Gene Therapy for Ornithine Transcarbamylase Deficiency (OTCD)
J Lawrence Merritt, II, Ultragenyx Pharmaceutical, USA

15:00 SP3-4 Emerging Nucleic Acid-Based Therapies for Congenital Disorders of Glycosylation (CDG)
Kent Lai, University of Utah, USA

Symposium 4: Transformation in Newborn Screening
Chairs: Susan Berry, USA
Sarah Viall, USA
14:00-15:45, Room 2 (Room A)

14:00 SP4-1 **Dissecting Mitochondrial Disease Mechanisms Through Integrative Multi-Omics**
Patrick Forny, Washington University School of Medicine in St. Louis, USA

14:20 SP4-2 **Toward the development of a novel newborn screening modality: In-depth high-throughput non-targeted proteome approach using dried blood spots**
Osamu Ohara, Kazusa DNA Research Institute, JAPAN

14:40 SP4-3 **Identifying circulating mechanistic biomarkers of mitochondrial disease**
Rohit Sharma, Massachusetts General Hospital, USA

15:00 SP4-4 **Whole Genome Sequencing Analysis: From Newborn Screening to Lifelong Insights**
Dau-Ming Niu, Institute of Clinical medicine, National Yang Ming Chiao Tung University / Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan

15:20 SP4-5 **Newborn Screening in Taiwan: Program Updates and Real-World Outcomes**
Yin-Hsiu Chien, National Taiwan University Hospital, Taiwan

Oral 6: Lysosomal Disorders 2
Chairs: Masahisa Kobayashi, Japan
Aya Narita, Japan
14:00-16:00, Room 3 (Room B-1)

14:00 OP6-1 **Blended Phenotypes in Gaucher Disease: Revealing Multi-Locus Disorders Shaping Precision Medicine**
Pramod Mistry, Yale School of Medicine, USA

14:15 OP6-2 **Unveiling Liver Cancer Risk in Gaucher Disease: Spatial Transcriptomics and Immune Profiling Reveal an Oncogenic Hepatic Milieu**
Pramod Mistry, Yale University School of Medicine, USA

14:30 OP6-3 **Targeting Gaucher disease via iminosugar chaperone-antioxidant conjugates: A dual-action therapeutic strategy**
Francesca Clemente, Department of Chemistry (DICUS), University of Florence, Italy

14:45 OP6-4 **Decoding Bone Involvement in Gaucher Disease through miRNA Profiles from plasmatic exosomes.**
Irene Serrano-Gonzalo, Fundación Española para el Estudio y Tratamiento de la Enfermedad de Gaucher y otras lisosomales (FEETEG), Spain / Grupo de Investigación de Enfermedad de Gaucher (GIIS-012). IIS Aragón, Spain / Grupo de Investigación Mecanismos de Enfermedad Crónica e Investigación Traslacional, Spain

15:00 OP6-5 **EXPLORING GBA1 GENE IN PARKINSON'S DISEASE: PREVALENCE AND VARIANT SPECTRUM FROM ASIA MINOR**
Merve Koc Yekeduz, Ankara University School of Medicine, Department of Pediatric Metabolism, Turkey / Harvard Medical School, Boston Children's Hospital, Department of Anesthesiology, Critical Care and Pain Medicine, USA

15:15 OP6-6 **Lentiviral Hematopoietic Stem Cell Gene Therapy for Late Juvenile Metachromatic Leukodystrophy: Interim Results of a Phase III Study**
Valeria Calbi, San Raffaele Telethon Institute for Gene Therapy (SR-TIGET), Italy / Pediatric Immunohematology Unit and BMT Program, IRCCS San Raffaele Scientific Institute, Italy

15:30 OP6-7 HYDROXYPROPYL-BETA-CYCLODEXTRIN FOR THE LONG-TERM TREATMENT OF NIEMANN PICK TYPE C1: EFFICACY AND SAFETY DATA FROM 4 CLINICAL STUDIES AND THE ONGOING EXPANDED ACCESS PROGRAM
Caroline Hastings, UCSF Benioff Children's Hospital Oakland, USA / Gazi University Hospital, Turkey

15:45 OP6-8 Identification of cepharanthine as a potential therapy of acid sphingomyelinase deficiency by reducing cellular sphingosylphosphorylcholine
Mengni Yi, Pediatric Endocrinology and Genetics, Xinhua Hospital, Shanghai Institute for Pediatric Research, Shanghai Jiao Tong University School of Medicine, China

Oral 7: Translational Research and Novel Diseases

Chairs: Yair Anikster, Israel and USA
Tetsuya Ito, Japan

14:00-16:00, Room 4 (Room B-2)

14:00 OP7-1 Graphene flakes for enhanced delivery of the enzyme to fibroblasts derived from the patients with lysosomal storage disorders
Sandra Vranic, Nano-Cell Biology Lab, Division of Cell Matrix Biology & Regenerative Medicine, School of Biological Sciences, Faculty of Biology, Medicine and Health, The University of Manchester, UK / Centre for Nanotechnology in Medicine, Faculty of Biology, Medicine and Health, The University of Manchester, UK

14:15 OP7-2 Nervonic Acid Targets Pathological Very Long Chain Fatty Acids in Adrenoleukodystrophy: Evidence from Preclinical Mouse Studies
Reena V Kartha, University of Minnesota, USA

14:30 OP7-3 One-carbon and NAD+ metabolic fluxes in ex vivo precision-cut liver slices of Glycogen Storage Disease type I.
Esther Boelina Homan, Laboratory of Pediatrics, University of Groningen, University Medical Center Groningen, Netherlands

14:45 Withdrawal

15:00 OP7-5 NMNAT3 DEFICIENCY: A NOVEL INBORN METABOLIC DISORDER CAUSING HEMOLYTIC ANEMIA
Judith J.M. Jans, Metabolic Diagnostics, Department of Genetics, University Medical Center Utrecht, Netherlands

15:15 OP7-6 Biallelic SIDT2 Loss-of-Function Associated with Cerebellar Ataxia and Lysosomal Dysfunction Mimics Impairment of SIDT2 in Mice
Grace Yoon, The Hospital for Sick Children, Canada

15:30 OP7-7 Phosphatidylinositol 4-kinases: their fundamentals, role on myelin formation, and their emergent relevance in undiagnosed diseases within pediatric populations.
Alejandro Alvarez-Prats, NICHD, National Institutes of Health, USA

15:45 OP7-8 Deoxythymidylate Kinase (DTYMK) Deficiency: A New Inherited Metabolic Disease with Intrafamilial Variability
Ching Wan Lam, The University of Hong Kong , China

Oral 8: Organic Acidemias
Chairs: Shamima Rahman, UK
Sabine Scholl-Bürgi, Austria
14:00-16:00, Room 5 (Room D)

14:00 OP8-1 **Clinical burden of Propionic Acidemia in Japan: A real-world evidence cohort study using a hospital-based healthcare database**
Yoko Nakajima, Fujita Health University, Japan

14:15 OP8-2 **Long-term complications in methylmalonic acidaemia in adults**
Samreen Safdar, Salford Royal Hospital, NCA, UK

14:30 OP8-3 **Outcome of dietary treatment of short-chain enoylCoA hydratase deficiency in a Pacific cohort**
Isaac Bernhardt, Adult and Paediatric National Metabolic Service, Starship Children's Health, Te Toka Tumai, Te Whatu Ora | Health New Zealand , New Zealand

14:45 OP8-4 **Cell-specific metabolic dependencies influence propionate catabolism in methylmalonic aciduria**
Caroline Tanja Frei, University Children's Hospital Zurich, Switzerland / University of Zurich, Switzerland

15:00 OP8-5 **Mitochondria-targeted therapies improve bioenergetics in Glutaric Acidemia type 1 patient's fibroblasts**
Bianca Seminotti, UPMC Children's Hospital of Pittsburgh, USA / Division of Genetic and Genomic Medicine, Department of Pediatrics, University of Pittsburgh School of Medicine, Pittsburgh, USA

15:15 OP8-6 **Rescue of Glutaric Aciduria Type I by GalNAC conjugated siRNA against Aminoadipate Semialdehyde Synthase**
Karl-Dimiter Bissig, Duke University, USA Minor Outlying Islands

15:30 OP8-7 **mRNA-3705 Therapy for Methylmalonic Acidemia: Interim Data from a Phase 1/2 Study**
Sabine Fuchs, University Medical Center Utrecht, Netherlands

15:45 OP8-8 **mRNA-3927 for the treatment of propionic acidemia: final results from mRNA-3927-P101 Part 1 dose-escalation cohorts and cumulative data from ongoing participants**
Andreas Schulze, Hospital for Sick Children and University of Toronto, Canada

Special Symposium 1: From Pathogenesis to Therapy, From Japan to the World: Global Translation, Metabolic Flux, and Gene Editing for Citrin Deficiency
Chairs: Kimitoshi Nakamura, Japan
Barbara Yu, Singapore and UK
14:00-16:00, Room 6 (Sakura)

14:00 SSP1-1 **Understanding Citrin Deficiency Within the Spectrum of Liver Metabolic Diseases and Urea Cycle Disorders**
Johannes Häberle, University Children's Hospital Zurich, Switzerland

14:25 SSP1-2 **Metabolic Flux Analysis and New Biochemical Insights in Citrin Deficiency (CD)**
Marc Kopel Hellerstein, University of California, Berkeley, USA

14:50 SSP1-3 **Correcting Genetic Liver Diseases by Prime Editing**
Gerald Schwank, University Zurich, Switzerland

15:15 SSP1-4 **The status of adult patients with citrin deficiency in Japan; From the nation-wide study of Japan**
Jun Kido, Department of Pediatrics, Faculty of Life Sciences, Kumamoto University, Japan

15:30 SSP1-5 **Proteomic Approaches to Newborn Screening for Citrin Deficiency**
Yoichi Wada, Tohoku University Hospital, Japan

15:45 SSP1-6 Financial Realities and the Way Forward for Novel Therapies in Rare Diseases
Barbara Yu, Citrin Foundation, Singapore and UK

Oral 9: Urea Cycle Disorders
Chairs: Nicola Brunetti-Pierri, Italy
Nithiwat Vatanavicharn, Thailand
14:00-16:00, Room 7 (Room E)

14:00 0P9-1 Systemic mRNA therapy crosses the blood-brain barrier and reverses the neurological phenotype in pre-clinical model of Argininosuccinic Aciduria
Sonam Gurung, Great Ormond Street Institute of Child Health, University College London, UK

14:15 0P9-2 Argininosuccinate lyase deficiency leads to impaired blood brain barrier through transcriptional and translational regulation of cell junctional proteins
Hsiang-Chun Chang, Department of Molecular and Human Genetics, Baylor College of Medicine, USA

14:30 Withdrawal

14:45 0P9-4 Durable efficacy and safety of DTX301: Long-term follow up of a phase 1/2 trial in adults with ornithine transcarbamylase deficiency
Tarekegn Geberhiwot, University of Birmingham, UK

15:00 0P9-5 Pharmacological chaperones as novel treatment strategy for ornithine transcarbamylase deficiency
Alexander Laemmle, Division of Pediatric Endocrinology, Diabetology and Metabolism, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland / University Institute of Clinical Chemistry, Inselspital, Bern University Hospital, University of Bern, Switzerland / Department of Biomedical Research, University of Bern, Switzerland

15:15 0P9-6 A Phase 1 and 1b study in healthy subjects and OTC heterozygotes with CMP-CPS-001-an investigational antisense oligonucleotide for the treatment of Urea Cycle Disorders (UCDs)
Yuri Maricich, CAMP4 Therapeutics, USA

15:30 0P9-7 Succinyl-CoA synthetase reaction (succinate-forming) is a reasonable target of anaplerotic therapy for hyperammonemia
Makoto Yoshino, Cognitive and Molecular Research Institute of Brain Disease, Kurume University, Japan

15:45 0P9-8 Prime editing achieves durable in vivo correction of phenylketonuria and citrullinemia type I
Gerald Schwank, University Zurich, Switzerland

Coffee Break
15:45-16:15, Annex Hall / Event Hall / Swan

Oral 10: Lysosomal Disorders 3
Chairs: Toya Ohashi, Japan
Takanobu Otomo, Japan
16:00-17:30, Room 1 (Main Hall)

16:00 0P10-1 Behavioural and biochemical validation of a novel acid ceramidase disorder mouse model with epileptiform presentation.
Luis Ángel Albarrán-Ponce, Neurolipidomics Laboratory and India Taylor Lipidomics Research Platform, University of Ottawa Brain and Mind Research Institute, Ottawa Institute of Systems Biology, Department of Biochemistry, Microbiology and Immunology, University of Ottawa, Canada

16:15 Withdrawal

16:30 OP10-3 Restoration of enzyme activity in mucopolysaccharidosis IVA fibroblasts using exosomes derived from mesenchymal stem cell overexpressing GALNS gene
Minji Im, Samsung Medical Center, Republic of Korea

16:45 OP10-4 Development of gene editing technologies to correct a mutation in GNPTAB of mucolipidosis type II/III patients
Ryunosuke Sanada, Center for Regenerative Medicine, National Center for Child Health and Development., Japan / Department of NCCHD Child Health and Development, Graduate School of Medical and Dental Sciences, Institute of Science Tokyo., Japan

17:00 OP10-5 Novel regulators of the Mannose 6-phosphate pathway
Sabrina Jabs, Institute of Clinical Molecular Biology, Kiel University/ University Hospital Schleswig-Holstein, Kiel, Germany

17:15 OP10-6 Derivation of iPSC-Based Human Neural Models for Investigating the Pathophysiology of Sialidosis
Rodolfo Tonin, Neuroscience and Medical Genetics Department, Meyer Children's Hospital IRCCS, Italy

Oral 11: Mitochondrial Disorders 1
Chairs: David R. Thorburn, Australia
Masaru Shimura, Japan
16:00-17:30, Room 2 (Room A)

16:00 OP11-1 Recent Advances in Genetic Diagnosis and Management of Mitochondrial Diseases in Japan
Kei Murayama, Juntendo University, Japan / Diagnostics and Therapeutics of Intractable Disease, Intractable Disease Research Center. Juntendo University Faculty of Medicine, Japan / Department of Metabolism, Chiba Children's Hospital, Japan

16:15 OP11-2 Tissue-Specific Mitochondrial DNA Variant Distribution and Its Clinical Implications in a Japanese Cohort
Atsuko Okazaki, Juntendo University, Graduate School of Medicine, Diagnostics and Therapeutics of Intractable Diseases, Japan

16:30 OP11-3 Publicly Funded Genomic Sequencing For Mitochondrial Disorders In Australia: Uptake And Outcomes
Megan Ball, Murdoch Children's Research Institute, Australia

16:45 OP11-4 Molecular Epidemiology of Primary Mitochondrial Diseases in Spain
Marcello Bellusci, Reference Centre for Inherited Metabolic Disorders, 12 de Octubre University Hospital, Spain / Biomedical Network Research Centre on Rare Diseases (CIBERER), Instituto de Salud Carlos III, Spain / Mitochondrial & Neuromuscular Disorders Laboratory, Instituto de Investigación Sanitaria 12 de Octubre (imas12), Spain

17:00 OP11-5 Feasibility of diagnosis of mitochondrial diseases by -omics technologies on whole blood
Bram Decru, Department of Laboratory Medicine, UZ Leuven, Belgium / Metabolics Expertise Center, VIB-KU Leuven, Belgium / Lab of applied Mass Spectrometry, KU Leuven, Belgium

17:15 OP11-6 Untargeted proteomics greatly aides the functional diagnosis of mitochondrial aminoacyl-tRNA synthetases (ARS2s)
Johan LK Van Hove, Department of Pediatrics, University of Colorado, USA

Oral 12: Clinical Studies and Patient-Reported Outcomes 1
Chairs: Markey McNutt, USA
Meow-Keong Thong, Malaysia
16:00-17:45, Room 3 (Room B-1)

16:00 OP12-1 **Ataxia-Telangiectasia: Treating Mitochondrial Dysfunction with a novel form of Anaplerosis**
David J Coman, Department of Metabolic Medicine, Queensland Children's Hospital, Australia / Child Health Research Centre, Faculty of Medicine, University of Queensland, Australia

16:15 OP12-2 **Potential benefits of L-serine in children with GRIN2B loss-of-function variants: randomized n-of-1 trials**
Clara van Karnebeek, Emma Center for Personalized Medicine, Netherlands / United for Metabolic Diseases, Netherlands

16:30 OP12-3 **The SIMPATHIC basket trial to target shared pathomechanisms for personalized medicine in metabolic disorders**
Annelieke R. Muller, Amsterdam University Medical Center, Emma Center for Personalized Medicine, Netherlands / Amsterdam UMC location University of Amsterdam, Department of Pediatrics and Human Genetics, Netherlands / Amsterdam UMC, Amsterdam Gastroenterology Endocrinology Metabolism; Amsterdam Public Health - Personalized Medicine and Methodology, Netherlands

16:45 OP12-4 **Development of a radiographic vertebral severity score for evaluation of disease progression in alkaptonuria**
Francis Rossignol, Human Biochemical Genetics Section, Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health, USA

17:00 OP12-5 **Long-term safety and efficacy of pegtibatinase for treatment of classical homocystinuria (HCU): data from the Phase 1/2 COMPOSE® open-label extension study**
Can Ficicioglu, Children's Hospital of Philadelphia, USA

17:15 OP12-6 **International Patient Reported Data Accelerating Research for Homocystinuria**
Brittany Parke, HCU Network America, USA

17:30 OP12-7 **Mid- and long-term evolution after liver +/- kidney transplantation for methylmalonic acidemia**
Margaux Gaschignard, Metabolic center Necker Hospital, France

Oral 13: Disorders of Vitamins, Cofactors and Trace Elements
Chairs: Julien Park, Germany
Anita Inwood, Australia
16:00-17:30, Room 4 (Room B-2)

16:00 OP13-1 **Characterisation of the Slc30a10 Knockout Mouse: A Preclinical Model for Manganese-Induced Neurotoxicity and Therapeutic Development**
Hendrik Vogt, University College London, UK

16:15 OP13-2 **Long-term Outcomes of Patients with Cobalamin C Deficiency Diagnosed Through Newborn Screening**
Yue Huang, University of California, San Diego, USA / Rady Children's Hospital, USA

16:30 OP13-3 **Biotinidase deficiency, a Treatable Neurometabolic Disorder: Largest cohort from India-Single Center Experience.**
Balamurugan Nagarajan, Pediatric Neurology Consultant, Rainbow Children's Hospital, India

16:45 OP13-4 **Lessons from 4 year diagnosing and monitoring pyridoxine dependent epilepsy by quantitative analysis of 2S,6S-/2S,6R-oxopropylpiperidine-2-carboxylic acid (2-OPP) and 6-oxopiperidine-2-carboxylic acid (6-oxoPIP)**
Anke P Willems, Radboudumc, Netherlands

17:00 OP13-5 The impact of beta-carotene accumulation on metabolism and neurodevelopment in rats
Xue Ma, Children's Medical Center, Peking University First Hospital, China

17:15 OP13-6 Integrated multi-omics identifies transcriptional dysregulation in remethylation disorders
Daphné Chopard, Department of Computer Science, ETH Zurich, Switzerland / Department of Intensive Care and Neonatology and Children's Research Center, University Children's Hospital Zurich, University of Zurich, Switzerland

Oral 14: Amino Acid Disorders 1

Chairs: Chika Takano, Japan
Carlo Dionisi-Vici, Italy
16:00-17:30, Room 5 (Room D)

16:00 OP14-1 Elevation of branched chain amino acids due to Branched Chain Amino Acid Transferase 2 deficiency (BCAT2): to treat or not to treat?
Guido Zago, Department of Paediatric Metabolic Medicine, Great Ormond Street Hospital NHS Trust , UK

16:15 OP14-2 Peripheral Neuropathy in variant MSUD new cases
Charles John Billington, University of Minnesota, USA

16:30 OP14-3 Alternative sources of valine and isoleucine for prompt reduction of plasma leucine in maple syrup urine disease patients
Maryam Ziadlou, Department of Food Science and Technology, Faculty of Agriculture, Science and Research Branch Islamic Azad University, Iran

16:45 OP14-4 Expanding the Therapeutic Landscape of MSUD: Insights from Chilean Dietary Management
Tracy Fuenzalida, Laboratorio de Genética y Enfermedades Metabólicas, Chile

17:00 OP14-5 An open-label, prospective, interventional study to determine the optimal treatment of classical homocystinuria (HCU) in infants identified through newborn screening in Qatar
Tawfeg Ben-Omran, Sidra Medicine, Qatar / Hamad Medical Corporation, Qatar

17:15 OP14-6 Identification and characterization of pharmacological chaperones for cystathionine beta-synthase-deficient homocystinuria
Tomas Majtan, Department of Pharmacology, Faculty of Science and Medicine, University of Fribourg, Switzerland

Oral 15: Newborn Screening 1

Chairs: Sarah Viall, USA
Yew Sing Choy, Malaysia
16:00-17:30, Room 7 (Room E)

16:00 OP15-1 Inclusion of metachromatic leukodystrophy in newborn screening program: development of a diagnostic algorithm and ad interim results of a prospective pilot
Giancarlo la Marca, Newborn Screening, Clinical Biochemistry and Clinical Pharmacy Laboratory, Meyer Children's Hospital IRCCS, Italy / Department of Experimental and Clinical Biomedical Sciences "Mario Serio", University of Florence, Italy

16:15 OP15-2 Prospective Newborn Screening for Metachromatic Leukodystrophy in Austria: Updated Results and Findings
Petra Oliva, ARCHIMEDlife GmbH, Austria

16:30 OP15-3 X-Linked Adrenoleukodystrophy and Newborn Screening: an experience of extended screening for families in the city of Sao Paulo
Fernanda De Castro Monti Rabelo, Jo Clemente Institute, Brazil / Neurometabolic Clinic, Neurology Department, Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo, Brazil

16:45 OP15-4 Newborn Screening for X-linked Adrenoleukodystrophy in Wisconsin: What We Have Learned So Far
Mei Baker, Division of Genetics and Metabolism, Department of Pediatrics, University of Wisconsin School of Medicine and Public Health, USA

17:00 OP15-5 California's Experience with Newborn Screening for Guanidinoacetate Methyltransferase Deficiency: Insights into Tier–1 and Tier–2 Testing and the Arginine/Guanidinoacetate Ratio
Kuldeep Dhillon, Genetic Disease Screening Program, California Department of Public Health, USA

17:15 OP15-6 A New Metabolic Disease Candidate for Newborn Screening Program: Pyridoxine Dependent Epilepsy (PDE)
Giancarlo la Marca, Newborn Screening, Clinical Biochemistry and Clinical Pharmacy Laboratory, Meyer Children's Hospital IRCCS, Italy / Department of Experimental and Clinical Biomedical Sciences "Mario Serio", University of Florence, Italy

Garrod Award Lecture
Chair: Matthias Baumgartner, Switzerland
17:30-18:00, Room 1 (Main Hall)

Neurological disease in argininosuccinic aciduria: a bedside-to-bench story
Julien Baruteau, Great Ormond Street Institute of Child Health, University College London, UK / Great Ormond Street Hospital for Children, UK

Poster 1: Best Poster Award
Chairs: Mitsuru Kubota, Japan
Kristina L. Elvidge, Australia
18:00-19:00, Exhibition & Poster 2 (Event Hall)

BP-01 Genetic Variants and Clinical Features of Patients With Glycogen Storage Disease Ib
Qiu Wenjuan, Department of Pediatric Endocrinology and Genetic Metabolism, Xinhua Hospital, Shanghai Institute of Pediatric Research, School of Medicine, Shanghai Jiao Tong University, China

BP-02 Dual Vascular Changes in Fabry Disease as Neuroimaging Marker
Chong Kun Cheon, DEPARTMENT OF PEDIATRICS, PUSAN NATIONAL UNIVERSITY CHILDREN'S HOSPITAL, Republic of Korea

BP-03 Report on the activities of JaSMIn (Japan Registration System for Metabolic & Inherited Diseases).
Satoko Tsushima, Nursing Department, Specialist Nursing Office, Genetic Coordinator,National Center for Child Health and Development, Japan

BP-04 Low– fat mother´s milk for infants with lipid metabolism disorders¦ promising results from a pre– clinical study
Susanne Dirne-van Alst, Department of Internal Medicine, division of Dietetics, Erasmus Medical Center, Netherlands

BP-05 An interactive, searchable database of LC-FAOD gene variants, genotypes and phenotypes
Aneal Khan, M.A.G.I.C. (Metabolics and Genetics in Canada) Clinic Ltd., Canada

BP-06 Individualized Dosing and Long-Term Outcomes of Givosiran in Recurrent Acute Hepatic Porphyria
Eliane Sardh, Centre for Inherited Metabolic Diseases, Porphyria Centre Sweden, Karolinska University Hospital, Department of Molecular Medicine and Surgery, Karolinska Institutet, Sweden

BP-07 The current status of clinical management and molecular features of congenital disorders of glycosylation in Japan
Nobuhiko Okamoto, Department of Medical Genetics, Osaka Women's and Children's Hospital, Japan / Department of Molecular Genetics and Endocrinology, Research Institute, Osaka Women's and Children's Hospital, Japan

BP-08 The role of Golgi apparatus in human brain development and neural stem cell fate choice: A lesson from the study of COG5-CDG
Elena Taverna, Human Technopole, Italy

Poster 2: Best Poster Award
Chairs: José Abdenur, USA
Kaustuv Bhattacharya, Australia
18:00-19:00, Exhibition & Poster 2 (Event Hall)

BP-09 Development of Small Molecule Therapeutics Targeting Aberrant RNA Splicing in Cardiac Variant Fabry Disease
Ya-Chi Chen, Genetic Consultant Center Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan / Department of Pediatrics, Taipei Veterans General Hospital, Taiwan

BP-10 In vivo editing of the transferrin locus for systemic correction of lysosomal storage diseases: proof-of-concept in mouse models of MPS1 and Pompe disease.
Paula J Waters, Medical Genetics Service, Dept. Laboratory Medicine, Centre hospitalier universitaire de Sherbrooke (CHUS), Canada / Department of Pediatrics, Faculty of Medicine and Health Sciences, Université de Sherbrooke, Canada

BP-11 AT845 gene replacement therapy for late-onset Pompe disease: an update on safety and preliminary efficacy data from FORTIS, a phase 1/2 open-label clinical study
Chieri Hayashi, Astellas Gene Therapies, USA

BP-12 Investigation of in vivo gene therapy for CNS symptoms of GM1 gangliosidosis mediated by blood-brain barrier-penetrating enzymes for clinical application
Saki Matsushima, Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine, Japan

BP-13 Neuro-specific peptides in alpha-mannosidosis patients
Nato Vashakmadze, Research Institute of Pediatrics and Children's Health in Petrovsky National ResearchCentre of Surgery, Russia

BP-14 Modeling Niemann-Pick Disease Type C by Directly Converted Neurons from Urine-Derived Cells
Keita Matsumoto, Department of Physiology, Keio University School of Medicine, Japan

BP-15 Nonclinical pharmacology of JR-446, a novel blood-brain barrier penetrant A-N-acetylglucosaminidase
Jun Ito, JCR Pharmaceuticals Co., Ltd., Japan

BP-16 Alterations in mitochondrial energy metabolism and mitochondria-lysosome contact in MPS I and MPS II knockout cells
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil / PPG Ciências Biológicas: Fisiologia, Universidade Federal do Rio Grande do Sul, Brazil / PPG Neurociências, Universidade Federal do Rio Grande do Sul, Brazil

Evening Reception & Networking following Poster Session - Japan Night
19:00-20:30, Japanese Garden, located inside ICC Kyoto

Thursday | 4 September 2025

Registration
07:30-19:00, Event Hall

Industry-Sponsored Symposium
08:00-09:00, Room 3 (Room B-1)

Speed Mentoring
08:00-09:00, Lobby

Snack Service
08:30-09:30, Annex Hall / Event Hall / Swan

Posters
09:00-19:00, Annex Hall / Event Hall

Keynote Lecture 2
Chair: Kimitoshi Nakamura, Japan
09:15-10:00, Room 1 (Main Hall)

09:15 KL2 **Hypoxia as a therapy for mitochondrial disease**
Vamsi Krishna Mootha, Massachusetts General Hospital, Harvard Medical School, USA

Plenary Session 1
Chairs: Marshall Summar, USA
Barbara Yu, Singapore and UK
10:00-10:30, Room 1 (Main Hall)

10:00 PS1 **Empowering Biomedical Discovery with "AI Scientists"**
Marinka Zitnik, Harvard University, Harvard Medical School, USA

Plenary Session 2
Chair: Sean Froese, Switzerland
10:30-11:00, Room 1 (Main Hall)

10:30 PS2 **Harnessing allosteric regulation of metabolic enzymes for novel drug discovery**
Wyatt Yue, Newcastle University, UK

Coffee Break
10:30-11:00, Annex Hall / Event Hall / Swan

Plenary Session 3
Chair: Nicholas Ah Mew, USA
11:00-11:30, Room 1 (Main Hall)

11:00 PS3 Role of urea cycle flux studies for preclinical research
Johannes Häberle, University Children's Hospital Zurich, Switzerland

Plenary Session 4
Chair: Charles P. Venditti, USA
11:30-12:00, Room 1 (Main Hall)

11:30 PS4 The Future of Specific Advanced Treatments for Inborn Errors of Metabolism
Emil D. Kakkis, Ultragenyx, USA

Industry-Sponsored Symposium
12:30-13:30, Room 2 (Room A)

Industry-Sponsored Symposium
12:30-13:30, Room 3 (Room B-1)

Industry-Sponsored Symposium
12:30-13:30, Room 4 (Room B-2)

Industry-Sponsored Symposium
12:30-13:30, Room 5 (Room D)

Industry-Sponsored Symposium
12:30-13:30, Room 6 (Sakura)

JIMD Communicating Editors Meeting
12:30-13:30, Meeting Room M1 (Room C-1) (Closed)

Lunch Service
12:45-13:45, Event Hall / Swan

Industry-Sponsored Symposium
13:45-14:45, Room 3 (Room B-1)

Industry-Sponsored Symposium
13:45-14:45, Room 4 (Room B-2)

Industry-Sponsored Symposium
13:45-14:45, Room 5 (Room D)

Industry-Sponsored Symposium
13:45-14:45, Room 6 (Sakura)

Coffee with the Editors
14:30-15:15, Main Hall Lobby

Coffee Break
14:45-15:15, Annex Hall / Event Hall / Swan

Special Symposium 2: Patient Advocacy: How Quality of Care Impacts Quality of Life
Moderator: Karen Reznik Dolins, USA
Speakers: Kirsty Hoyle, Metabolic Support UK, UK,
Tresa Warner, National Urea Cycle Disorders Foundation, USA
15:00-17:00 Room 7 (Room E)

15:00 Presenter introductions & Workshop objectives
15:15 Presentations covering key topics "How Quality of Care Impacts Quality of Life"
16:15 Priority setting activity & interactive discussion

Symposium 5: Cutting-Edge Advances in Lysosomal Disorders
Chairs: Marc Patterson, USA
Nicola Brunetti-Pierri, Italy
15:15-17:30, Room 1 (Main Hall)

15:15 SP5-1 Beyond ERT/SRT : Brain, Lung, and Mesenteric Lymph Node Spatial-Omics Atlas Redefines Cellular Pathology and Informs Immunotherapy in nGD
Pramod K. Mistry, Yale University School of Medicine, USA

15:35 SP5-2 Lessons Learned in Pompe disease in the era of Newborn Screening
Priya Kishnani, Duke University Medical Center, USA

15:55 SP5-3 Mucopolysaccharidoses: Recent Advances and Future Prospects
Roberto Giugliani, UFRGS, HCPA, DASA, CASA DOS RAROS, Brazil

16:15 SP5-4 From Disease Discovery to Treatment in DEGS1 leukodystrophy
Aurora Pujol, Bellvitge Biomedical Research Institute (IDIBELL) , Spain

16:35 SP5-5 Advancing an Innovation Strategy for Lysosomal Storage Disorders through Digital Health and Data-Driven Collaboration
Maurizio Scarpa, Azienda Sanitaria Universitaria Friuli Centrale, Italy

16:55 SP5-6 Human iPSC-derived brain models to unravel disease mechanisms of neuronopathic lysosomal storage disorders
Isaac Canals, University of Zurich - Kinderspital Zürich, Switzerland

Symposium 6: Frontiers in Amino Acid Metabolism Disorders

Chairs: Tetsuya Ito, Japan
Philippa Mills, UK

15:15-17:15, Room 2 (Room A)

15:15 SP6-1 Cerebral creatine deficiency syndromes.
Nicola Longo, University of California Los Angeles (UCLA), USA

15:35 SP6-2 The biochemistry of nonketotic hyperglycinemia in a mouse and development of new treatments
Johan L.K. Van Hove, University of Colorado, USA

15:55 SP6-3 MSUD: Update on treatment and long-term follow-up.
Verónica Cornejo, Laboratory of Genetics and Metabolic Diseases, INTA, University of Chile, Chile

16:15 SP6-4 A protein-based engineering of human fibroblast-derived hepatic progenitors applicable for a gene-cell therapy to tyrosinemia type-1
Yukihito Ishizaka, Japan Institute for Health Security, Japan

16:35 SP6-5 Personalized therapeutic genome editing for hepatic inborn errors of metabolism
Rebecca Ahrens-Nicklas, The Children's Hospital of Philadelphia and University of Pennsylvania, USA

Oral 16: Fatty Acid Oxidation and Ketone Body Disorders

Chairs: Hironori Kobayashi, Japan
Ute Spiekerkoetter, Germany

15:15-17:15, Room 3 (Room B-1)

15:15 OP16-1 Adult-Diagnosed Fatty Acid Oxidation Disorders Identified by Acylcarnitine Analysis: Diagnostic Cues and Clinical Characteristics from a Japanese Single-Center Retrospective Study
Hironori Kobayashi, Laboratory Division,Shimane University Hospital, Japan

15:30 OP16-2 Rate of LCHADD chorioretinopathy progression in a prospective cohort of 40 participants.
Melanie B Gillingham, Oregon Health & Science University, USA

15:45 OP16-3 Ceramide-The Unmasked Driver of Heart Failure In Long-Chain Fatty Acid Oxidation Disorders (LC-FAODs)
Marie Kristine Norris, University of Utah, USA

16:00 OP16-4 Adipyl- and methylmalonyl-D-dipeptide conjugates normalize lysine succinylation and improve bioenergetics in fatty acid β-oxidation deficient patient cells
Al-Walid Mohsen, Dept of Pediatrics, School of Medicine, University of Pittsburgh, USA / Dept of Human Genetics, School of Public Health, University of Pittsburgh, USA

16:15 OP16-5 PPARδ agonists improve protein lysine succinylation and bioenergetics in the absence of glucose and ameliorate elevated ROS in VLCAD and LCHAD deficient patients' fibroblasts
Anuradha Karunanidhi, Dept of Pediatrics, School of Medicine, University of Pittsburgh, USA

16:30 OP16-6 Discovery and Thermal Shift-guided SAR analysis of a novel class of pharmacological chaperones targeting MCAD Deficiency
Lydia Gerber, University Children's Research, UCR@Kinder-UKE, University Medical Center Hamburg-Eppendorf, Germany / Organic Chemistry, Department of Chemistry, Faculty of Mathematics, Informatics and Natural Sciences, Universität Hamburg, Germany / German Center for Child and Adolescent Health (DZKJ), Germany

16:45 OP16-7 Gene therapy for long-chain hydroxyacyl-coA dehydrogenase deficiency
Paul Schurmann, University Medical Center Utrecht, Netherlands

17:00 OP16-8 Comparison of treatment efficacy of very long-chain acyl-CoA (VLCAD) deficiency with an AAV9.hVLCAD vector, synthetic VLCAD mRNA, and triheptanoin in a mouse model
Jerry Vockley, University of Pittsburgh School of Medicine, USA

Oral 17: Emerging Trends in IEMs

Chairs: Ee Shien Tan, Singapore
Hudson H. Freeze, USA

15:15-16:45, Room 4 (Room B-2)

15:15 OP17-1 Long-term clinical outcomes of patients with AHP who were not attack-free after 6 months of givosiran treatment: subgroup analysis of the phase 3 ENVISION study
Eliane Sardh, Centre for Inherited Metabolic Diseases, Porphyria Centre Sweden, Department of Molecular Medicine and Surgery, Karolinska Institutet, Karolinska University Hospital, Sweden

15:30 OP17-2 A Genome-Wide Association Study Identifies GRHL2 as a Potential Modifier of Severity of Photosensitivity in Erythropoietic Protoporphyria
Isabella Suijker, Porphyria Expert Center Rotterdam, Department of Internal Medicine, Erasmus University Medical Center, Netherlands

15:45 OP17-3 The CRISPR-Cas9 knockout DDC SH-SY5Y in vitro model for AADC deficiency provides insight into two severe catalytic variants: a cross-sectional structural and functional analysis
Mariarita Bertoldi, University of Verona, Department of Neuroscience, Biomedicine and Movement Sciences, Italy

16:00 OP17-4 tRNA at the Frontier: Developing Medicines to Treat Stop Codon Disease
Nerissa C. Kreher, Alltrna, USA

16:15 OP17-5 Maternal health outcomes in ornithine transcarbamylase deficiency: A comparative analysis of pregnancies in symptomatic and asymptomatic heterozygotes
Margo Sheck Breilyn, Icahn School of Medicine at Mount Sinai, USA

16:30 Withdrawal

Oral 18: Carbohydrate Disorders

Chairs: Sunita Bijarnia-Mahay, India
Yoichi Wada, Japan

15:15-17:15, Room 5 (Room D)

15:15 OP18-1 Results from a pivotal phase 3 double-blind placebo-controlled trial of DTX401 for the treatment of individuals with glycogen storage disease type Ia (GSDIa)
John J. Mitchell, Montreal Children's Hospital, Canada

15:30 OP18-2 Bempedoic acid prolongs fasting time in patients with glycogen storage disease type 1a
Anibh Martin Das, Hannover Medical School, Germany

15:45 OP18-3 Empagliflozin in Chinese GSDIb Patients: Efficacy, Safety, and Machine Learning-Assisted Dose Adjustment Model
Yu Xia, Department of Pediatric Endocrinology and Genetic Metabolism, Xinhua Hospital, Shanghai Institute of Pediatric Research, School of Medicine, Shanghai Jiao Tong University, China

16:00 OP18-4 New Avenues to Treat Neutropenia in Glycogen Storage Disease type Ib (GSD1b) and G6PC3-deficient Patients.
Maria Veiga-da-Cunha, de Duve Institute and UCLouvain, Belgium

16:15 OP18-5 **Improving dietary protocols in Glycogenosis type III: experience from an Italian cohort**
Marco Scaglione, Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health, University of Genova, Italy

16:30 OP18-6 **Hepatic glycogen storage disease type IX: long-term outcomes in the UK from 91 patients**
Rebecca K Halligan, Department of Paediatric Inherited Metabolic Diseases, Evelina London Children's Hospital, UK

16:45 OP18-7 **Galactose Response to Lactose Intake in Four Japanese Children with GALM Deficiency**
Chika Takano, Department of Pediatrics and Child Health, Nihon University School of Medicine, Japan / Division of Microbiology, Department of Pathology and Microbiology, Nihon University School of Medicine, Japan

17:00 OP18-8 **Preserving Fertility in Classic Galactosemia: Window of Opportunity and Gene Therapy Solutions**
E. Naomi Vos, MosaKids Children's Hospital, Maastricht University Medical Centre+, Netherlands / GROW, Maastricht University, Netherlands / United for Metabolic Diseases (UMD), Netherlands / European Reference Network for Hereditary Metabolic Disorders (MetabERN), Italy / Department of Clinical Genetics, Maastricht University Medical Centre+, Netherlands

Special Symposium 3: Facing complexity in metabolic nutrition and dietetics globally

Moderators: Julio Cesar Rocha, Portugal
Anita MacDonald, UK

15:15-17:15, Room 6 (Sakura)

15:15 SSP3-1 **Medium-chain fatty acid therapy for citrin deficiency.**
Chikahiko Numakura, Saitama Medical University, Japan

15:45 SSP3-2 **Nutritional treatment in 12 Chilean patients older than 6 years with glutaric aciduria type 1: metabolic, anthropometric, and neurological outcomes.**
Felipe Penaloza, INTA, Universidad de Chile, Chile

16:05 SSP3-3 **Parenteral nutrition (PN) use during acute illness in children with inherited metabolic disorders (IMD)**
Jennifer Dunne, Dietetics and Metabolic Medicine, Great Ormond Street Hospital for Children, UK

16:25 SSP3-4 **Nutritional Counseling and optimization of metabolic control in phenylketonuria**
Sara Giorda, Department of Pediatrics, University of Torino, Italy

16:45 SSP3-5 **ADHD incidence in PKU patients and its impact on dietary management**
Angela Harris, Royal Children's Hospital, Australia

17:05 SSP3-6 **The challenge of dietary assessment in PKU: Rethinking new measures for research**
Rani H Singh, Emory University, USA

Recon4IMD Clinical Connect F2F

16:45-18:00, Meeting Room M1 (Room C-1) (Closed)

SIMD Membership/Business Meeting

17:30-19:30, Room 7 (Room E) (Closed/ only for SIMD Members)

Galactosemia Network (GalNet) symposium

17:30-19:30, Meeting Room M2 (RoomC-2) (Closed/ only for GalNet members)

Poster 3: Best Poster Award

Chairs: Tomoko Nakanishi, Japan
Yew Sing Choy, Malaysia

17:30-18:30, Exhibition & Poster 2 (Event Hall)

BP-17 Non-clinical Safety Evaluation of Ex Vivo Gene Therapy for Hunter Disease Using Mouse Model
Takashi Higuchi, The Jikei University, Japan

BP-18 Pathological analysis of sialidosis /galactosialidosis model mice and gene therapy
Jun Tsukimoto, Graduate School of Pharmaceutical Sciences, Tokushima University, Japan

BP-19 Arimoclomol upregulates expression of genes belonging to the coordinated lysosomal expression and regulation (CLEAR) network
Hadeel Shammas, Zevra Therapeutics Inc., USA

BP-20 OXA1L deficiency leads to mitochondrial myopathy via ROS mediated NFkB signaling pathway
Yongkun Zhan, Department of Clinical Genetics Center, Shanghai Institute for Pediatric Research, Xinhua Hospital affiliated to Shanghai Jiao Tong University School of Medicine, China

BP-21 Withdrawal

BP-22 Pilot study for rapid gene panel test in neonates with abnormal newborn screening result of inherited metabolic diseases
Jinsup Kim, Department of Pediatrics, Barowell Hospital, Republic of Korea

BP-23 The use of a second-tier method, combining citrulline levels and gene analysis, significantly improved the detection rate of Citrin deficiency in newborn screening
Hsiao-Jan Chen, The Chinese Foundation of Health, Taipei, Taiwan

BP-24 Newborn CAH screening using a single 17-OHP cut-off value in the primary screening followed by a 2nd tier steroid assay
James Soon Chuan Lim, Biochemical genetics and national expanded newborn screening programme, KK Women's and Children's Hospital, Singapore

Poster 4: Best Poster Award

Chairs: Simon Allan Jones, UK
Nithiwat Vatanavicharn, Thailand

17:30-18:30, Exhibition & Poster 2 (Event Hall)

BP-25 Transcriptomic analyses of gene expression following CRISPR/Cas9 knockout of ACY1 in HepG2 cells.
Lil Klaas, Research Group Inborn Errors of Metabolism, Department of Natural Sciences & Institute for Functional Gene Analytics (IFGA), Bonn-Rhein-Sieg University of Applied Sciences, Germany

BP-26 Emerging Non-Canonical Pathogenic Variants Disrupting Chromatin Architecture in Inborn Errors of Metabolism
Belen Perez, Centro de Diagnóstico de Enfermedades Moleculares, Centro de Biología Molecular Severo Ochoa, CIBERER, IdiPAZ. Universidad Autónoma de Madrid., Spain

BP-27 Cholesterol ester accumulation in ABCD1-deficient HeLa cells
Masashi Morita, Graduate School of Medicine and Pharmaceutical Sciences, University of Toyama, Japan

BP-28 Decreased Bone Density in Phenylketonuria: PHEFREE- The National Institutes of Health Rare Disease Consortium for Hyperphenylalaninemia
Georgianne Lee Arnold, University of Pittsburgh Medical Center, USA

BP-29 Neuroinflammation in brain of rats submitted to chronic chemically-induced model of hyperphenylalaninemia: L-carnitine protection
Jessica Lamberty Faverzani, Universidade Federal do Rio Grande do Sul, Brazil / Serviço de Genetica Medica, Hospital de Clinicas de Porto Alegre, Brazil

BP-30 Initial Findings from the First Four Patients Enrolled in OTC-HOPE Clinical Trial: No Hyperammonemic Events in First Participant to Complete 24-week Study
Julien Baruteau, Great Ormond Street Hospital for Children, UK

BP-31 Modeling, characterization and therapeutic screening of Citrin deficiency
Toni Vukovic, University Children's Hospital, Switzerland

BP-32 Short-term Therapeutic Effects of UX007 (Triheptanoin) in Citrin Deficiency Mouse Model
Eri Imagawa, Department of Pediatrics, The Jikei University School of Medicine, Japan

Friday | 5 September 2025

Registration
07:30-18:00, Event Hall

Industry-Sponsored Symposium
08:00-09:00, Room 3 (Room B-1)

Speed Mentoring
08:00-09:00, Lobby

Snack Service
08:30-09:00, Annex Hall / Event Hall / Swan

Posters
09:00-17:00, Annex Hall / Event Hall

Keynote Lecture 3
Chair: Fumio Endo, Japan
09:15-10:05, Room 1 (Main Hall)

09:15 KL3 Molecular mechanisms and physiological roles of autophagy
Noboru Mizushima, The University of Tokyo, Japan

Coffee Break
10:00-10:30, Annex Hall / Event Hall / Swan

Symposium 7: Frontiers in Organic Acidemias and Fatty Acid Oxidation Disorders
Chairs: Chong Kun Cheon, Republic of Korea
Shirou Matsumoto, Japan
10:15-11:55, Room 1 (Main Hall)

10:15 SP7-1 Comparison of treatment efficacy of very long-chain acyl-CoA (VLCAD) deficiency with an AAV9.hVLCAD vector, synthetic VLCAD mRNA, and triheptanoin in a mouse model
Jerry Vockley, University of Pittsburgh School of Medicine, USA

10:35 SP7-2 Benefits and limitations of newborn screening for organic acidemias and fatty acid oxidation disorders
Stefan Kölker, Heidelberg University Hospital and Heidelberg University, Medical Faculty, Centre for Pediatrics and Adolescent Medicine, Germany

10:55 SP7-3 Carglumic Acid Shows Long-Term Benefits in Organic Acidurias: Evidence Supports Early and Ongoing Treatment
Sufin Yap, Sheffield Children's Hospital, UK

11:15 SP7-4 Unraveling energetic dysregulation and rewiring in methylmalonic aciduria
Sean Froese, University Children's Hospital Zurich, Switzerland

11:35 SP7-5 New Insights into the Pathophysiology of isolated MMA
Charles P Venditti, National Institutes of Health, USA

Symposium 8: Advances in iPS Cell Research
Chairs: Hiroshi Kobayashi, Japan
Andrea Dardis, Italy
10:15-11:45, Room 2 (Room A)

10:15 SP8-1 Targeting Cholesterol Biosynthesis in ALS: Insights from iPSC-Derived Motor Neurons and Polygenic Risk Analysis
Hideyuki Okano, Keio University Regenerative Medicine Research Center (KRM), Japan

10:35 SP8-2 The iPSC Neurodegeneration Initiative: Integrating Patient Cohorts and iPSC Models
Haruhisa Inoue, Center for iPS Cell Research and Application (CiRA), Kyoto University, JAPAN

10:55 SP8-3 Studying for abnormal phenotype and drug development in ganglioside-accumulated disease
Takumi Era, Department of Cell Modulation, Institute of Molecular Embryology and Genetics, Kumamoto University, Japan

11:15 SP8-4 The neurological roles of G-quadruplexes in iPS neurons derived from ATR-X syndrome
Norifumi Shioda, Kumamoto University, Japan

Oral 19: Innovative Therapies
Chairs: Yohta Shimada, Japan
Johanna M.P. van den Hout, Netherlands
10:15-11:45, Room 3 (Room B-1)

10:15 OP19-1 Engineering Targeted Lipid Nanoparticles for Bone Marrow Delivery of Base and Prime Editors
Jose Castro-Alpizar, Metabolic Diseases, Division Pediatrics Wilhemina Children's Hospital, University Medical Center Utrecht, Netherlands

10:30 OP19-2 Development of a clinical product based on autologous genome-edited hematopoietic stem and progenitor cells to treat Mucopolysaccharidosis type 1
Edina Poletto, Stanford University, USA

10:45 OP19-3 CAMPSIITE phase I/II/III: Interim clinical update of clemidsogene lanparvovec (RGX-121), an investigational gene therapy for treatment of neuronopathic mucopolysaccharidosis type II (MPS II)
Roberto Giugliani, Medical Genetics Service, HCPA, Brazil

11:00 OP19-4 AMT-191 investigational gene therapy in adult males with classic Fabry disease; initial safety and biomarker results of phase 1/2 study
Maryam Banikazemi, NYC Health + Hospitals, USA

11:15 OP19-5 Lessons learned from the first-in-human homologous recombination gene editing clinical study in pediatric patients with methylmalonic acidemia
Jirair Krikor Bedoyan, UPMC Children's Hospital of Pittsburgh, USA / University of Pittsburgh School of Medicine, USA

11:30 OP19-6 Extensive metabolic detoxification and favorable clinical outcomes sustained through 5 years post-treatment in Hematopoietic Stem Cell Gene Therapy (OTL-203) for Mucopolysaccharidosis Type I-Hurler
Francesca Tucci, San Raffaele Telethon Institute for Gene Therapy (SR-Tiget), Italy

Oral 20: Lipid and Peroxisomal Disorders
Chairs: Motomichi Kosuga, Japan
Yoriko Watanabe, Japan
10:15-11:45, Room 4 (Room B-2)

10:15 OP20-1 **Evaluation of Clinical, Radiological, and Genetic Features in Cases of X- Linked Adrenoleukodystrophy**
Sebnem Kilic, Istanbul Univercity, Istanbul Faculty of Medicine Pediatric Nutrition and Metabolism Department, Turkey

10:30 OP20-2 **Shifts in Positive Results for Smith Lemli Opitz Syndrome Reflect early Detection of Fetal Fentanyl Syndrome**
Silvia Tortorelli, Mayo Clinic, USA

10:45 OP20-3 **When Blood Turns Milky: Lipemia as a Gateway to Diagnosing Pediatric IEMs - A Case Series from a Tertiary Care Pediatric Hospital in Sri Lanka**
Nishani Matara Mahavidanage De Silva, Lady Ridgeway Hospital for Children- Department of chemical pathology, Sri Lanka

11:00 OP20-4 **Mind the gap- Can current homozygous familial hypercholesterolaemia management achieve therapeutic low-density lipoprotein cholesterol target levels in paediatric patients?**
Mike Champion, Paediatric Inherited Metabolic Medicine, Evelina London Children's Hospital, UK

11:15 OP20-5 **Safety and tolerability of chenodeoxycholic acid in pediatric patients with cerebrotendinous xanthomatosis (RESTORE): An open-label phase 3 study**
Vikram Prakash, Arnold Palmer Children's Hospital and Orlando Health, USA

11:30 OP20-6 **Modeling Rare Metabolic Disease Using Humanized Yeast and CRISPR Myoblasts to Investigate TANGO2 Function**
Aaliya Naaz, Concordia University, Canada

Oral 21: Phenylketonuria 2
Chairs: Cary O. Harding, USA
Shoji Yano, USA
10:15-11:45, Room 5 (Room D)

10:15 OP21-1 **AMPLIPHY: A Phase 3 Study Comparing the Efficacy and Safety of Sepiapterin and Sapropterin in Children and Adults with Phenylketonuria**
Anita Inwood, Queensland Lifespan Metabolic Medicine Service, Queensland Children's Hospital, Australia

10:30 OP21-2 **Pegvaliase treatment in 18 adult patients with classical phenylketonuria, part 1: efficacy, patterns of responsiveness and complement dynamics**
Erika Ogawa, Department of Pediatrics and Child Health, Nihon University School of Medicine, Japan / Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital, Japan

10:45 OP21-3 **Safety and Efficacy of Pegvaliase in Adolescents with Phenylketonuria: Primary Results from PEGASUS, a Phase 3 Open-label Randomized Controlled Study**
Stephanie Sacharow, Boston Children's Hospital, USA

11:00 OP21-4 **Effect of Long-Term Sepiapterin Treatment on Dietary Phenylalanine Tolerance in Japanese Participants with Phenylketonuria: Interim Results from the APHENITY Extension Study**
Takashi Hamazaki, Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine, Japan

11:15 OP21-5 Use of Phenylalanine-Free L-Amino Acid Mixtures During Illness Episodes in BH4-Responsive Phenylketonuria Patients on Unrestricted Diet:A Retrospective Follow-Up Study
Selin Akbulut, İstanbul University-Cerrahpaşa, Turkey

11:30 OP21-6 A prolonged release compared to an amino acid protein substitute in classical PKU effect on morning phenylalanine and tyrosine concentrations
Anne Daly, Department of Dietetics, Birmingham Women's and Children's Hospital, UK

Oral 22: Nursing in Metabolism

Chairs: Anita Inwood, Australia
Ina Knerr, Ireland

10:15-11:45, Room 6 (Sakura)

10:15 OP22-1 Seasonal fluctuations in disease-specific quality of life among adult patients with Fabry disease: an observational study
Yuta Koto, Faculty of Nursing, Kansai Medical University, Japan

10:30 OP22-2 The Role of Nursing in Multidisciplinary Clinic for Inborn Errors of Metabolism: Delivering Holistic Patient Care
Souwaluck Ratanamalaya, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand

10:45 OP22-3 30 Years of Metabolic Nursing Experience at the National Centre for Inherited Metabolic Disorders in Dublin, Ireland
Maria Christine O Regan Dinnan, CHI At Temple Street, Ireland

11:00 OP22-4 Human milk feeding in inherited metabolic disorders: a systematic review of clinical outcomes, practices, challenges and facilitators
Alexander Hoeller, Division of Nutrition and Dietetics, University Hospital Innsbruck, Austria

11:15 OP22-5 Exploring Family Perspectives on Home vs. Inpatient Profiling for Children with a Ketotic Glycogen Storage Disease - A Single UK Centre Experience
Jordan Cox, Evelina London Children's Hospital, UK

11:30 OP22-6 Evaluating the Impact of Orthotopic Liver Transplant (OLT) on Quality of Life (QoL) for 15 Children with Metabolic Disorders and Their Families
Catherine Atthow, Queensland Lifespan Metabolic Medicine Service, Australia

Oral 23: Newborn Screening 2

Chairs: Wendy Smith, USA
Yin-Hsiu Chien, Taiwan

10:15-11:45, Room 7 (Room E)

10:15 OP23-1 Six years of application of a highly multiplexed 2nd tier test to improve performance metrics in Newborn Screening Program
Michela Perrone Donnorso, LABSIEM-Pediatric Clinic Unit-DINOGMI, University of Genova, IRCCS Istituto Giannina Gaslini, Italy

10:30 OP23-2 Breaking Barriers in Newborn Screening: Reactomics as a Game-Changer for Inherited Metabolic Disorders Detection
Raquel Yahyaoui, Hospital Regional Universitario de Malaga., Spain / Instituto de Investigacion Biomedica de Málaga. IBIMA-Plataforma BIONAND., Spain

10:45 OP23-3 DEVELOPMENT AND VALIDATION OF MULTI-OMIC NEWBORN BLOODSPOT SCREENING IN A SOUTH AUSTRALIAN COHORT, THE NEWBORNSINSA STUDY
Drago Bratkovic, Women's and Children's Hospital, Australia

11:00 OP23-4 A Multiplexed LC-MS/MS Method With Built-In Reflex Capabilities Allows For Rapid and Comprehensive Newborn Screening Results While Increasing Testing Capacity.
Craig Houghton Seymour, Mayo Clinic, USA

11:15 OP23-5 Global Updates on Taiwan's Exclusive Newborn Screening for Mucopolysaccharidosis Type IVA
Hsiang-Yu Lin, Department of Pediatrics, MacKay Memorial Hospital, Taiwan / International Rare Disease Center, MacKay Memorial Hospital, Taiwan / Department of Medicine, Mackay Medical College, Taiwan / Department of Medical Research, MacKay Memorial Hospital, Taiwan / Mackay Junior College of Medicine, Nursing and Management, Taiwan

11:30 OP23-6 California's three–tiered approach to MPS screening: A comprehensive analysis of MPS I and MPS II implementation and outcomes
Rajesh Sharma, Genetic Disease Screening Program, California Department of Public Health, USA

Industry-Sponsored Symposium
12:00-13:00, Room 2 (Room A)

Industry-Sponsored Symposium
12:00-13:00, Room 3 (Room B-1)

Industry-Sponsored Symposium
12:00-13:00, Room 4 (Room B-2)

Industry-Sponsored Symposium
12:00-13:00, Room 5 (Room D)

Industry-Sponsored Symposium
12:00-13:00, Room 6 (Sakura)

Lunch Service
12:15-13:15, Event Hall / Swan

Oral 24: Lysosomal Disorders 4
Chairs: Roberto Giugliani, Brazil
Torayuki Okuyama, Japan
13:30-15:00, Room 1 (Main Hall)

13:30 OP24-1 Intracerebroventricular enzyme replacement therapy in patients with neuronopathic mucopolysaccharidosis type II: Final report of 5-year results from a Japanese open-label phase 1/2 study
Joohyun Seo, Department of Clinical Genomics, Saitama Medical University, Japan

13:45 OP24-2 Long-term (up to 5 years) efficacy of pabinafusp alfa on neurocognition in patients with mucopolysaccharidosis type II: a pooled, post hoc analysis of clinical trials
Roberto Giugliani, Universidade Federal do Rio Grande do Sul and Hospital de Clinicas de Porto Alegre, Brazil / Casa dos Raros, Brazil

14:00 OP24-3 Risk and clinical characteristics of spinal cord compression across different mucopolysaccharidosis types: A retrospective cohort study
Minji Im, Samsung Medical Center, Republic of Korea

14:15 OP24-4 MORQUIO A DISEASE: UNDERSTANDING THE EYE PATHOLOGY THROUGH GLYCOSAMINOGLYCAN DISTRIBUTION
Adriana M Montano, Saint Louis University, USA

14:30 OP24-5 Allogeneic Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis IV A (Morquio A): An international retrospective study of 40 children
Sandhya Kharbanda, University of California, USA

14:45 OP24-6 Early Diagnosis and Intervention of Mucopolysaccharidosis in Infants diagnosed by Expanded Newborn Screening: Enzyme Replacement Therapy and Cord Blood Transplantation in Three Case
Hirotoshi Sakaguchi, Children's Cancer Center, National Center for Child Health and Development, Japan

Oral 25: Mitochondrial Disorders 2

Chairs: Shamima Rahman, UK
Kei Murayama, Japan

13:30-15:00, Room 2 (Room A)

13:30 OP25-1 Impaired bioenergetics in FBXL4-deficient fibroblasts is restored by aminolevulinate and iron (ALA/Fe)
Alexandra Latini, CHOC Children's Hospital, USA / LABOX - Universidade Federal de Santa Catarina, Brazil

13:45 OP25-2 Identification of a De Novo RNU4-2 Variant Implicated in Spliceosomal Dysfunction and Mitochondrial Disease
Kohta Nakamura, Diagnostics and Therapeutics of Intractable Diseases, Intractable Disease Research Center, Graduate School of Medicine, Juntendo University, Japan

14:00 OP25-3 Role of BOLA3 in the Mitochondrial Fe-S Cluster Clarified by Metabolomic Analysis
Hiroyuki Iijima, National Center for Child Health and Development, Japan

14:15 OP25-4 Nicotinamide riboside and L-serine treatment improves objective clinical performance in methionyl-tRNA formyltransferase deficiency (MTFMT)
Johan LK Van Hove, University of Colorado, USA

14:30 OP25-5 Isotope tracing reveals anaplerotic mechanisms of medium-chain fatty acids in malate dehydrogenase 2 deficiency
Judith J.M. Jans, Dept. Genetics, University Medical Center Utrecht, Netherlands

14:45 OP25-6 Oral Supplementation with NMN Improves Cardiac Function in Aged Mice by Promoting Sirt1 Activation and Enhancing Mitochondrial Function
Yuya Kinoshita, Department of Pediatrics, Kumamoto University Hospital, Japan / Department of Molecular Genetics, Kumamoto University, Japan

Oral 26: Clinical Studies and Patient-Reported Outcomes 2

Chairs: Irene De Biase, USA
Hsiang-Yu Lin, Taiwan

13:30-15:00, Room 3 (Room B-1)

13:30 OP26-1 Leriglitazone Achieved a Primary Endpoint Based on Disease Arrest in Patients with Childhood Cerebral Adrenoleukodystrophy in the NEXUS Open-Label Phase 2/3 Study
Caroline Sevin, Pediatric Neurology Department, Reference Center for Leukodystrophies, Assistance Publique-Hôpitaux de Paris, Hôpitaux Universitaires Paris Saclay, Bicêtre Hospital, Le Kremlin Bicêtre, France

13:45 OP26-2 Women With X-Linked Adrenoleukodystrophy: Disease Burden and Risk of Cerebral Disease
Florian S. Eichler, Massachusetts General Hospital, USA

14:00 OP26-3 Trends and outcomes of hospital admissions for inherited metabolic disorders at a single quaternary care center
Maria Paula Silva, Division of Genetics, Genomics, and Metabolism, Ann & Robert H. Lurie Children's Hospital of Chicago, USA / Department of Pediatrics, Northwestern University Feinberg School of Medicine, USA

14:15 OP26-4 Report on two decades of activity of the Brazilian Information Service on Inborn Errors of Metabolism
Mariana Lima Scortegagna, Postgraduate Program in Clinical Sciences, UFRGS, Brazil / Casa dos Raros, Brazil / SIEM, Information Service on Inborn Errors of Metabolism, UFRGS, Brazil

14:30 OP26-5 Prevalence and Characterization of Eating Disorders among Patients with Inborn Errors of Metabolism Managed with Diet
Mary Kate LoPiccolo, Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, USA

14:45 OP26-6 Long term effects of cholic acid therapy in children with defects of primary bile acid synthesis
Virginija Bambalaite, Theravia, France

Oral 27: Inborn Errors of Metabolism in Adults

Chairs: Mitsuru Kubota, Japan
Ken Sakurai, Japan

13:30–15:00, Room 4 (Room B-2)

13:30 OP27-1 Assesment of Cardiovascular Risk in Early-Treated Adult Patients with Phenylketonuria – a Comparative Population-based Study in Hungary
Kata Rebeka Utassy, Department of Internal Medicine and Oncology, Semmelweis University, Hungary / Faculty of Medicine, Semmelweis University, Hungary

13:45 OP27-2 Hepatic outcomes in adult patients with glycogen storage disease type III: a multi-centre UK cohort study
Kevin Kuriakose, Mark Holland Metabolic Unit, Salford Royal Hospital, UK

14:00 OP27-3 Serum high–sensitivity troponin to detect Fabry Cardiomyopathy
Subadra Wanninayake, Department of Inherited Metabolic Disorders, Queen Elizabeth Hospital Birmingham, UK

14:15 OP27-4 Biliary Lithiasis in Gaucher Disease: Insights on Risk Factors
Lucas Ferreira Teixeira, School of Medicine, Universidade Federal do Rio Grande do Sul, Brazil

14:30 OP27-5 Bone involvement in acid sphingomyelinase deficiency (ASMD)
Andrea Bordugo, Regional Centre for Rare Diseases, Azienda Ospedaliera Friuli Centrale-ASUFC, Italy

14:45 OP27-6 Clinical Outcomes in Adult Survivors of Nonketotic Hyperglycinemia: Insights from a Patient Registry
Brian Shayota, University of Utah, USA / Primary Children's Center for Personalized Medicine, USA

Oral 28: Amino Acid Disorders 2
Chairs: Ina Knerr, Ireland
Risto J. Lapatto, Finland
13:30-15:00, Room 5 (Room D)

13:30
OP28-1 The functional analysis of the variants of human glutamine synthetase
Hideki Matsumoto, Department of Pediatrics, Graduate School of Medicine, Gifu University, Japan

13:45
OP28-2 Investigation of miRNAs Associated with Nonketotic Hyperglycinemia
Harun Bayrak, Department of Molecular Medicine, Graduate School of Health Sciences, TOBB University of Economics and Technology, Turkey

14:00
OP28-3 Tuberous sclerosis complex as a new form of lysine-related inborn errors of metabolism
Felix Chan, University of Birmingham, UK / Birmingham Center for Neurogenetics, University of Birmingham, UK / Birmingham Center for Human Brain Health, University of Birmingham, UK

14:15
OP28-4 Low-Dose Nitisinone Therapy in Hereditary Tyrosinemia Type I: A Cost-Effective and Clinically Effective Approach
Sudipto Das, Division of Genetics, Department of Pediatrics, AIIMS, India

14:30
OP28-5 Evaluation of Nitisinone Treatment in a Cohort of Adult Patients with Alkaptonuria
Luis Baena-Ariza, Hospital Universitario Virgen del Rocio, Spain

14:45
OP28-6 Alkaptonuria and Nitisinone: A Cross-sectional Study of Patients in Germany
Anibh Martin Das, Hannover Medical School, Germany

Oral 29: Glycosylation and Protein Modification Disorders
Chairs: Takashi Hamazaki, Japan
Tamas Kozicz, USA
13:30-15:00, Room 6 (Sakura)

13:30
OP29-1 Predicting disease-overarching therapeutic approaches for Congenital Disorders of Glycosylation using multi-OMICS
Irena Josephina Johanna Muffels, Icahn School of Medicine at Mount Sinai, USA

13:45
OP29-2 Exploring Post-Translational Modifications of Laminin-211 in Cardiac Architecture: Insights from Inherited Phosphoglucomutase-1 Deficiency
Bijina Balakrishnan, Division of Medical Genetics, Department of Pediatrics, University of Utah, USA

14:00
OP29-3 Restoring Glycosylation and Metabolic Function in PMM2-CDG Human Cortical Organoids via GSK Inhibition
Rameen Shah, Icahn School of Medicine at Mount Sinai, USA

14:15
OP29-4 Discover intermediate metabolism in congenital disorders of glycosylation
Miao He, Children's Hospital of Philadelphia, USA / University of Pennsylvania, USA

14:30
OP29-5 Accumulation of UFMylated RPL26 and enhanced UFL1 interaction due to hypomorphic bi-allelic UFC1 gene variants provide new insights in causing profound infantile encephalopathy
Sunita Bijarnia-Mahay, Sir Ganga Ram Hospital, India

14:45
OP29-6 Preliminary results from an ongoing phase 2, open-label study evaluating the effects of GLM101 on ataxia improvement, safety, tolerability, and pharmacokinetics in patients with PMM2-CDG
Mercedes Serrano, Hospital Sant Joan de Deu, Spain

Model Organisms in Inherited Metabolic Disease Research
13:30-15:00, Room 7 (Room E) (Registration at MO-IMD)

Networking Evening
18:30-20:30, Kyoto Railway Museum

Networking Evening
19:30-21:30, THE SODOH HIGASHIYAMA KYOTO

Registration
08:30-12:45, Event Hall

Memorial Session for Prof. Toshiyuki Fukao: The Global Path of Ketone Body Metabolism Research
Chairs: Hiroyuki Ida, Japan
Grant A Mitchell, Canada
8:30 - 10:00, Room 5 (Room D)

8:30 MMS1-1 Memory of Prof. Toshiyuki Fukao — His remarkable contributions to inherited metabolic disorders and their legacy
Kimitoshi Nakamura, Kumamoto University, Japan

8:40 MMS1-2 A clinical approach to IEMs of ketone body metabolism
Grant A Mitchell, CHU Sainte-Justine, University of Montreal, Canada

9:10 MMS1-3 Continuing and Expanding the Scope of Research on Ketone Body Metabolism
Hideo Sasai, Department of Pediatrics / Department of Early Diagnosis and Preventive Medicine for Rare and Intractable Pediatric Diseases, Graduate School of Medicine, Gifu University, Japan

9:30 MMS1-4 The Future of Ketone Body Research
Jörn Oliver Sass, Bonn-Rhein-Sieg-University of Applied Sciences, Germany

Snack Service
08:30-09:00, Annex Hall / Event Hall / Swan

Posters
09:00-12:00, Annex Hall / Event Hall

Plenary Session 5
Chair: Kei Murayama, Japan
09:00-09:30, Room 1 (Main Hall)

09:00 PS5 Finding mitochondrial disease-associated proteins through systems biochemistry
David J. Pagliarini, HHMI/Washington University School of Medicine in St. Louis, USA

Plenary Session 6
Chair: Takashi Hamazaki, Japan
09:30-10:00, Room 1 (Main Hall)

09:30 PS6 Long-Term Efficacy of Genome Editing in Infant Mice With Glycogen Storage Disease Type Ia
Dwight Koeberl, Duke University School of Medicine, USA

Plenary Session 7
Chair: Eva Morava, USA
10:00-10:30, Room 1 (Main Hall)

10:00 PS7 Mucolipidosis II-related diseases: impact of the mannose 6-phosphate pathway
Thomas Braulke, University Medical Center Hamburg-Eppendorf; Institute of Osteology & Biomechanics; Cell Biology of Rare Diseases, Germany

Coffee Break
10:00-10:30, Annex Hall / Event Hall / Swan

Symposium 9: Late-Breaking Research
Chairs: Risto J. Lapatto, Finland
Andreas Schulze, Canada
10:30-12:00, Room 1 (Main Hall)

10:30 SP9-1 Stalling of Oligodendrocyte Progenitor Differentiation by Guanidino Compounds Is the Cause of Dysmyelination in Arginase Deficiency
Gerry Lipshutz, University of California, USA

10:45 SP9-2 Sustained biochemical correction and improved neurological outcomes at 36-months post hematopoietic Stem Cell Gene Therapy for Sanfilippo Disease
Simon Allan Jones, Manchester University NHS Foundation Trust, UK

11:00 SP9-3 Mitochondrial disease enhances influenza pathogenesis by upregulating de novo sialic acid biosynthesis
Peter J. McGuire, National Human Genome Research Institute, NIH, USA

11:15 SP9-4 Novel approach to safe PKU therapy: development of an adenovirus vector for cleavage-free genomic knock-in by Cas9 nickase-based gene editing realizing "one disease, one vector"
Tomoko Nakanishi, Center for Biomedical Research Resources, Juntendo University Graduate School of Medicine, Japan

11:30 SP9-5 Unifying neuronopathic IEMs under childhood dementia through an international Delphi study
Kristina L. Elvidge, Childhood Dementia Initiative, Australia

11:45 SP9-6 Membrane Lipid Composition Regulates Cellular Binding and Uptake of Pegunigalsidase Alfa Across Diverse Cell Types, including Fabry Fibroblasts: Possible implications for multiorgan treatment
Abdullah Hoter, Department of Biochemistry, University of Veterinary Medicine Hannover, Germany / Clinic for Pediatric Kidney-, Liver- and Metabolic Diseases, Hannover Medical School, Germany

Symposium 10: Asian Young Investigator Award
Chairs: Meow-Keong Thong, Malaysia
Thanyachai Sura, Thailand
10:30-12:00, Room 2 (Room A)

10:30 SP10-1 Gene Editing Therapeutic Strategies Targeting Fabry Disease Cardiac Variant IVS4+919 G>A in Taiwan
Yu-Ting Chiang, Institute of Clinical Medicine, National Yang Ming Chiao Tung University, Taiwan

10:45 SP10-2 Genotype, Phenotype and Outcome of Enzyme Replacement Therapy for 50 cases with Infant-Onset Pompe disease
Mai Thi Thanh Do, Vietnam National Children's Hospital, Vietnam

11:00 SP10-3 Deep phenotyping of patients with citrin deficiency in Singapore- Single centre experience
Mildrid Yeo, Genetics Service, Department of Paediatrics, KK Women's and Children's Hospital, Singapore

11:15 SP10-4 Safety and Effectiveness of Triheptanoin in Korean Patients with Fatty Acid Oxidation Disorder
Ji-Hee Yoon, Department of pediatrics, Bundang Jesaeng Hospital, Republic of Korea

11:30 SP10-5 Aminoacyl-tRNA Synthetase Deficiencies: Clinical Spectrum, Functional Characterization, and Treatment Opportunities
Parith Wongkittichote, Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand

11:45 SP10-6 The changing landscape of management of inherited metabolic disorders in India
Swasti Pal, Sir Ganga Ram Hospital , India

Special Symposium 4: Japanese Patient Advocacy

Chairs: Motomichi Kosuga, Japan
Masahisa Kobayashi, Japan
10:30 - 12:00, Room 7 (Room E)

10:30 SSP4-1 指定難病の根治に向けて患者会ができること / The Role of Patient Advocacy Groups in Aiming for a Cure for Designated Intractable Diseases
Yu Onozawa, NPO HPP HOPE

10:45 SSP4-2 患者会の紹介 / Introduction to the Activities of Patient Organizations
Koji Maeda, The Japan Cystinosis Patients and Fmailies Association

11:00 SSP4-3 我が国の在宅酵素補充療法の取組について ～患者サイドから～ / Efforts Toward Home Enzyme Replacement Therapy in Japan: A Patient's Perspective
Hisao Harada, Japan Lysosome Disease Patient Family Association Counci

11:15 SSP4-4 新生児スクリーニングへの取組と新たな患者参画の活動～ 3 つの動きを経て～ / Initiatives for Newborn Screening: Through Three Phases of Development
Ikuko Kaku, Japan Fabry Disease Patients and Family Association

11:30 SSP4-5 日本のポンペ病患者を取り巻く現状と課題 / Current Situation and Challenges Surrounding Pompe Disease Patients in Japan
Junjiro Saeki, Japan Pompe Association

12:00 SSP4-6 NPO AYA の日本における取組について / Introdacution of NPO AYA, Challenges for disabled children in Japan
Yusuke Hamada, NPO AYA

Closing Ceremony

12:00-12:45, Room 1 (Main Hall)

Posters

BP-01 ~ BP-32 ➡ Exhibition & Poster 2 (Event Hall)
P-01 ~ P-557 ➡ Poster 1 (Annex Hall)
P-558 ~ P-668 ➡ Exhibition & Poster 2 (Event Hall)

1. Best Poster Award

BP-01 Genetic Variants and Clinical Features of Patients With Glycogen Storage Disease Ib
Qiu Wenjuan, Department of Pediatric Endocrinology and Genetic Metabolism, Xinhua Hospital, Shanghai Institute of Pediatric Research, School of Medicine, Shanghai Jiao Tong University, China

BP-02 Dual Vascular Changes in Fabry Disease as Neuroimaging Marker
Chong Kun Cheon, DEPARTMENT OF PEDIATRICS, PUSAN NATIONAL UNIVERSITY CHILDREN'S HOSPITAL, Republic of Korea

BP-03 Report on the activities of JaSMIn (Japan Registration System for Metabolic & Inherited Diseases).
Satoko Tsushima, Nursing Department, Specialist Nursing Office, Genetic Coordinator,National Center for Child Health and Development, Japan

BP-04 Low- fat mother´s milk for infants with lipid metabolism disorders¦ promising results from a pre- clinical study
Susanne Dirne-van Alst, Department of Internal Medicine, division of Dietetics, Erasmus Medical Center, Netherlands

BP-05 An interactive, searchable database of LC-FAOD gene variants, genotypes and phenotypes
Aneal Khan, M.A.G.I.C. (Metabolics and Genetics in Canada) Clinic Ltd., Canada

BP-06 Individualized Dosing and Long-Term Outcomes of Givosiran in Recurrent Acute Hepatic Porphyria
Eliane Sardh, Centre for Inherited Metabolic Diseases, Porphyria Centre Sweden, Karolinska University Hospital, Department of Molecular Medicine and Surgery, Karolinska Institutet, Sweden

BP-07 The current status of clinical management and molecular features of congenital disorders of glycosylation in Japan
Nobuhiko Okamoto, Department of Medical Genetics, Osaka Women's and Children's Hospital, Japan / Department of Molecular Genetics and Endocrinology, Research Institute, Osaka Women's and Children's Hospital, Japan

BP-08 The role of Golgi apparatus in human brain development and neural stem cell fate choice: A lesson from the study of COG5-CDG
Elena Taverna, Human Technopole, Italy

BP-09 Development of Small Molecule Therapeutics Targeting Aberrant RNA Splicing in Cardiac Variant Fabry Disease
Ya-Chi Chen, Genetic Consultant Center Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan / Department of Pediatrics, Taipei Veterans General Hospital, Taiwan

BP-10 In vivo editing of the transferrin locus for systemic correction of lysosomal storage diseases: proof-of-concept in mouse models of MPS1 and Pompe disease.
Paula J Waters, Medical Genetics Service, Dept. Laboratory Medicine, Centre hospitalier universitaire de Sherbrooke (CHUS), Canada / Department of Pediatrics, Faculty of Medicine and Health Sciences, Université de Sherbrooke, Canada

BP-11 AT845 gene replacement therapy for late-onset Pompe disease: an update on safety and preliminary efficacy data from FORTIS, a phase 1/2 open-label clinical study
Chieri Hayashi, Astellas Gene Therapies, USA

BP-12 Investigation of in vivo gene therapy for CNS symptoms of GM1 gangliosidosis mediated by blood-brain barrier-penetrating enzymes for clinical application
Saki Matsushima, Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine, Japan

BP-13 Neuro-specific peptides in alpha-mannosidosis patients
Nato Vashakmadze, Research Institute of Pediatrics and Children's Health in Petrovsky National ResearchCentre of Surgery, Russia

BP-14 Modeling Niemann-Pick Disease Type C by Directly Converted Neurons from Urine-Derived Cells
Keita Matsumoto, Department of Physiology, Keio University School of Medicine, Japan

BP-15 Nonclinical pharmacology of JR-446, a novel blood-brain barrier penetrant A-N-acetylglucosaminidase
Jun Ito, JCR Pharmaceuticals Co., Ltd., Japan

BP-16 Alterations in mitochondrial energy metabolism and mitochondria-lysosome contact in MPS I and MPS II knockout cells
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil / PPG Ciências Biológicas: Fisiologia, Universidade Federal do Rio Grande do Sul, Brazil / PPG Neurociências, Universidade Federal do Rio Grande do Sul, Brazil

BP-17 Non-clinical Safety Evaluation of Ex Vivo Gene Therapy for Hunter Disease Using Mouse Model
Takashi Higuchi, The Jikei University, Japan

BP-18 Pathological analysis of sialidosis /galactosialidosis model mice and gene therapy
Jun Tsukimoto, Graduate School of Pharmaceutical Sciences, Tokushima University, Japan

BP-19 Arimoclomol upregulates expression of genes belonging to the coordinated lysosomal expression and regulation (CLEAR) network
Hadeel Shammas, Zevra Therapeutics Inc., USA

BP-20 OXA1L deficiency leads to mitochondrial myopathy via ROS mediated NFkB signaling pathway
Yongkun Zhan, Department of Clinical Genetics Center, Shanghai Institute for Pediatric Research, Xinhua Hospital affiliated to Shanghai Jiao Tong University School of Medicine, China

BP-21 Withdrawal

BP-22 Pilot study for rapid gene panel test in neonates with abnormal newborn screening result of inherited metabolic diseases
Jinsup Kim, Department of Pediatrics, Barowell Hospital, Republic of Korea

BP-23 The use of a second-tier method, combining citrulline levels and gene analysis, significantly improved the detection rate of Citrin deficiency in newborn screening
Hsiao-Jan Chen, The Chinese Foundation of Health, Taipei, Taiwan

BP-24 Newborn CAH screening using a single 17-OHP cut-off value in the primary screening followed by a 2nd tier steroid assay
James Soon Chuan Lim, Biochemical genetics and national expanded newborn screening programme, KK Women's and Children's Hospital, Singapore

BP-25 Transcriptomic analyses of gene expression following CRISPR/Cas9 knockout of ACY1 in HepG2 cells.
Lil Klaas, Research Group Inborn Errors of Metabolism, Department of Natural Sciences & Institute for Functional Gene Analytics (IFGA), Bonn-Rhein-Sieg University of Applied Sciences, Germany

BP-26 Emerging Non-Canonical Pathogenic Variants Disrupting Chromatin Architecture in Inborn Errors of Metabolism
Belen Perez, Centro de Diagnóstico de Enfermedades Moleculares, Centro de Biología Molecular Severo Ochoa, CIBERER, IdiPAZ. Universidad Autónoma de Madrid., Spain

BP-27 Cholesterol ester accumulation in ABCD1-deficient HeLa cells
Masashi Morita, Graduate School of Medicine and Pharmaceutical Sciences, University of Toyama, Japan

BP-28 Decreased Bone Density in Phenylketonuria: PHEFREE- The National Institutes of Health Rare Disease Consortium for Hyperphenylalaninemia
Georgianne Lee Arnold, University of Pittsburgh Medical Center, USA

BP-29 Neuroinflammation in brain of rats submitted to chronic chemically-induced model of hyperphenylalaninemia: L-carnitine protection
Jessica Lamberty Faverzani, Universidade Federal do Rio Grande do Sul, Brazil / Serviço de Genetica Medica, Hospital de Clinicas de Porto Alegre, Brazil

BP-30 Initial Findings from the First Four Patients Enrolled in OTC-HOPE Clinical Trial: No Hyperammonemic Events in First Participant to Complete 24-week Study
Julien Baruteau, Great Ormond Street Hospital for Children, UK

BP-31 Modeling, characterization and therapeutic screening of Citrin deficiency
Toni Vukovic, University Children's Hospital, Switzerland

BP-32 Short-term Therapeutic Effects of UX007 (Triheptanoin) in Citrin Deficiency Mouse Model
Eri Imagawa, Department of Pediatrics, The Jikei University School of Medicine, Japan

2. AI Based Research

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P-02 Circadian-Aware Deep Learning for Predicting Time-Dependent Metabolite Toxicity in Inborn Errors of Metabolism via Multi-Timescale Plasma Profiling and Genomic Variant Integration
Rifaldy Fajar, Mathematical and Computational BioMedicine Research Group, The Integrated Mathematical, Computational, and Data Science for BioMedicine Research Foundation, Indonesia

P-03 Breaking Barriers in Lysosomal Storage Disorder Diagnosis: Using Machine Learning and No-Cost Genetic Testing to Improve Early Detection and Access
Robert J. Hopkin, Cincinnati Children's Hospital Medical Center, USA

P-04 A symptom-based analysis on rare diseases in children: from the view of knowledge graph and large language model
Zhehui Chen, Department of Pediatrics, Women and Children's Hospital, School of Medicine, Xiamen University, China / National Institute for Data Science in Health and Medicine, Xiamen University, China

P-05 AI restores key missing nanopore sequencing metadata to enable large-scale pangenome reference construction
Zhixing Feng, Xinhua Hospital affiliated to Shanghai Jiao Tong University School of Medicine, China

P-06 Federated AI for Scalable, Collaborative Rare Disease Research and Real – World Evidence Generation
Christian Johannes Hendriksz, A Rare Cause, UK / SYMETRYML Inc, USA / North-West University, South Africa

P-07 Enhancing Early Diagnosis of Fabry Disease: A Comparative Study of Machine Learning Models and Feature Selection Strategies
Jalal Khan, Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, UAE

P-08 Evaluating GeneReviews as a Resource for Machine Learning Applications in Treatment and Management of Inborn Errors of Metabolism (IEM)
Tabeer Fatima, United Arab Emirate University, UAE

3. Amino Acid Disorders

P-09 Cohort study of Korean MSUD patients
Sook Za Kim, KSZ Children's Hospital/Korea Genetics Research Center, Republic of Korea

P-10 HIGH-DOSE VALINE, ISOLEUCINE, AND ALANINE SUPPLEMENTATION DURING METABOLIC DECOMPENSATION IN PATIENTS WITH MAPLE SYRUP URINE DISEASE (MSUD) - A SINGLE CENTER EXPERIENCE
Kim Ng, Children's Hospital of Philadelphia, USA

P-11 BCKDK Deficiency Mimicking Autism: Early Diagnosis and Treatment Outcomes
Hanim Aghakishili, Istanbul University- Cerrahpasa, Cerrahpasa Faculty of Medicine, Child Nutrition and Metabolism, Turkey

P-12 Branched-Chain Amino Acid Transferase Type 2 (BCAT2) Deficiency: Three Cases from Slovenia and a Systematic Review
Barbka Repic Lampret, Clinical Institute for Special Laboratory Diagnostics, University Children's Hospital, University Medical Centre , Slovenia

P-13 Difficulties in the management of CBS deficiency in adult patients: overprescription of pyridoxine and under recognition of pyridoxine related complications
Jeremy Clark, Royal Melbourne Hospital, Australia

P-14 Clinical characterization and haplotype analysis in Brazilian patients with Classical Homocystinuria carrying the p.Trp323Ter variant
Gabriela Garcia Silvano, Laboratório Basic Research and Advanced Investigations in Neurosciences (BRAIN) – Centro de Pesquisa Experimental - Hospital de Clínicas de Porto Alegre (HCPA), Brazil

P-15 An update of the Classical Homocystinuria genetic landscape in Brasil
Gabriela Garcia Silvano, Laboratório Basic Research and Advanced Investigations in Neurosciences (BRAIN) – Centro de Pesquisa Experimental - Hospital de Clínicas de Porto Alegre (HCPA), Brazil

P-16 Lessons Learned from following a Large Cohort of Tyrosinemia type-I (HC-I) Patients, One-Center Experience
Ruqaiah Altassan, Medical Genomics, Genomic Medicine Centre of Excellence, King Faisal Specialist Hospital and Research Center, Saudi Arabia

P-17 Tyrosinemia type I complicated with hepatocellular carcinoma
Assel Tulebayeva, Scientific Centre of Paediatric and Paediatric Surgery, Kazakhstan / Asfendiyarov Kazakh National Medical University, Kazakhstan

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P-19 Overview of the situation in the diagnosis and treatment of Tyrosinemia type 1 in Latin America
Veronica CORNEJO, LabGEM, Instituto de Nutrición y Tecnología de los Alimentos(INTA), University of Chile, Chile

P-20 Prevalence, Manifestation, and Genetic Analysis of Hypermethioninemia Cases in Newborns-A Single Center over 20 Years
Jeongho Lee, Department of Pediatrics, Soonchunhyang University Seoul Hospital, Republic of Korea

P-21 A rare cause of methionine demethylation disorder: Glycine N-methyltransferase deficiency
Meryem Karaca, Istanbul University Medical Faculty, Turkey / Pediatric nutrition and Metabolism Department, Turkey

P-22 A case of preterm infant with alkaptonuria associated with a novel variant
Yusuke Hattori, Department of Pediatrics, Kumamoto University Hospital, Japan

P-23 Alkaptonuria in Two Colombian Patients: Identification of HGD Variants Including a Novel Finding
Maria Camila Leon Sanabria, Institute of Human Genetics, Pontificia Universidad Javeriana, Colombia / Servicio de Genética y Clinica de Errores Innatos del Metabolismo, Hospital Universitario San Ignacio, Colombia

P-24 Systemic lupus erythematosus as the primary symptom in patients with lysinuric protein intolerance
Yifan Li, Department of Rheumatology, Children's Hospital of Fudan University, National Center for Children's Health, China

P-25 Investigation of Serum Acid Sphingomyelinase Activity in Patients with Lysinuric Protein Intolerance
Atsuko Noguchi, Akita University GraduateSchool of Medicine, Japan

P-26 An Atypical Case of Lysinuric Protein Intolerance: Unveiling a Hidden Diagnosis Through Pneumocystis Pneumonia
Chern Yan Tan, The Willink Metabolic Unit, UK

P-27 Do Inborn Errors of Metabolism Affect Social-Emotional Competencies in Early School-Aged Children?
Amanda Natalia Krzywdzinska-Rogowska, Department of Inborn Errors of Metabolism and Pediatrics, Institute of Mother and Child, Poland

P-28 The Clinical Screening Strategy to overcome challenges of Early Detection Treatable Inborn Errors of Matabolisms In Limited Resources Area
Damayanti Rusli Sjarif, Div Pediatric Nutrition And Metabolic Diseases Faculty Of Medicine Universitas Indonesia And Dr Cipto Mangunkusumo National Referral Hospital Al Hospital, Indonesia

P-29 A Devastating Neurodevelopmental Disorder: Glutaminase Deficiency with a Novel GLS Variant
Merve Yoldas Celik, Adana City Training and Research Hospital, Department of Pediatric Metabolism, Turkey

P-30 3-Hydroxyisobutyryl-CoA Hydrolase (HIBCH) Deficiency-First Case Report from Pakistan
Bushra Afroze, Department of Paediatrics & Child Health, Aga Khan University, Pakistan / Chemical Pathology, Aga Khan University, Pakistan

P-31 High anion gap metabolic acidosis, dare to think rare: 5-Oxoprolinuria-Novel variant in Glutathione Synthetase Deficiency in two Sri Lankans
Mihika Samindi Fernando, Department of Chemical Pathology, Lady Ridgeway Hospital for Children, Sri Lanka

P-32 Inhibition of SLC6A19 Normalizes Toxic Amino Acid Levels Associated with Inborn Errors of Metabolism
Heike J Wobst, Jnana Therapeutics, USA

P-33 Aminoacidopathies in Tunisia: A pilot study from the Tunisian registry
Mouna Bouchouicha, Rabta hospital, Tunisia

P-34 Application of metabolomics in differential diagnosis of amino aciduria
Chunhua Zhang, MILS International, Japan

P-35 Dual model nutriphenomics to identify precision dietary therapies for inherited metabolic disorders
Jemma Gasperoni, La Trobe University , Australia

P-36 Untargeted nutrigenomics in Drosophila: Dietary insights for inherited disorders of amino acid metabolism
Zoriana Novosiadla, Department of Biochemistry and Chemistry, La Trobe Institute for Molecular Science, La Trobe University, Australia

P-37 FOCAD variant in an infant: A rare cause of acute liver failure
Doaa Ali Alsultan, Genetics and Metabolic Medicine Division, Department of Pediatrics,Prince Sultan Military Medical City, Saudi Arabia

P-38 From developmental delay to three different rare diagnoses
Ayse Nur Altun, Gazi Yasargil Training and Research Hospital, Department of Pediatrics, Division of Pediatric Metabolic Diseases, Turkey

P-39 Effects of the NMDA Receptor Antagonist Memantine in a Neonatal Animal Model of Nonketotic Hyperglycinemia
Guilhian Leipnitz, Post Graduation Program in Biological Sciences: Biochemistry, Universidade Federal do Rio Grande do Sul, Brazil / Post Graduation Program in Biological Sciences: Physiology, Universidade Federal do Rio Grande do Sul, Brazil / Post Graduation Program in Neurosciences, Universidade Federal do Rio Grande do Sul, Brazil

P-40 A rare diagnosis of prolidase deficiency (missed on short-read genome analysis) made by integrating long-read sequencing and multi-omic validation
Ivan De Dios, Institute for Clinical and Translational Science, USA

P-41 NEONATAL PYROGLUTAMIC ACIDURIA: ABOUT 3 CASES
Kamel Monastiri, Teaching Hospital of Monastir, Tunisia

P-42 PROLINE DEFICIENCY DISORDERS: ABOUT TWO CASES
Kamel Monastiri, Department of Intensive Care and Neonatal Medicine, Teaching Hospital of Monastir, Tunisia

4. Carbohydrate Disorders

P-43 Continuous Glucose Monitoring (CGM) and Psychological Assessment in Adults with Inherited Metabolic Disorders (IMDs) Prone to Hypoglycemia
Nicola Vitturi, Division of Metabolic Diseases, Department of Medicine, University Hospital of Padova, Italy

P-44 Comprehensive Clinical, Biochemical, and Genetic Characterization of Glycogen Storage Diseases in an Iranian Cohort Using Targeted Gene Sequencing and Metabolomics
Zahra Beyzaei, Shiraz University of Medical Sciences, Iran

P-45 Nutritional management and fasting tolerance in children with Glycogen Storage Disorders
Erin Mullane, Royal Children's Hospital, Australia

P-46 Overview of Glycogen Storage Disease in Brazil: Data from the Brazilian Rare Disease Network
Mariana Lima Scortegagna, Hospital de Clínicas de Porto Alegre, Brazil

P-47 Effect of empagliflozin treatment on laboratory and clinical findings of patients with glycogen storage disease type Ib: first study from Turkey
Engin Kose, Department of Pediatric Metabolism, Ankara University Faculty of Medicine, Turkey

P-48 Clinical outcomes and molecular spectrum of Indian patients with Glycogen storage disorder type 1b
Neerja Gupta, All India Institute of Medical Sciences, India

P-49 Empagliflozin for the treatment of metabolic disorders in children - a single centre experience
Chern Yan Tan, The Willink Metabolic Unit, UK

P-50 The NAD^+ secretion pathway is consistently upregulated in hepatic Glycogen Storage Disease Type I mice
Candelas Gross-Valle, Department of Pediatrics, University of Groningen, University Medical Center Groningen, Netherlands

P-51 The use of Modified Atkins Diet in the treatment of a 5-year-old patient with Glycogen Storage Disease type III
Maria Pekala, Department of Pediatrics, Nutrition and Metabolic Disorders, Children's Memorial Health Institute, Poland

P-52 Infant formula composition affects tetraglucoside (Glc4) excretion: the impact on the diagnostics of inborn errors of metabolism
Silvia Radenkovic, Department of Genetics, Section Metabolic Diagnostics, Wilhemina Children's Hospital, University Medical Center Utrecht, Netherlands

P-53 Nine-Year Outcome of a High-Protein, High-Fat Diet in a Paediatric Patient with Glycogen Storage Disease Type IIIa: A Case Report
Siew Li Ting, Willink Metabolic Unit, Genomic Medicine, St Mary's Hospital, Manchester University NHS Foundation Trust, UK

P-54 CLINICAL SPECTURM OF GSD IV/ GLYCOGEN BRANCHING ENZYME DEFICIENCY; AN EXPERIENCE FROM A TERTITARY CENTRE IN THE UK.
Nirubhan Veeraraghavan, Evelina Children Hospital, UK

P-55 Discordant phenotype in Glycogenosis XI secondary to a de novo mutation in the SLC2A2 in Colombian families
Patricia Ruiz Navas, Peadiatric Hepatologist, Colombia

P-56 Understanding Glycogen Storage Disease type IX: A Systematic Review with Clinical Focus : Why It Is Not Benign and Re-quires Vigilance
Egidio Candela, Pediatric Unit, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Italy

P-57 The feasibility, acceptability, and clinical outcomes of a structured exercise intervention in children and young adults with McArdle disease.
Kiera Batten, Department of Nutrition and Dietetics, The Children's Hospital at Westmead, Australia / Genetic Metabolic Disorders Service, The Children's Hospital at Westmead, Australia / School of Health Sciences, Faculty of Medicine and Health, The University of New South Wales, Australia

P-58 Clinical and epidemiological profile of galactosemia: An analysis from the Brazilian Rare Diseases Network
Monique Sartori Broch, Hospital de Clínicas de Porto Alegre (HCPA), Brazil / Rede Nacional de Doenças Raras, Brazil

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P-60 The multifaceted aspects of bone health in classical galactosemia - a cross-sectional analysis combining high resolution quantitative CT scans (HR-pQCT) and whole body DXA
Michela Motti, Department of Diabetes, Endocrinology, Nutritional Medicine and Metabolism, Inselspital, Bern University Hospital and University of Bern, Switzerland

P-61 Co-occurrence of Autism Spectrum Disorders in Pediatric Patients with Classical Galactosemia: A Polish Cohort Study
Amanda Natalia Krzywdzinska-Rogowska, Department of Inborn Errors of Metabolism and Pediatrics, Institute of Mother and Child, Poland

P-62 Cognitive and Executive Function Impairment in Children with Classical Galactosemia: Insights from a Pilot Study of Polish Children
Amanda Natalia Krzywdzinska-Rogowska, Department of Inborn Errors of Metabolism and Pediatrics, Institute of Mother and Child , Poland

P-63 Hidden Congenital Porto-Systemic Shunt and Hypergalactosemia at Newborn Screening
Laura Fiori, Department of Pediatrics, Vittore Buzzi Children's Hospital, Italy

P-64 A Rare Dual Diagnosis: Fructose-1,6-Bisphosphatase Deficiency and D-2-Hydroxyglutaric Aciduria in a Child with Hypoglycemia and Developmental Delay
Merve Yoldas Celik, Adana City Training and Research Hospital, Department of Pediatric Metabolism, Turkey

P-65 Hereditary fructose intolerance: an often unrecognised cause of hepatic steatosis
Dinusha Pandithan, Evelina London Children's Hospital, UK

P-66 Symptom Variability in Transaldolase Deficiency: Three Unrelated Families with an Identical Pathogenic Variant
Dmitry Igorevich Grebenkin, National Medical Research Center for Children's Health, Ministry of Health of Russia, Russia

P-67 Sucrase-Isomaltase Gene variants in Irritable Bowel Syndrome: Impact on Protein Trafficking and Enzyme Function
Stephanie Tannous, University of Veterinary Medicine Hannover, Germany

P-68 A Study on the Clinical Characteristics and the Therapeutic Effect of Recombinant Human Growth Hormone in 21 Chinese Children with Gitelman Syndrome
Yaqing Lu, Children's Hospital Affiliated to Zhengzhou University, China

P-69 MOLECULAR GENETIC CHARACTERISTICS OF PATIENTS WITH ACUTE LIVER FAILURE DUE TO WILSON'S DISEASE
Huong T.M Nguyen, Vietnam National Children's Hospital, Vietnam

P-70 Solid organ transplantation for patients with Alstrom syndrome – a case series of organ transplanted patients in the United Kingdom
Subadra Wanninayake, Department of Inherited Metabolic Disorders, University Hospitals Birmingham NHS Trust, UK

P-71 Newborn Screening for Lysosomal Diseases: Insights from the LysoNeo Study
Soumeya Bekri, Department of Metabolic Biochemistry, Referral Center for Lysosomal Diseases, Normandie Univ, UNIROUEN, CHU Rouen, AIMS, SysMedLab, Filière G2M, France

P-72 Phase III, Open-label, Switch-over Trial of the Efficacy and Safety of Agalsidase Beta Biosidus (Agalzyme ®) in Fabry Disease Patients Previously Stabilized with Fabrazyme ®
Viridiana Berstein, Departamento de Investigación Clínica, Biosidus S.A.U., Argentina

P-73 Enzyme replacement therapy with Olipudase Alfa (Xenpozyme) in Niemann-Pick Disease Type AB: A case series showing promising results
Hadeel ayman Alrabee, Prince Sultan Military Hospital, Saudi Arabia

P-74 Changes in clinical manifestations in patients with alpha-mannosidosis treated with velmanase alfa therapy in the French Etoile Alpha registry
Nathalie Guffon, Reference Center for Hereditary Metabolic Diseases, Hospices Civils of Lyon (HCL), France

P-75 Trappsol®Cyclo™: OPEN LABEL TREATMENT IN THE TRANSPORTNPC™ SUB-STUDY IN PATIENTS UNDER THE AGE OF 3 DIAGNOSED WITH NIEMANN PICK DISEASE TYPE C1
Orna Staretz Chacham, Soroka Medical Center, Faculty of Health Sciences, Ben-Gurion University of the Negev, Israel

P-76 Arimoclomol for the treatment of Niemann-Pick disease type C in a real-world setting: Long-term data from an expanded access program in the United States
Caroline Hastings, UCSF Benioff Children's Hospital Oakland, USA Minor Outlying Islands

P-77 Implementation of a Novel Guideline for Rhabdomyolysis in a Patient with VLCAD Deficiency
Pamela Maree Tucker, Children's Institute of Pittsburgh, USA

P-78 INBORN ERRORS OF METABOLISM IN UNEXPLAINED ACUTE ENCEPHALOPATHY: CHALLENGES IN DIAGNOSIS AND MANAGEMENT
Muhammed Shabeer P, Institute of Medical Genetics and Genomics, Sir Ganga Ram Hospital, India

P-79 Prospective Open Label Administration of Mefenamic Acid in Patient with KCNK9 Imprinting Syndrome: 1-year Report of Behavioral and Developmental Outcomes of Two Interrupted Therapeutic Regimens
Khalsa Amur Al Sulaimi, Oman Medical Specialty Board, Oman

P-80 Case Management of Patients from a Large Brazilian Family with Segawa Syndrome Using Patient-Reported Outcome Measures
Ailton Cezario Alves Junior, Postgraduate Programme in Health Sciences - Faculty of Medicine - Federal University of Minas Gerais, Brazil

P-81 Clinical Challenges of Pregnancy and Postpartum in a Woman with Segawa Disease and Severe Restless Legs Syndrome Managed by Case Management Technology
Eugenia Ribeiro Valadares, Postgraduate Programme in Health Sciences - Faculty of Medicine - Federal University of Minas Gerais, Brazil

P-82 Preliminary results of the International Cooperative Assessment of Ataxia Scale (ICARS) from an ongoing PMM2-CDG Natural History Study
Mercedes Serrano, Neuropediatric Department, Hospital Sant Joan de Déu, Spain

P-83 Prevalence of inherited metabolic disorders in epilepsy: Do we still need to perform biochemical genetic investigations in epilepsy genetic clinics?
Saadet Mercimek-Andrews, Department of Medical Genetics, University of Alberta, , Canada

P-84 Clinical Spectrum of Carnitine-Acylcarnitine Translocase Deficiency Associated with the SLC25A20 c.199-10T>G Variant: Experience from Children's Hospital 1, Vietnam
Nhung Tran, Children' hospital 1, Vietnam

P-85 Cholestatic Jaundice in a Neonate: Coexisting Citrin Deficiency and Biliary Atresia - A Rare Diagnostic Challenge
Nhung Tran, Children's Hospital 1, Vietnam

P-86 Clinical characteristics and genetic analysis of 3 children with cblB type methylmalonic acidemia
Zixia Zhang, Affiliated Children's Hospital of Zhengzhou University, China

P-87 Amino Acid Deficiency Secondary to Continuous Veno-Venous Hemofiltration in Cases of Hyperammonemia: An Anabolic Dead End?
Chloe Grosyeux, Pediatric Nephrology Department, University Hospital of Nancy, France

P-88 Blood cobalt ion level in patients of methylmalonic acidemia with cobalamins injection
Huiting Zhang, Peking University First Hospital, China

P-89 Miglustat as a treatment for adults with Tangier Disease Neuropathy: the MUSTANG N–of–1 trial with 21 months clinical observation
Andrew Cook, Clinical Trials Unit, University of Southampton, UK

P-90 Genomic Insights into Rare Pediatric and Adult-Onset Metabolic and Neurodevelopmental Disorders: A Case Series from a Molecular Diagnostic Laboratory in Nepal
Sandeep Thapa, Kathmandu Center for Genomics and Research Laboratory (KCGRL), Nepal

P-91 The 3FM-Serious Request National Campaign by Metakids & United for Metabolic Diseases: A Unique Partnership Raising National Funds and Awareness for IMDs
Clara van Karnebeek, Emma Center for Personalized Medicine, Amsterdam UMC, Home of United for Metabolic Diseases , Netherlands

P-92 ADOLESCENT PERCEPTIONS OF HEALTHCARE SERVICES FOR INHERITED METABOLIC DISORDERS AND GENETIC DISEASES: INSIGHTS FROM A SURVEY IN A PEDIATRIC HOSPITAL
Albina Tummolo, Department of Metabolic Diseases and Clinical Genetics, Giovanni XXIII Children Hospital, Italy

P-93 Psychosocial Perspectives on Relationships and Reproductive Decision-Making in Late Adolescents and Young Adults with Inherited Metabolic Disorders
Albina Tummolo, Children Hospital Giovanni XXIII, Department of Metabolic Diseases and Clinical Genetics, Italy

P-94 Comparative Analysis of Two Major Children's Hospitals in the United States: Similarities and Differences in Metabolism Programs
Kimberly A Chapman, Children's Hospital Los Angeles, USA

P-95 Review of 830 published patients with PSACH or EMD1: clinical and mutational spectrum
Xiaolin Ni, Beijing Children's Hospital, China

P-96 Burden and Outcomes of Hospitalization in Inherited Metabolic Disorders: Insights from a Single Center
Carlos Enrique Prada, Division of Genetics, Genomics, and Metabolism, Ann & Robert H. Lurie Children's Hospital of Chicago, USA / Department of Pediatrics, Northwestern University Feinberg School of Medicine, USA

P-97 Screening and Registry Project for hereditary Urolithiasis Using Urine Mass spectrometry
Seiji Tanaka, Department of Pediatrics and Child Health Kurume University School of Medicine, Japan

P-98 Neonatal Ammonia Sepsis Screening (NASS) in Oregon Birthing Hospitals
Amy Chun-Yao Yang, Molecular and Medical Genetics, Oregon Health & Science University (OHSU), USA

P-99 Four cases of KBG syndrome due to 16q24.3 microdeletion and literature review
Miao Miao Li, Department of Endocrinology and Inborn Error of Metabolism,Children's Hospital Affiliated to Zhengzhou University,Henan Children's Hospital,ZhengzhouChildren's Hospital, China

P-100 Thyroxine binding globulin deficiency in three families and review of the literature
Miao Miao Li, Department of Endocrinology and Inborn Error of Metabolism,Children's Hospital Affiliated to Zhengzhou University,Henan Children's Hospital,Zhengzhou Children's Hospital, China

P-101 Insulin Resistance Patterns and Risk Factor Associations Across Tanner Stages in Girls with Central Precocious Puberty
Jin-bo Li, Department of Pediatric Endocrinology, Genetics and Metabolism, The Third Affiliated Hospital of Zhengzhou University, China

P-102 Phenotypic and Genotypic Analysis of 186 Chinese Patients with Congenital Adrenal Hyperplasia: a retrospective, multicentre study
Lianjing Huang, Department of Pediatrics, Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, China / Hubei Provincial Key Laboratory of Pediatric Genetic Metabolic and Endocrine Rare Diseases, Wuhan, China / Hubei Provincial Clinical Research Center for Children's Growth and Development and Metabolic Diseases, Wuhan, China

P-103 How Small Teams Can Succeed in Sponsored Research: Building a Strategic Framework for Clinical Trial Excellence
Farideh Aljallad Oberheu, University of South Florida, USA / Tampa General Hospital, USA

P-104 Immunization coverage and timeliness of vaccination in young patients with inborn errors of metabolism: a French multicentric study
Anne-Sophie RENOUS, ToTeM, CHRU TOURS, France

P-105 NOD-M: SYSTEMIC INDICATORS OF INHERITED METABOLIC DISORDERS IN HEARING LOSS
Fatma Tuba EMINOGLU, Ankara University Faculty of Medicine, Department of Pediatric Metabolism, Turkey

P-106 Challenges in Genetic Diagnosis of Extreme Early-Onset Obesity in Children Under 5
Si Chen, Chengdu Women's and Children's Central Hospital, China

P-107 Association Between Folic Acid Levels and Physiological and Developmental Indicators in Children with Developmental Delays
Si Chen, Chengdu Women's and Children's Central Hospital, China

P-108 Clinical - Experimental Study on Melatonin Deficiency and Central Precocious Puberty: Correlation, Mechanism and Intervention
Shuxian Yuan, Children's Hospital Affiliated to Zhengzhou University, Henan Provincial Children's Hospital, China

P-109 DIAGNOSTIC APPROACH TO CARDIOMYOPATHIES:
A STUDY OF A TUNISIAN PAEDIATRIC SERIES
Zeineb Najar, Pediatric Department of La Rabta hospital, Tunisia

P-110 PAH genotype and their impact on responsiveness to sapropterin treatment in Thai patients with phenylketonuria
Dhachdanai Dhachpramuk, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand

6. Dietetics and Nutrition

P-111 Therapeutical implications of rethinking inborn errors of intermediate metabolism as "euphenic" or "non-euphenic"
Francesco Porta, Department of Clinical and Biological Sciences, University of Torino, Italy / Department of Pediatrics, University of Torino, Italy

P-112 Registered Dietitians in Metabolic Units in Spain
Marta Suarez-Gonzalez, Hospital Universitario Central de Asturias, Spain

P-113 Scenario of Dietary practices in a Resource Limited Setting: Perspective of a remote clinic in India
Madhavi Harikrishna Kulkarni, Butterflies Child Development Centre, India / NIRMAN, India

P-114 Does medication for treatment of Attention Deficit Hyperactivity Disorder in Children With Phenylketonuria affect growth and Metabolic Control?
Angela Harris, Royal Children's Hospital, Australia

P-115 Substantial Dietary Intake Variation in Phenylketonuria Highlights Implications for Clinical Trial Design
Rani H Singh, Department of Human Genetics, Emory University School of Medicine, USA

P-116 Longitudinal assessment of body composition in an infant with phenylketonuria treated at an early age
Júlio César Rocha, Nutrition and Metabolism, NOVA Medical School, Faculdade de Ciências Médicas, Universidade NOVA de Lisboa, Portugal / Reference Centre of Inherited Metabolic Diseases, ULS São José, Portugal / CINTESIS - Center for Health Technology and Services Research, NOVA Medical School, Portugal / CHRC - Comprehensive Health Research Center, NOVA Medical School, Portugal

P-117 Phenylalanine Metabolism in Premature, Very Low Birthweight (VLBW) Infants - Observations From The New Zealand Newborn Metabolic Screening Programme
Helen Prunty, Auckland City Hospital, New Zealand

P-118 Nutritional management of a successful multifetal PKU Pregnancy
Suzanne Ford, North Bristol NHS Trust, UK

P-119 Dietitian Prescribers in Inherited Metabolic Disorders; experience prescribing Sapropterin Dihydrochloride for Phenylketonuria
Camille Newby, University Hospitals Bristol and Weston NHS Foundation Trust, UK

P-120 Acceptability of Ready-to-Use Valine and Isoleucine Supplements in the Dietary Management of MSUD: A Prospective Study
Anne Daly, Department of Dietetics, Birmingham Women's and Children's Hospital, UK

P-121 Ornithine transcarbamylase (OTC) deficiency. A psychological-dietetic perspective
Gabriele Skacel, Department of Pediatrics and Adolescent Medicine, Division of Pediatric Pulmonology, Allergology and Endocrinology, Medical University of Vienna, Austria

P-122 Utilization of a Modified Adkins Diet for a child with argininosuccinate lyase deficiency and intractable epilepsy
Erin L MacLeod, Children's National Rare Disease Institute, Children's National Hospital, USA

P-123 Dietary treatment of adults with Methylmalonic acidaemia (MMA):A survey to examine the dietetic practice of adult metabolic Dietitians in the UK and Ireland.
Melanie Claire Hill, Northern General hospital NHS Foundation Trust, UK

P-124 Dietetic Management of 41 Galactosemia Patients: Initial Feeding Practices and Challenges in Identifying Suitable Galactose-Free Formulas.
Eleana Petropoulou, Institute of Child Health, Greece

P-125 Effects of nutritional counseling by telemedicine in pediatric patients with Inherited Metabolic Diseases diagnosed by Newborn Screening during their first year of life: preliminary results
Veronica Maria Tagi, Department of Biomedical and Clinical Science, University of Milan, Italy / Department of Pediatrics, Vittore Buzzi Children's Hospital, Italy

P-126 Skimmed breast milk in an infant with long chain 3-hydroxyacyl-coA dehydrogenase deficiency
Delia Barrio-Carreras, Refence Center for Inherited Metabolic Disorders (MetabERN) Hospital Universitario 12 de Octubre, Spain

P-127 Healthcare provision and patients outcomes in adolescents with Phenylketonuria: A UK centre experience.
Alex Pinto, Birmingham Children's Hospital, UK

P-128 More evidence of declining blood phenylalanine control in paediatric and adult centres in the UK
Alex Pinto, UK longitudinal project on blood Phe control in PKU, UK

P-129 Dietary intake and overweight in patients with MCADD
Alex Pinto, Birmingham Children's Hospital, UK

P-130 Implementation of a Ketogenic Diet to Manage Severe Hypertrophic Cardiomyopathy in an Infant with Glycogen Storage Disease Type IIIa
Katherine Arduini, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

P-131 Long-Term Follow-Up of Targeted Nutritional Intervention in LARS1 Deficiency: Reduced Hepatic Episodes and Improved Clinical Outcome
Damla Kocaman, Marmara University, Pendik Education and Research Hospital, Nutrition and Dietetics, Turkey

P-132 Gut Microbiota and cytokine in Children With Nonalcoholic Fatty Liver Disease
Xin Yuan, Fuzhou First General Hospital Affiliated with Fujian Medical University, Fuzhou Children's Hospital of Fujian Medical University, China

P-133 The underlying mechanism of brain damage caused by hypercarotenemia
Xue Ma, Children's Medical Center, Peking University First Hospital, China

7. Disorders of Fatty Acid Oxidation and Ketone Body Metabolism

P-134 Cohort of Patients with Fatty Acid β-Oxidation Disorders in the Adult Metabolic Disorders Unit of the Virgen del Rocio University Hospital (HUVR)
Eva Venegas-Moreno, Hospital Universitario Virgen del Rocio, Spain

P-135 Newborn screening leads to an unexpected diagnosis
Sara Lopes Ferreira, Local Health Unit of Coimbra, Portugal / Member of metabERN, Portugal

P-136 Fatty Acid Oxidation Disorders in Southern Brazil: Insights from Rio Grande do Sul state
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David Olsson, Department of Women's and Children's Health, Unit for Pediatric Endocrinology Karolinska Institutet, Sweden / Department of Pediatric endocrinology and inborn errors of metabolism, Astrid Lindgren Children's Hospital, Karolinska University Hospital, Sweden

P-138 Resolving Complex Acylcarnitine Chromatography: High-Resolution Mass Spectrometry for Identifying Interferences and Isomers
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Justin Dutta, Division of Genetic and Genomic Medicine, Department of Pediatrics, University of Pittsburgh School of Medicine, USA / Dept of Human Genetics, School of Public Health, University of Pittsburgh , USA

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Tomoki Saito, Department of Endocrinology and Metabolism, Hyogo Prefectural Kobe Children's Hospital, Japan

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Troy von Beck, Division of Medical Genetics, Department of Pediatrics, Duke University Medical Center, USA

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Carolyn Bursle, Queensland Lifespan Metabolic Medicine Service, Australia

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Andrea Pascual-Ramirez, Internal Medicine Department, Hospital Clínic de Barcelona , Spain

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David Benjamin Manser, Department of Genetic Medicine, Westmead Hospital, Australia

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Shirou Matsumoto, Department of Neonatology, Kumamoto University Hospital, Japan

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Seiya Oshima, Tohoku University, Japan

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Blanka Stiburkova, Institute of Rheumatology, Czech Republic

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Fatma Tuba Eminoglu, Department of Pediatric Metabolism and Nutrition, Ankara University Faculty of Medicine, Turkey / Rare Diseases Application and Research Center, Ankara University, Turkey

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Beata Vekeriotaite, Laboratory of Nutrition and Metabolic Epigenetics, Institute of Food, Nutrition and Health, Department of Health Sciences and Technology, ETH, Switzerland / Division of Metabolism and Children's Research Center, University Children's Hospital, University of Zurich, Switzerland

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P-174 A novel therapy for pyridoxine dependent epilepsy due to biallelic pathogenic variants in ALDH7A1: secondary mitochondrial energy deficiency and improvements of neurodevelopmental outcome on triheptanoin
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Rebekah Barrick, Division of Metabolic Disorders, Children's Hospital of Orange County, USA

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Tingting Hu, Department of Pediatric Endocrinology and Genetics, Xinhua Hospital Affiliated to Shanghai Jiao Tong University School of Medicine; Shanghai Institute for Pediatric Research, China

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Anne Mei-Kwun KWOK, Department of Pediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong

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Patricia L Hall, Mayo Clinic, USA

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Hicham Alaoui, Glycomine, USA

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Huiting Zhang, Peking University First Hospital, China

P-190 Phenotype and genotype Analysis of PMM2-CDG in a Tunisian Cohort
Thouraya Ben Younes, LR18SP04 and Department of Pediatric Neurology, National Institute Mongi Ben Hmida of Neurology, Tunisia

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James Nurse, Southampton General Hospital, UK

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Veronika Holubova, Department of Pediatrics and Inherited Metabolic Disorders, First Faculty of Medicine, Charles University and General University Hospital in Prague, Czech Republic

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Julien Park, University Hospital Muenster, Department of General Pediatrics, Germany

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Julien H. Park, University Hospital Muenster, Department of General Pediatrics, Germany

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Alessandra Verde, Department of Clinical and Experimental Sciences, University of Brescia, Italy

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Irena Josephina Johanna Muffels, Icahn School of Medicine at Mount Sinai, USA

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Lisa Siegal, Wilhelmina Children's Hospital, University Medical Center Utrecht, Netherlands

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Wongsathorn Eiumtrakul, Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand

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Milena Greczan, Department of Pediatrics, Nutrition and Metabolic Diseases, Children's Memorial Health Institute, Poland

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Jie Ming Yeo, Department of Paediatric Inherited Metabolic Diseases, Evelina London Children's Hospital, UK

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Hana Hansikova, Department of Paediatrics and Inherited Metabolic Disorders, First Faculty of Medicine, Charles University and General University Hospital, Czech Republic

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Michel Tchan, Westmead Hospital, Australia

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Ayako Matsunaga, St. Marianna University School of Medicine, Japan

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Maximilian Pichler, Endocrinology and Metabolism, IM III, Medical University of Vienna, Austria

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Roxanne Janica Evangelista Merencilla, University of the Philippines Manila College of Medicine, Philippines / Institute of Human Genetics - National Institutes of Health, University of the Philippines Manila, Philippines

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Shagun Kaur, Phoenix Children's, USA / University of Arizona College of Medicine - Phoenix, USA

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Vivek Pant, Samyak Diagnostic Pvt Ltd, Nepal

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Samuel MF Bradbrook, Department of Medical Genetics, University of Calgary, Canada

P-215 FGF-21 Elevation with Associated Clinical and Biochemical Changes in Citrin deficiency (CD) Patients
Jiaqi Liang, Department of Paediatrics and Adolescent Medicine, The University of Hong Kong, Hong Kong

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Rencia van der Sluis, Biomedical and Molecular Metabolism Research (BioMMet), North-West University, South Africa

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Michela Perrone Donnorso, LABSIEM-Pediatric Clinic Unit-DINOGMI, University of Genova, IRCCS Istituto Giannina Gaslini, Italy

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Rawan Mohammad Alqahtani, Prince Sultan Military Medical City, Saudi Arabia

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Ryuta Takase, Dept.Pediatrics and Child Health, Kurume University, Japan

P-220 Why is genetic confirmation of the diagnosis important? A case of double diagnosis of propionic acidemia due to mosaic uniparental isodisomy and X-linked craniofrontonasal dysplasia
Laura Maaria Tanner, Department of Medical and Clinical Genetics, University of Helsinki, Finland

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Josefina Olivares, Endocrinology and Nutrition Department, Spain / Son Espases University Hospital , Spain / IdISBa, Spain

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Tasneem Binte Raheel, University of Manchester, UK

P-223 Clinical Characteristics of Adult Fabry Disease Diagnosed via Newborn Screening Compared to Non-Screening Diagnoses
Masanori Hirose, Chiba University Hospital, Japan

P-224 Investigation of Modifier Genes Associated with Clinical Manifestation Variability in Fabry Disease Patients with the GLA IVS4+919 G>A Mutation
Chien-Hui Yen, Institute of Clinical Medicine, National Yang Ming Chiao Tung University, Taiwan / Genetic Consultant Center Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan

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Yu-Ping Hsieh, Genetic Consultant Center Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan / Department of Pediatrics, Taipei Veterans General Hospital, Taiwan

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Kairit Joost, East-Tallinn Central Hospital, Estonia

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Ryosuke Bo, Department of Pediatrics, Kobe University Graduate School of Medicine, Japan

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Subadra Wanninayake, Department of Inherited Metabolic Disorders, Queen Elizabeth Hospital Birmingham, UK

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Raymond Yu-Jeang Wang, Children's Hospital of Orange County, USA

P-233 Ambroxol Hydrochloride as a Therapeutic Approach in SanFlippo Syndrome (MPSIII)
Ozlem Goker-Alpan, Lysosomal and Rare Disorders Research and Treatment Center, USA

P-234 In Vivo Direct Lentiviral Gene Therapy Improves Disease Pathology in a Mucopolysaccharidosis IVA Murine Model
Shunji Tomatsu, Nemours Children's Health, USA

P-235 CRISPR/nCas9-edited CD34+ cells rescue MPS IVA fibroblasts phenotype
Shunji Tomatsu, Nemours Children's Health, USA

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Shunji Tomatsu, Nemours Children's Health, USA

P-237 In vitro assessment of cytosine base editing and prime editing for correction of the common p.N409S and p.L483P GBA1 pathogenic variants
Chloe Christensen, Children's Hospital of Orange County, USA

P-238 Lentiviral gene therapy for Fabry disease: 5 year end of study results from the Canadian FACTS trial.
Aneal Khan, M.A.G.I.C. Clinic (Metabolics and Genetics in Canada), Canada

P-239 Invention of an oral medication for cardiac Fabry disease caused by RNA mis-splicing for GLA c.639+919G>A variant
Tomonari Awaya, Department of Anatomy and Developmental Biology, Graduate School of Medicine, Kyoto University, Japan / Center for Anatomical Studies, Graduate School of Medicine, Kyoto University, Japan

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George Aldersley, Department of Paediatric Neurology, Royal Manchester Children's Hospital, UK

P-241 An Innovative Platform Approach for the Parallel Development of HSC-GT for Rare/Ultra-Rare Lysosomal Storage Disorders with Severe Skeletal and Neurological Manifestations
Stefania Crippa, San Raffaele Telethon Institute for Gene Therapy (SR-TIGET), Italy

P-242 Nuclease-facilitated homology directed repair (HDR)-mediated gene insertion in liver corrects hyperphenylalaninemia in murine phenylketonuria (PKU)
Cary O Harding, Oregon Health & Science University, USA

P-243 Analysis of Premature Termination Codons (PTCs) in Phenylketonuria (PKU): Genotype/Phenotype Correlations
Nerissa C. Kreher, Alltrna, USA

P-244 CRISPR base editing for OTC stabilization
Sven Klassa, University Children's Hospital Zurich, Switzerland

P-245 Ass1 Targeted Gene Insertion in Newborn Mice Provides Effective, Long Term Disease Correction in a Lethal Mouse Model of Citrullinemia Type I
Evangelos Pefanis, Regeneron Pharmaceuticals, USA

P-246 mRNA-3745 Therapy for GSD1a: Interim reported data from Phase 1/2 Ba1ance Study
Nicola Longo, University of California Los Angeles, USA

P-247 mRNA therapy for Glycogen Storage Disease type 1b
Lucia De Stefano, Telethon Institute of Genetics and Medicine , Italy

P-248 Evaluation of efficacy and safety of AAV8-ATP7B gene therapy in a mutant mouse model of Wilsons disease
Chunhua Zeng, Guangzhou Women and Children's Medical Center, Guangzhou Medical University, China

P-249 Liver gene therapy with optimized lentiviral vectors provides long-term efficacy, safety and extra-hepatic benefit in a mouse model of methylmalonic acidemia
Elena Barbon, San Raffaele Telethon Institute for Gene Therapy, IRCCS San Raffaele Scientific Institute, Italy

P-250 Personalized Splice-modulating Antisense Oligonucleotide Therapy for PEX1-related Zellweger Spectrum Disorder (ZSD)
Didem Demirbas, Boston Children's Hospital/Harvard Medical School, USA

P-251 Differential Temporal Effects of Immunosuppressants on Clinical Outcomes in GAMT Deficient Mice Following Treatment with scAAV9.hGAMT
Robyn Binsfeld, Queen's University, Canada

P-252 Long-term LCAT replacement in a patient with familial LCAT deficiency in first in human clinical trial of ex vivo gene/cell therapy using autologous preadipocytes
Masayuki Kuroda, Center for Advanced Medicine, Chiba University Hospital, Japan

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Robert Thompson, Department of Neurology, Massachusetts General Hospital, USA / Center for Genomic Medicine, Massachusetts General Hospital, USA

P-254 Enhancing Cardiac Stem Cell Therapy via Mitochondria-Targeted Nanocarriers: A Novel Strategy for Myocardial Regeneration
Jiro Abe, Department of Pediatrics and Neuromascular Center, National Hospital Organization Hokkaido Medical Center, Japan / Department of Pediatrics, Hokkaido University Hospital, Japan / Laboratory for Molecular Design of Pharmaceutics, Faculty of Pharmaceutical Science, Hokkaido University, Japan

13. Lysosomal Disorders

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Akhil Bhalla, Denali Therapeutics Inc., USA

P-256 Breaking Barriers in Lysosomal Disorder Screening: A Novel simultaneous LC-MS/MS Approach for Tay-Sachs, Sandhoff, and GM1 Gangliosidosis
Stephan T. Hold, ARCHIMEDlife GmbH, Austria

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Marc Patterson, IntraBio Inc, USA

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Marc Patterson, IntraBio Inc, USA

P-259 An Analysis of Biomarkers for the Evaluation of Gene Therapy for Niemann-Pick Disease Type C
Chika Watanabe, Jichi Medical University, Japan

P-260 Variations in cholesterol storage, intracellular localization and trafficking pattern of NPC1 in Niemann-Pick type C
Kathrin Zotter, University Children's Hospital Zurich - Eleonorenstiftung, Switzerland

P-261 Safety and efficacy of arimoclomol in a pediatric substudy of Niemann-Pick disease type C patients aged 6 to < 24 months at study enrolment
Laila Arash-Kaps, SphinCS, Clinical Science for LSD, Germany

P-262 Disease severity analysis in the treatment of early onset Niemann Pick disease Type C1
Rebecca Jaeger, Division of Metabolic Disorders, Children's Hospital of Orange County, Rady Children's Health, USA / Graduate Program in Genetic Counseling, Feinberg School of Medicine, Northwestern University, USA / Department of Pediatrics, Rush University Medical Center, USA

P-263 Niemann Pick Type C associated IBD: A paediatric case series from a single centre
Chern Yan Tan, The Willink Metabolic Unit , UK

P-264 Efficacy results across an observational trial, a double-blind randomized trial of arimoclomol in Niemann-Pick type C patients treated with miglustat, and an open-label extension phase
Laila Arash-Kaps, SpinCS, Clinical Science for LSD, Germany

P-265 Clinical and Genetic Characterization of Acid Sphingomyelinase Deficiency Type AB in the French Cohort
Nathalie Guffon, Reference center for inherited metabolic disease, France

P-266 Early Diagnostic Trends in Acid Sphingomyelinase Deficiency Following the Implementation of Tandem Mass Spectrometry-Based Enzyme Assay in Japan: A Single-Center Experience
Akie Kato, Department of Pediatrics, Akita University Graduate School of Medicine, Japan

P-267 Long-term 24-month findings of N-acetyl-L-leucine for Niemann-Pick disease type C
Marc Patterson, IntraBio Inc, USA / Hospital of the Ludwig Maximilians University, Munich, Dept. of Neurology, Germany

P-268 Improvement in quality of life and general functions in pediatric acid sphingomyelinase deficiency patients after receiving olipudase alfa: a single-center experience in Taiwan
Yu-Wen Pan, National Cheng Kung University Hospital, Taiwan

P-269 Preemptive Diagnosis of Nieman-Pick disease type C and Rapid Response of Gelastic Cataplexy by N-Acetyl-l-Leucine: a Case Report
Kitiwan Rojnueangnit, Thammasat University, Thailand / Center of Medical Genomics, Thammasat University Hospital, Thailand

P-270 Alternative approaches for the pre-diagnosis and follow-up of lysosomal storage diseases: label free pre-diagnosis of Niemann Pick C using Raman and surface-enhanced Raman spectroscopy
Martino Calamai, European Laboratory for Non-Linear Spectroscopy (LENS), Italy / National Institute of Optics - National Research Council, Italy

P-271 Ten Years of Newborn Screening for Acid Sphingomyelinase Deficiency: Follow Up Data from the Illinois Cohort
Joshua J Baker, Ann & Robert H Lurie Children's Hospital of Chicago, USA

P-272 LSD Brazil Network: Advancing the Diagnosis of Lysosomal Storage Diseases in Brazil and Latin America
Franciele Barbosa Trapp, Casa dos Raros, Brazil

P-273 Sphingolipids, lyso-sphingolipids and their analogues as first-line biomarkers in the diagnosis, treatment and research of lysosomal storage disorders: utility of LC-MS lipidomics
Ladislav Kuchar, Research Unit for Rare Diseases, Department of Paediatrics and Inherited Metabolic Disorders, First Faculty of Medicine, Charles University and General University Hospital, Czech Republic

P-274 Enzyme Replacement Therapy in Resource-Limited Settings: Challenges and Outcomes in Paediatric Lysosomal Storage Disorders
Shilpa Jauhari, Command Hospital Air Force , India

P-275 Genetic Landscape, Founder Variants and Birth Prevalence of Lysosomal Storage Disorders in Qatar
Tawfeg Ben-Omran, Genetic and Genomic Medicine, Sidra Medicine, Qatar

P-276 Key lipid biomarkers in dried blood spots for the diagnosis and monitoring of lysosomal storage disorders
Isidro Arevalo-Vargas, Fundación para el Estudio y la Terapéutica de la Enfermedad de Gaucher y Otras Lisosomales (FEETEG), Spain / GIIS-012. Instituto de Investigación Sanitaria Aragón (IIS Aragón), Spain / Universidad de Zaragoza, Departamento de Bioquímica, Biología Molecular y Celular, Spain / Corporación Unificada Nacional de Educación Superior CUN, Colombia

P-277 Unlocking New Frontiers in LSD Biomarker Detection: Innovation from Matrices to Reference Ranges.
Amber Van Baelen, University of Antwerp, Belgium

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P-279 Nuclear transcription factor NF- K B in lymphocyte populations from children with Gaucher disease
Matvey Valerievich Konyashin, Federal State Autonomous Institution "National Medical Research Center for Children's Health" of the Ministry of Health of the Russian Federation, Russia

P-280 Increased contacts between lysosome and mitochondria: a potential pathogenic mechanism in neuronopathic Gaucher disease
Silvana Zanlungo, Silvana Zanlungo, Chile

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P-282 Massive Disease Burden in Infants with Gaucher Disease Type 1 and Type 3: Long-term Outcomes of Early Imiglucerase Therapy
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Ashlee R Stiles, Division of Medical Genetics, Department of Pediatrics, Duke University Medical Center, USA

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Clara Camacho dos Reis, Programa de Pós Graduação em Genética e Biologia Molecular da Universidade Federal do Rio Grande do Sul, Brazil

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Kagan calisgan, Istanbul University-Cerrahpasa, Cerrahpasa Medical School, Child Nutrition and Metabolism, Turkey

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Rohit Sadanand, All India Institute of Medical Sciences, India

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Ayse Yuksel Yanbolu, Ege University Faculty of Medicine Children's Hospital, Department of Pediatric Metabolism and Nutrition, Turkey

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Yanjun Liu, University of Galway, Ireland

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Hyo-Sang Do, Department of Pediatrics, University of Ulsan College of Medicine, Republic of Korea

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Beatrice Dufrusine, Center for Advanced Studies and Technology (CAST), University 'G. d'Annunzio' of Chieti-Pescara, Italy

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MOTOMICHI KOSUGA, Center for Medical Genetics, National Center for Child Health and Development, Japan

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Marzia Pasquali, Department of Pathology, University of Utah, USA / ARUP Laboratories, USA

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Tsubasa Oguni, Laboratories Division, Shimane University Hospital, Japan

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Norio Sakai, Center for Promoting Treatment of Intractable Diseases, ISEIKAI International General Hospital, Japan

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Clara Robin, Internal medicine department, INSERM 1253 « iBraiN », University of Tours, France

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Janey Roxanna Phelps, University of North Carolina School of Medicine, Dept of Anesthesiology, USA

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Anil B Jalan, NIRMAN, India

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Tetsumin So, National Center for Child Health and Development, Japan

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Burcu Ozturk-Hismi, Marmara University School of Medicine, Department of Pediatrics, Division of Pediatric Metabolic Diseases, Turkey

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Ebru Aypar, Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Cardiology, Turkey

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Julien Van Gils, Department of Medical Genetics, University Hospital of Bordeaux, France

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Mireia del Toro, Pediatric Neurology, Hospital Universitario Vall d'Hebron, Spain

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CARMEN CURIATI, REFERENCE CENTER IN INBORN ERROS OF METABOLISM/UNIVERSIDADE FEDERAL DE SAO PAULO, Brazil

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Maria Fuller, Genetics and Molecular Pathology, SA Pathology, Australia

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Huan Liang, Xinhua Hospital, Shanghai Institute for Pediatric Research, Shanghai Jiao Tong University School of Medicine, China

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Natalie Engmann, Denali Therapeutics Inc., USA

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Shunji Tomatsu, Nemours Children's Health, USA

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Shunji Tomatsu, Nemours Children's Health, USA

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Marie Daentl, ARCHIMEDlife GmbH, Austria

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Lucia Segura Schmitz, University Medical Center Mainz, Villa Metabolica, Germany

P-328 Genotypes, and Their Clinical and Biochemical Features of 179 Japanese patients with Fabry disease- Can clinical types be predicted based on genotypes?
Kaoru Eto, Tokyo Women's Medical University Adachi Medical Center, Japan / Advanced Clinical Research Center & Asian LSD Center, Institute of Neurological disorders, Japan

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Tomas Ripoll-Vera, Son LLatzer Universitary Hospital & IdISBa, Spain

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John A. Bernat, University of Iowa Health Care, USA

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Irene Koulinska, Chiesi Global Rare Diseases, USA

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Uma Ramaswami, Royal Free London NHS Foundation Trust, University College London, UK

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John A. Bernat, University of Iowa Health Care, USA

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P-336 Pegunigalsidase alfa cellular uptake, stability, lysosomal delivery and impact on mitochondrial and energetic dysfunction in Fabry cells
Jessica Gambardella, Federico II University of Naples, Italy

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Maryam Banikazemi, Department of Pediatrics, Division of Genetics, Metropolitan Hospital/New York Medical College, USA

P-338 A Decade of Fabry Disease in Japan: Insights from the Jikei Fabry Registry
Toshiki Tsunogai, The Jikei University School of Medicine, Department of Pediatrics, Japan

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P-340 Serum Immune Response to Globotriaosylceramide and Galabiosylceramide in Fabry Disease Patients Reveals Complement is Associated with Treatment Response and Cardiac Pathology
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P-341 Phenotypic Expression and Drug screening using NGN2-induced Neurons derived from Niemann Pick C (NPC)-iPSCs
Yoshikatsu Eto, Advanced Clinical Research Center, Southern Tohoku Institute for Neuroscience, Japan

P-342 Late-Onset Fabry disease as Major Clinical Phenotype-Clinical, Biochemical and Molecular Features in Japanese Fabry Patients with Late Onset.
Yoshikatsu Eto, Southern Tohoku Institute for Neuroscience/Tokyo Jikei University, Japan

P-343 High Risk and Newborn Screening (NBS) for Early Diagnosis of Neuronal Ceroid Lipofuscinosis (NCL) Types 1 and 2 Using Dried Blood Spot (DBS)
Yoshikatsu Eto, Advanced Clinical Research Center, Southern Tohoku Institute for Neuroscience/ Jikei University, Japan

P-344 Long term follow up of LysoGb3 in a Fabry pediatric cohort diagnosed by newborn screening
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Chung Lin Lee, Department of Pediatrics, MacKay Memorial Hospital, Taiwan

P-346 Extracellular Vesicles as New Mediators of Renal Pathology in Anderson Fabry Disease
Livia Lenzini, Department of Medicine, University of Padova, Italy

P-347 Cardiorespiratory Fitness and Physical Function in adult patients with Fabry Disease: A Cross-Sectional Multicenter Study
Nicola Vitturi, Division of Metabolic Diseases, Department of Medicine, University Hospital of Padova, Italy

P-348 Demoralization and Psychological Distress in Adult Patients with Fabry Disease: A Preliminary Study
Nicola Vitturi, Division of Metabolic Diseases, Department of Medicine, University Hospital of Padova, Italy

P-349 Development of a mouse-human-chimeric anti-α-galactosidase A monoclonal antibody as a reference for measuring antidrug antibody titers in patients with Fabry disease
Takahiro Tsukimura, Meiji Pharmaceutical University, Japan

P-350 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
Patricio Aguiar, Inborn Errors of Metabolism Reference Center, Portugal / Faculty of Medicine, Lisbon University, Portugal

P-351 Age-related progression of clinical symptoms and the importance of early intervention in 100 heterozygous females with Fabry disease
Kazuya Tsuboi, LSD Center, Nagoya Central Hospital, Japan

P-352 Dissecting the neuronal features of Fabry Disease
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Martha Caterina Faraguna, Pediatrics, Fondazione IRCCS San Gerardo dei Tintori, Monza, Italy / Center for Lysosomal and Metabolic Diseases, Department of Pediatrics, Erasmus MC University Medical Center, Netherlands

P-355 Can alpha-glucosidase in plasma and leukocytes serve as a surrogate marker for future gene therapy studies in classic infantile Pompe disease?
Martha Caterina Faraguna, School of Medicine and Surgery, University of Milano-Bicocca, Italy / Pediatrics, Fondazione IRCCS San Gerardo dei Tintori, Monza, Italy / Center for Lysosomal and Metabolic Diseases, Department of Pediatrics, Erasmus MC University Medical Center, Rotterdam, Netherlands

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Martina Bon, Regional Coordinating Center for Rare Diseases, University Hospital of Udine, Italy

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Sara Lucas Del Pozo, Charles Dent Metabolic Unit, The National Hospital for Neurology and Neurosurgery, UK

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Yohta Shimada, Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine, Japan

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William Kerr, Amicus Therapeutics UK Ltd, UK

P-360 Ambroxol hydrochloride monotherapy as pharmacological chaperone therapy in patients with neuronopathic Gaucher disease
Aya Narita, Department of Pediatrics, ISEIKAI International General Hospital , Japan / Division of Child Neurology, Institute of Neurological Science, Tottori University Faculty of Medicine, Japan

P-361 ATB200-19: an open-label, expanded access study of the safety and effectiveness of cipaglucosidase alfa plus miglustat in adults with late-onset Pompe disease in Japan
Aya Narita, Department of Pediatrics, ISEIKAI International General Hospital, Japan / Division of Child Neurology, Institute of Neurological Science, Tottori University Faculty of Medicine, Japan

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Priya S Kishnani, Duke University Medical Center, USA

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Rodolfo Tonin, Neuroscience and Medical Genetics Department, Meyer Children's Hospital IRCCS, Italy

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P-365 A Retrospective Study in a Large Cohort to Identify Predictive Clinical and Biological Markers of Lysosomal Acid Lipase Deficiency.
Irene Serrano-Gonzalo, Fundación Española para el Estudio y Tratamiento de la Enfermedad de Gaucher y otras lisosomales (FEETEG), Spain / Grupo de Investigación de Enfermedad de Gaucher (GIIS-012). IIS Aragón, Spain / Grupo de Investigación Mecanismos de Enfermedad Crónica e Investigación Traslacional (MECIT), Spain

P-366 Evaluating Lipocalin-2 as a Potential Biomarker of Liver Damage for Lysosomal Acid Lipase Deficiency
Irene Serrano-Gonzalo, Fundación Española para el Estudio y Tratamiento de la Enfermedad de Gaucher y otras lisosomales (FEETEG), Spain / Grupo de Investigación de Enfermedad de Gaucher (GIIS-012). Instituto de Investigación Sanitaria de Aragón, Spain / Grupo de Investigación Mecanismos de Enfermedad Crónica e Investigación Traslacional (MECIT), Spain

P-367 Testing for Metachromatic Leukodystrophy (MLD) in the Central and Eastern European (CEE) region
Cecilia Marinova, Medasol, Czech Republic

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Valeria Calbi, San Raffaele Telethon Institute for Gene Therapy (SR-Tiget), Italy / Pediatric Immunohematology Unit and BMT Program, IRCCS San Raffaele Scientific Institute, Italy

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Dominik Doerfel, University Children's Hospital, University Medical Center Hamburg-Eppendorf, Germany

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Takashi Watanabe, Department of Pathophysiology and Metabolism, Kawasaki Medical School, Japan

P-371 A quantitative UPLC-MS/MS-based oligosaccharide assay in dried blood spots for the diagnosis of alpha-mannosidosis
Simona Murko, University Children's Hospital, University Medical Center Hamburg-Eppendorf, Germany

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Goksu Demirbas, Basaksehir Cam and Sakura City Hospital, Turkey

P-373 The Real-World Journey of Patients with Alpha-Mannosidosis: A Retrospective US Claims Database Study
Robert J. Hopkin, Cincinnati Children's Hospital Medical Center, USA

P-374 Effectiveness and Tolerability of Migalastat in Adult Fabry Disease: A Single Regional Centre Experience
Eamon P McCarron, Adult Inherited Metabolic Diseases, Salford Care Organisation, Northern Care Alliance NHS Foundation Trust, UK

P-375 Endocrine Dysfunction in adults with Glycoproteinoses
Eamon P McCarron, Adult Inherited Metabolic Diseases, Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust, UK

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Adriana M Montano, Saint Louis University, USA

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Adriana M Montano, Saint Louis University , USA

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Chamal Palingu Imalke Kankanan Arachchige, Faculty of Medicine, University of Ruhuna, Sri Lanka

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Phillip Garretson, UPMC Division of Genetic and Genomic Medicine, Department of Pediatrics, University of Pittsburgh, USA

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Libo Wang, Shanghai Children's Medical Center School of Medicine, Shanghai Jiao Tong University, China

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Diego Martinelli, Division of Metabolic Diseases and Hepatology, Bambino Gesù Children's Hospital IRCCS, Italy

P-382 Interferon stimulated gene expression is a biomarker for primary mitochondrial disease
Nandaki Keshavan, UCL GOS Institute of Child Health, UK / Metabolic Unit, Great Ormond Street Hospital, UK

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Tomohiro Ebihara, Department of Neonatology, Chiba Children's Hospital, Japan

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Sayaka Ajihara, Department of Clinical Genomics, Saitama Medical University, Japan / Department of Pediatrics, Saitama Medical University, Japan

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Siew Li Ting, Willink Metabolic Unit, Genomic Medicine, St Mary's Hospital, UK

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Megan Dorman, Great Ormond Street Hospital, UK

P-387 Mild presentation of MT-ATP6: m.9176 T>G p.(L217R) Homoplasmic Pathogenic Variant (PV): spectral but not strictly causal
Anne Chun-Hui Tsai, University of Illinois, USA

P-388 Application value of urinary metabolic markers in the disease diagnosis and condition assessment of Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency
Yang Liu, Yang Liu,Department of Neurology,Beijing Children's Hospital,Capital Medical University,National Center for Children's Health,China, China

P-389 Molecular analysis of a mitochondrial disorder linked to a pathogenic variant in the ECHS1 (Enoyl-CoA Hydratase, Short-chain, 1) gene
Sanjhi Paliwal, Department of Pediatrics, Postgraduate Institute of Medical Education and Research (PGIMER), India

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Ha Thu Nguyen, Vietnam National Children's Hospital, Vietnam

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James Nurse, Southampton General Hospital, UK

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Matthew Thomas Snyder, University of Virginia - Pediatric Genetics, USA

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Wakako Kikuchi, Akita University Graduate School of Medicine, Japan

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Emma Louise Coombes, Genetic Metabolic Disorders Service, The Children's Hospital at Westmead, Australia

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Antri Savvidou, Department of Pediatrics, Institute of Clinical Sciences, Sahlgrenska Academy, University of Gothenburg, Sweden / Region Västra Götaland, Sahlgrenska University Hospital, Department of Pediatrics, Sweden

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Joél Smet, Ghent University, Mitochondrial Investigations Laboratory, Belgium

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Amna Ahmed, Department of Genetics, Sidra Medicine, Qatar / College of Health Sciences, Qatar University, Qatar

P-403 Fatal infantile presentation in a patient with MTO1 deficiency. Novel variant and discordant family phenotype.
Arianna Ko, Keck Graduate Institute, USA

P-404 Clinical challenges and importance of the genomic diagnosis of mitochondrial diseases in a cohort of Brazilian patients
Clarissa Bueno, Division of Child Neurology, Department of Neurology, University of Sao Paulo , Brazil

P-405 Clinical Spectrum and Long-Term Treatment Outcomes in Primary Coenzyme Q10 Deficiency-4 Due to COQ8A Mutations: A Case Series
Nuriye Ece Mintas, Muğla Sıtkı Koçman University Research Hospital, Department of Pediatric Metabolic Diseases, Turkey

P-406 Two Rare Cases: Mitochondrial DNA Depletion Syndrome in Siblings with a Novel Pathogenic SLC25A4 Variant
Goksu Demirbas, Basaksehir Cam and Sakura City Hospital, Turkey

P-407 SLC25A42 related mitochondrial disorder, case series from 19 new families.
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P-408 A novel and severe neonatal-onset phenotype of NDUFAF6
Sarah Hulley, Sheffield Children's Hospital, UK

P-409 Expanding the Clinical Spectrum of PARS2-Related Disorders: A Case Series and Functional Insights into Mitochondrial Dysfunction
Shubhnita Singh, Children's Hospital of Philadelphia, USA

P-410 3-methylglutaconic aciduria associated with deficiency in the mitochondrial i-AAA protease YME1L1
Petros P. Petrou, The Cyprus Institute of Neurology and Genetics/Biochemical Genetics Department, Cyprus

P-411 Clinical Characteristics and Influencing Factors of Children with MT-TL1 Gene m.3243A>G Mutation: A Phenotypic Study Based on 11 Han Chinese Families
Fan Yang, Shanghai Children's Medical Center, School of Medicine, Shanghai Jiao Tong University, China

P-412 L-citrulline treatment of nitric oxide deficiency in MELAS: a phase 1 dose-finding and safety study
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P-413 Genotype-phenotype correlations and clinical heterogeneity in Russian patients with single large-scale mitochondrial DNA deletions
Yulia Itkis, Research Centre for Medical Genetics, Russia

P-414 Results from a European multicenter study on single large-scale mitochondrial DNA deletion syndromes with adult onset
Kristoffer Bjorkman, University of Gothenburg, Institute of Clinical Sciences, Department of Pediatrics, Sweden

P-415 Clinical Spectrum of COXPD10 Due to MTO1 Variants: A Single-Center Experience from Turkey Including a Case of Uneventful Delivery
Asli Durmus, Trabzon Kanuni Training and Research Hospital, Turkey

P-416 COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 23: TWO TUNISIAN NEONATAL CASES
Kamel Monastiri, Teaching Hospital of Monastir, Tunisia

P-417 Combined oxidative phosphorylation deficiency 58 (COXPD58) caused by homozygous TEFM c.469C>G variant in multiple Indian patients, signatures of a founder mutation?
Sunita Bijarnia-Mahay, Sir Ganga Ram Hospital, India

P-418 Recognizing Primary Carnitine Deficiency: A Treatable Metabolic Cardiomyopathy with Variable Clinical Presentation
Sakina Mammadova, Ege University Medical Faculty Department of Pediatrics Division of Inborn Error of Metabolism and Nutrition, Turkey

P-419 Untargeted metabolomics and targeted analysis of sulfur metabolites in plasma of patients with mitochondrial optic neuropathies
Marketa Tesarova, Department of Pediatrics and Inherited Metabolic Disorders, First Faculty of Medicine, Charles University, and General University Hospital in Prague, Czech Republic

P-420 Surgical treatment of pediatric patients with intractable epilepsy with mitochondrial dysfunction
YOUNG-MOCK LEE, Department of Pediatrics, Gangnam Severance Hospital, Yonsei University College of Medicine, Republic of Korea

P-421 Current global vitamin and cofactor prescribing practices for primary mitochondrial diseases: Results of a European reference network survey
Karit Reinson, Department of Genetics and Personalized Medicine, Institute of Clinical Medicine, University of Tartu, Estonia

P-422 RETROSPECTIVE NATURAL HISTORY STUDY OF POLG DISEASE IN A MITOCHONDRIAL MEDICINE CLINICAL RESEARCH CENTER
Amy Goldstein, Children's Hospital of Philadelphia, USA

P-423 Integrative Clinical and Zebrafish Model Analysis of RARS2-Related Neonatal Mitochondrial Disease
Adiana Mutamsari Witaningrum, Department of Biochemistry and Molecular Genetics, Oita University Faculty of Medicine, Japan

P-424 Ethylmalonic and methylsuccinic acids impair mitochondrial bioenergetics and induce permeability transition in rat striatum
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil / PPG Ciências Biológicas: Fisiologia, Universidade Federal do Rio Grande do Sul, Brazil / PPG Neurociências, Universidade Federal do Rio Grande do Sul, Brazil

P-425 Mitochondrial Encephalopathy: First Indonesian Case Report of a MTND3 Causative Variant
Klara Yuliarti, Nutrition and Metabolic Disease Division, Department of Child Health, Cipto Mangunkusumo Hospital, Faculty of Medicine Universitas Indonesia, Indonesia

16. Neurotransmitter and Creatine Related Disorders

P-426 Early treatment in Sepiapterin Reductase Deficiency results in normal neurocognitive outcome: a case report
Maria Novelli, Sapienza University, Italy

P-427 A novel TH variant causes tyrosine hydroxylase deficiency in siblings from an isolated Cree community in northern Canada: a possible founder variant?
Lauren Badalato, Department of Pediatrics, Queen's University, Canada

P-428 A 19-months-old female patient with recurrent hypoglycemia, motor developmental delay and ptosis due to dopamine beta-hydroxylase deficiency: clinical and biochemical effects of treatment with Droxidopa
Bianca Panis, Department of Pediatrics, MosaKids Children's Hospital, Maastricht University Medical Centre, Netherlands

P-429 Cerebral Palsy as presenting misdiagnosis of Autosomal Recessive GCH-1 Dopa-Responsive Dystonia
Sandi Nugraha, Sandi Nugraha, Indonesia

P-430 Genetic Analysis in Cryptogenic Cerebral Palsy: A Prospective Study including Neurotransmitter genes
Oscar Mauricio Espitia Segura, HOMI Fundacion Hospital Pediatrico la Misericordia, Colombia / RICCNEP, Colombia

P-431 Non-Ketotic Hyperglycinemia: Case Series of Nine Neonates in a Tunisian NICU
Kamel Monastiri, Teaching Hospital of Monastir, Tunisia

P-432 Methenyltetrahydrofolate synthease deficiency (MTHFS deficiency): expanding the clinical and biochemical phenotype.
Sherry Fang, Department of Metabolic Medicine, Great Ormond Street Hospital, UK

P-433 Genotype-phenotype correlation and treatment outcome in dihydropteridine reductase (DHPR) deficiency across a single Paediatric UK centre
Sherry Fang, Department of Paediatric Inherited Metabolic Disease, Great Ormond Street Hospital for Children, UK

P-434 Tetrahydrobiopterin-Responsive Hyperphenylalaninemia: Treatment Approaches and Dietary Management with Identified Variants in Saudi Patients
Mai Saleh Labani, Department of Medical Genomics, Centre for Genomic Medicine, King Faisal Specialist Hospital and Research Centre (KFSHRC), Saudi Arabia

P-435 Eight Year Clinical Experience of Madopar in the Treatment of Tyrosine Hydroxylase Deficiency Caused by TH Gene Variation: A Single-Center Retrospective Cohort Study
Wanjun Lin, Women and Children's Medical Center Affiliated to Guangzhou Medical University, China

P-436 Diagnosis, management, and outcomes in patients with Hereditary Folate Malabsorption from a single centre.
Megan Dorman, Great Ormond Street Hospital, UK

P-437 The value of CSF neurotransmitter monitoring in the outcome of gene therapy in Aromatic Aminoacid Decarboxylase (AADC) defect
Filippo Manti, Department of Human Neuroscience, Sapienza University of Rome, Italy

17. New Diseases

P-438 Recurrent Paragangliomas Associated with a Novel Germline Variant in the Tricarboxylic Acid Cycle Gene DLST
Mickey Justin Myles Kuo, Ann & Robert H. Lurie Children's Hospital of Chicago, USA / Northwestern University Feinberg School of Medicine, USA / Eunice Kennedy Shriver National Institute of Child Health and Human Development, USA

P-439 A rare disease candidate for treatment in the differential diagnosis of congenital microcephaly, microcalcifications, hydrocephalus, and CP-like phenotype: FLVCR2-related vasculopathy and choline transport defect
Emine Genc, Zeynep Kamil Training and Research Hospital, Turkey

P-440 Biallelic SEPP1 Loss-of-Function is Associated to a Novel Adult-Onset Neurological Syndrome with Selenium Deficiency, Ataxia, Optic Atrophy, Neuropathy, and Deafness
Matheus Augusto Araujo Castro, Hospital das Clinicas, University of Sao Paulo School of Medicine, Brazil / Mendelics Genomic Analysis, Brazil

P-441 GSK3B-Dependent Dysregulation ofNeuronal Progenitor Proliferation and neurogenesis in OTUD5-Patient Induced Pluripotent Stem Cell Model
Na Xu, Shanghai Children's Medical Center, Shanghai jiao Tong University School of Medicine, China / Cell and Molecular Biology Laboratory, Zhoushan Hospital of Wenzhou Medical University, China

P-442 Systemic Selenoprotein Deficiency due to SECISBP2 mutation is a potentially treatable cause for 3-methylglutaconic aciduria type IV
Yew Sing Choy, Prince Court Medical Centre, Malaysia

P-443 De novo mutation in RAB11A is associated with neurodevelopmental disorder accompanied by variable multisystem disorders
Huiting Zhang, Peking University First Hospital, China

P-444 Combined malonic and methylmalonic aciduria: a novel cause of hyperinsulinism
Vincenza Gragnaniello, University of Padua, Italy

P-445 Heterozygous LONP1 Variant in a Patient with Mitochondrial Epileptic Encephalopathy: Broadening the Phenotypic Landscape
Maria Camila Solano Manzano, Postgraduate Student, Specialization in Medical Genetics, Faculty of Medicine, Pontificia Universidad Javeriana, Colombia

P-446 Expanded Newborn Screening by Tandem Mass Spectrometry in Thailand: From Early Detection to Treatment of Rare Diseases
Nithiwat Vatanavicharn, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand

P-447 AZ Experience of Increased UNSAT Samples With Expansion of NBS
Shagun Kaur, Phoenix Children's, USA / Department of Child Health, University of Arizona College of Medicine, USA

P-448 Expanded newborn screening in Health Regions 5 (Ratchaburi) and 13 (Bangkok) under Thailand´s Universal Health Coverage scheme supported by SiNBS webbased platform
Somporn Liammongkolkul, Division of Medical Genetics,Siriraj Hospital, Mahidol University, Thailand

P-449 Expanding newborn screening for treatable inherited disorders by using targeted genetic analysis
Nikki Wen Yan Fong, Genetics Service, Department of Paediatrics, KK Women's and Children's Hospital, Singapore / SingHealth Duke-NUS Genomic Medicine Centre, Singapore

P-450 Expanded Neonatal Screening for Inherited Metabolic Disorders in Russia: Predicted and Observed Incidence and Regional Differences
Daria Aleksandrova, Research Centre for Medical Genetics, Russia

P-451 Liquid chromatography-tandem mass spectrometry measurement of screening markers in dried blood spots for external quality control to ensure consistency among screening laboratories
Yosuke Shigematsu, Uji-Tokushukai Medical Center, Japan / University of Fukui, Japan

P-452 Enhancing Newborn Screening Systems in Tokyo: From Program Expansion to Consortium Formation.
Nobuyuki Ishige, Tokyo Health Service Association, Japan

P-453 Newborn Screening Quality Indicators and Outcomes in Thailand: Insights from a Newly Established Screening Center at Ramathibodi Hospital
Thipwimol Tim-Aroon, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand

P-454 Over 3 Decades of Organizing Services for Inherited Metabolic Disorders in Thailand
Pornswan Wasant, Faculty of Medicine Siriraj Hospital, Mahidol University , Thailand

P-455 Recommendation of 12 common IEMs for NBS Program in India: 25 Years of Mass-Spectrometry Experience in Metabolomic Screening in High-Risk Cases
Usha Pinakin Dave, MILS International India, India

P-456 Selective screening for inborn errors of metabolism in Vietnam: experience for 20 years
Dung Chi Vu, Vietnam National Children's Hospital, Vietnam

P-457 Two Years Experience of Inherited Metabolic Disorders Newborn Screening from Pakistan
Bushra Afroze, Department of Paediatrics and Child Health Aga Khan University Hospital , Pakistan

P-458 FALSE NEGATIVE IN NEWBORN SCREENING: BEWARE OF VITAMIN SUPPLEMENTATION
Francesco Tagliaferri, Foundation IRCCS Ca' Granda Ospedale Maggiore Policlinico, Clinical Metabolic Center, Italy

P-459 Communication of an abnormal metabolic newborn screening result in the Netherlands: the General Practitioner´s Perspective
Sietske Haitjema, Division of Metabolic Diseases, Beatrix Children's Hospital, University Medical Center Groningen, University of Groningen, Netherlands

P-460 A Nationwide Survey Investigating the Current Status of Genetic Counseling in Newborn Screening
Eri Sakai, Department of Medical Genetics, Osaka Metropolitan University Graduate School of Medicine, Japan

P-461 Review of infants diagnosed with an inborn error of metabolism (IEM) by newborn screening despite parental prenatal or preconception genomic carrier screening in Victoria, Australia
Bianca Morriss, Royal CHidrens Hospital, Australia

P-462 Expanded newborn screening for lysosomal disease in Kanagawa. Experience of a single institution.
Minami Ozawa, Department of Pediatrics,St. Marianna University School, Japan / Department of Genetic Medicine,St. Marianna University School of Medicine, Japan

P-463 Introduction of Neonatal Screening for Six Lysosomal Storage Diseases in Korea: Progress, Outcomes, and Challenges During the First Year of Implementation
A Young Park, Department of Pediatrics, Hallym University Hospital, Republic of Korea

P-464 A Decade of Newborn Screening for Mucopolysaccharidoses in Taiwan: Progress, Challenges, and Global Leadership Prospects
Chih-Kuang Chuang, MacKay Memorial Hospital, Taiwan

P-465 An innovative analytical platform to decode novel IDUA variants of uncertain significance identified by newborn screening for mucopolysaccharidosis type 1
Silvia Valentinuzzi, University G. d'Annunzio of Chieti-Pescara, Italy

P-466 Nine Years of Newborn Screening for Mucopolysaccharidosis Type II: Taiwan's Experience
Hsiang-Yu Lin, Department of Pediatrics, MacKay Memorial Hospital, Taiwan / International Rare Disease Center, MacKay Memorial Hospital, Taiwan / Department of Medicine, Mackay Medical College, Taiwan / Department of Medical Research, MacKay Memorial Hospital, Taiwan / Mackay Junior College of Medicine, Nursing and Management, Taiwan

P-467 Withdrawal

P-468 Screening for Fabry disease: not just for newborns
Alessandra Verde, University of Brescia, Italy

P-469 Ten Years of Pompe Disease Newborn Screening: A Single Center Experience
Allison M. Paltzer, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

P-470 Variant-Specific PCR Assays for Rapid Risk Assessment of Infantile-Onset Pompe Disease in Japanese Newborn Screening
Takaaki Sawada, Kumamoto University, Japan

P-471 Elevated ASM and IDS Activities as Novel Biomarkers for Mucolipidosis II/III in Newborn Screening
Kuan-Chi Tseng, National Taiwan University Hospital, Taiwan

P-472 Krabbe Disease in Infants Screened through Korean Newborn Screening since 2024: First Year Experience in a Single Center
Naeun Kwak, Department of Pediatrics, Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Republic of Korea

P-473 Regional differences in MCADD prevalence and ACADM gene variants: Insights from the first two years of newborn screening in Russia
Nikolai Bakin, Research Centre for Medical Genetics, Russia

P-474 Newborn screening for MCAD deficiency: 22 years of experience in Tuscany-Italy
Jacopo Venanzi, Neuroscience and Medical Genetics Department, Meyer Children's Hospital IRCCS, Italy

P-475 C14:1 isomers can be potential 2nd tier biomarkers to improve VLCADD positive predictive value in Newborn Screening
Michela Perrone Donnorso, LABSIEM-Pediatric Clinic Unit-DINOGMI, University of Genova, IRCCS Istituto Giannina Gaslini, Italy

P-476 Direct prediction of very-long chain acyl-CoA dehydrogenase deficiency (VLCADD) severity using newborn screening analyte data
Maaike Lenderink, Department of Metabolic diseases, University Medical Center Utrecht, Netherlands

P-477 Elevated C0 and C0/(C16+C18) ratio in pantothenate kinase-associated neurodegeneration: Possible biochemical markers in newborn screening
Emel Yilmaz-Gumus, Marmara University School of Medicine, Department of Pediatrics, Division of Inherited Metabolic Diseases, Turkey / Kocaeli City Hospital, Department of Pediatrics, Division of Inherited Metabolic Diseases, Turkey

P-478 Case Series of Six Patients with Positive Newborn Screening for CPT1 Deficiency: Investigating False Positive and Equivocal Results
Christopher Alexander Hardy, Department of Metabolic Medicine, Royal Children's Hopsital , Australia

P-479 Evaluating the use of propionyl-carnitine (C_3) in newborn screening in North Florida 2021-2024
Klaas J Wierenga, University of Florida, USA

P-480 Evaluation of newborn screening for diseases using 3-hydroxyisovalerylcarnitine (C5-OH) as a marker: systematic review and evaluation of 17 years of C5-OH screening in the Netherlands
Ryan Aukes, Department of Pediatrics, Division of Metabolic Disorders, Amsterdam Gastroenterology Endocrinology Metabolism, Amsterdam UMC, University of Amsterdam, Netherlands

P-481 New developments in neonatal mass screening for homocystinuria in Japan
Masayoshi Nagao, NHO Hokkaido Medical Center, Japan

P-482 International Survey on Phenylketonuria Newborn Screening
Domen Trampuz, Clinical Institute for Special Laboratory Diagnostics, University Children's Hospital, Ljubljana University Medical Center, Slovenia

P-483 Newborn screening for galactosemia in Hiroshima area of Japan
Reiko Kagawa, Hiroshima University Hospital, Azerbaijan

P-484 Early recognition of citrin deficiency: a newborn with elevated citrulline detected in newborn screening and acute liver failure
Hoi Yin Chan, Department of Pediatrics and Adolescent Medicine, Hong Kong Children's Hospital, Hong Kong / Medical Faculty, Radboud University Nijmegen, Netherlands

P-485 Newborn screening for citrin deficiency
Teck Wah Ting, Genetics Service, Department of Paediatrics, KK Women's and Children's Hospital, Singapore

P-486 Establishing CK-MM reference ranges for Low-Birth-Weight Infants for Duchenne Muscular Dystrophy Newborn Screening
Mari Mori, Ohio State University, USA

P-487 Advancing neonatal screening for pyridoxine-dependent epilepsy through combined analysis of butylated 2-OPP, 6-oxo-pipecolate and pipecolate
Joseph Paul Dewulf, Biochemical Genetics and Newborn Screening Laboratory, Department of Laboratory medicine, Cliniques universitaires Saint-Luc, UCLouvain, Belgium / Institut des Maladies Rares, Cliniques universitaires Saint-Luc, UCLouvain, Belgium

P-488 IFI44L and other Interferon Signaling Cytokines are Detectable in Aicardi Goutieres Syndrome Blood Spots.
Adeline Vanderver, Division of Neurology, Department of Pediatrics, Childrens Hospital of Philadelphia, USA

P-489 Detecting amino acid disorders in the Philippines via pre-column derivatization followed by reversed-phase UPLC: a 10-year experience
Dahlia Del Castillo Apodaca, University of the Philippines-Manila, Philippines

P-490 The impact of gestational diabetes mellitus on neonatal screening results via tandem mass spectrometry
Nopporn Sawatjui, Clinical Laboratory Section, Srinagarind Hospital, Faculty of Medicine, Khon Kaen University, Thailand / Srinagarind Excellent Laboratory, Faculty of medicine, Khon Kaen University, Thailand

P-491 Genetic diagnosis and genotype-phenotype association in 126 Brazilian individuals with reduced biotinidase activity
Devora Natalia Randon, Postgraduate Program in Genetics and Molecular Biology, Universidade Federal do Rio Grande do Sul, Brazil / Laboratory of Basic Research and Advanced Investigations in Neurosciences (BRAIN), Experimental Research Service, Hospital de Clínicas de Porto Alegre, Brazil

P-492 Amino acid and acylcarnitine profiles in infants with Down syndrome identified through expanded newborn screening
Kanda Sornkayasit, Center of Excellence in Precision Medicine, Srinagarind Hospital, Khon Kaen University, Thailand

P-493 Enhancing Neonatal Screening Strategies: A Machine Learning Approach for Improved Diagnostic Process
Maria Lucia Tommolini, University G. d'Annunzio of Chieti-Pescara, Italy

P-494 Newborn screening and the diagnosis of rare diseases: an ambispective study from the Brazilian Rare Diseases Network
Julia Cordeiro Milke, Universidade Federal do Rio Grande do Sul (UFRGS), Brazil / Hospital de Clínicas de Porto Alegre, Brazil / Rede Nacional de Doenças Raras, Brazil

P-495 Ten-Year Experience with Expanded Newborn Screening for Inborn Errors of Metabolism at a Malaysian University Hospital: Challenges, Outcomes, and Opportunities
Everlyn Coxin Siew, Genetic and Metabolism Unit, University Malaya, Malaysia

19. Novel Diagnostic/Laboratory Methods Including Omics

P-496 Untargeted metabolomic and proteomic profiling on dried blood spot to shed light on acid sphingomyelinase deficiency and the effects of enzyme replacement therapy
Silvia Valentinuzzi, University G. d'Annunzio of Chieti-Pescara, Italy

P-497 Untargeted metabolomics analysis of dried blood spots to identify potential metabolic biomarkers in Biotinidase deficiency
Ahmad Alodaib, Metabolomics Section, Precision Medicine Laboratory Department, Genomic Medicine Center of Excellence, King Faisal Specialist Hospital and Research Centre (KFSHRC), Saudi Arabia / The Department of Biochemistry and Molecular Medicine, College of Medicine, Alfaisal University, Saudi Arabia / 3 Genetic and Genomic Medicine Division, Department of Pediatrics, UPMC Children's Hospital of Pittsburgh, Saudi Arabia

P-498 Untargeted metabolomics profiling in dried blood spots as a tool to identify potential metabolic biomarkers in Carnitine palmitoyltransferase I
Ahmad Alodaib, Metabolomics Section, Department of Clinical Genomics, Center for Genomics Medicine, King Faisal Specialist Hospital and Research Centre (KFSHRC), Saudi Arabia / The Department of Biochemistry and Molecular Medicine, College of Medicine, Alfaisal University, Saudi Arabia / Genetic and Genomic Medicine Division, Department of Pediatrics, UPMC Children's Hospital of Pittsburgh, Saudi Arabia

P-499 Untargeted metabolomics analysis of dried blood spots of primary carnitine deficiency to identify potential metabolic biomarkers
Ahmad Alodaib, Metabolomics Section, Department of Clinical Genomics, Center for Genomics Medicine, King Faisal Specialist Hospital and Research Centre (KFSHRC), Saudi Arabia / The Department of Biochemistry and Molecular Medicine, College of Medicine, Alfaisal University, Saudi Arabia / Genetic and Genomic Medicine Division, Department of Pediatrics, UPMC Childrens Hospital of Pittsburgh, USA

P-500 Proteomic Analysis of Blood as a Novel Tool for Diagnosis and Screening of Citrin Deficiency
Yoichi Wada, Tohoku University Hospital, Japan

P-501 Spatial Proteomics for Enhanced Diagnosis of Mitochondrial Disease
Simon Wetzel, Division of Molecular Metabolism, Department of Medical Biochemistry and Biophysics, Karolinska Institute, Sweden / Centre for inherited metabolic diseases (CMMS), Karolinska University Hospital, Sweden

P-502 Towards Routine Application of Blood RNA-Seq in Genetic Testing: Evaluating the Impact of Globin Depletion, Inter-Batch Variability and the Choice of Specimen Collection
Xinyi Lu, Clinical Genetics Center, Shanghai Institute for Pediatric Research, Xinhua Hospital affiliated to Shanghai Jiao Tong University School of Medicine, China

P-503 Additional diagnostic yield through the analysis of short tandem repeats based on exome sequencing data
Shiyi Xu, Clinical Genetics Center, Xinhua Hospital, Shanghai Jiao Tong University School of Medicine, Shanghai, China., China / Shanghai Institute for Pediatric Research, Shanghai Jiao Tong University School of Medicine, China

P-504 Clinical Applications of a Rapid Real-Time Whole Genome Sequencing Analysis System
Yun-Ru Chen, Genetic Consultant Center Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan / Department of Pediatrics, Taipei Veterans General Hospital, Taiwan

P-505 Lipid signature in a mitochondrial trifunctional protein deficiency mouse model
Eduardo Vieira Neto, Division of Genetic and Genomic Medicine, School of Medicine, University of Pittsburgh, USA / UPMC Children's Hospital of Pittsburgh, USA

P-506 Diagnostic tool for patients with ornithine transcarbamylase deficiency lacking genetic confirmation
Alexander Laemmle, Division of Pediatric Endocrinology, Diabetology and Metabolism, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland / University Institute of Clinical Chemistry, Inselspital, Bern University Hospital, University of Bern, Switzerland / Department of Biomedical Research, University of Bern, Switzerland

P-507 Methylcitrate to Citrate Ratio as a Novel Biomarker for Inborn Errors of Propionate Metabolism: Preliminary Report on Clinical Utility
Osama Y Al-Dirbashi, Department of Laboratory Medicine and Pathology, Hamad Medical Corporation, Qatar / College of Health Sciences, Qatar University, Qatar / College of Health & Life Sciences, Hamad Bin Khalifa University, Qatar

P-508 LC-MS-based Proteomics on iPS Cell-Derived Neurons of Niemann-Pick Type C (NPC)
Takeo Iwamoto, Advanced Clinical Research Center, Southern Tohoku Research Institute for Neuroscience, Japan

P-509 Application of N-hexadecanoyl-sulfatide analysis by ultra-performance liquid chromatography-tandem mass spectrometry in dried blood spots, serum/plasma for mass screening of metachromatic leukodystrophy (MLD) in Japan
Takeo Iwamoto, Advanced Clinical Research Center, Institute of Neurological Disorders, Japan

P-510 Next-Generation Biomarker Analysis: A 6-Minute LC-MS/MS Assay for Alpha-Mannosidosis Oligosaccharide Monitoring
Stephan T. Hold, ARCHIMEDlife GmbH, Austria

P-511 High resolution mass spectrometry as a tool for semi-targeted next-generation metabolic profiling - challenges and solutions
Mihkel Ilisson, Department of Laboratory Genetics, Laboratory of Metabolic Diseases, Genetics and Personalized Medicine Clinic, Tartu University Hospital, Estonia / Department of Genetics and Personalized Medicine, Institute of Clinical Medicine, University of Tartu, Estonia

P-512 Retrospective comparative analysis of inborn errors of metabolism diagnosis before and after genomic sequencing availability in limited resources country
Cut Nurul Hafifah, Division of Nutrition and Metabolic Disease, Dept of Child Health, Faculty of Medicine, Universitas Indonesia, Indonesia / Human Genetic Research Cluster Indonesian Medical Education and Research Institute Faculty of Medicine Universitas Indonesia, Indonesia

20. Nursing in Metabolism

P-513 On-Call Service Demand for Paediatric Inherited Metabolic Disorders
Troy Dalkeith, Faculty of Medicine & Health, The University of Sydney, Australia

P-514 Dietary Management and the Patient Assessment of Chronic Illness Care (PACIC) Among Families of Children with Inborn Errors of Metabolism: A Fact-Finding Survey in Japan
Yuko Matsumoto, Department of nursing, Kagawa Prefectural University of Health Sciences, Japan

21. Organic Acidurias

P-515 Efficacy of liver transplantation in propionic acidemia : long-term follow-up in a French pediatric reference center
Tristan Mekdade, Reference Center for Inherited Metabolic Diseases, Necker Enfants Malades Hospital, APHP, Filière G2M, MetabERN, Paris Cité University, France / Pediatric Metabolic Unit, Pediatrics, Woman-Mother-Child Department, Lausanne University Hospital, Switzerland

P-516 Application of Carglumic acid in the prevention and treatment of hyperammonemia caused by organic acidemias
Hui Dong, Children's Medical Center, Peking University First Hospital, China

P-517 Initial Presentation and Long-Term Outcomes of Propionic Acidemia: A Retrospective Study from a Tunisian Pediatric Cohort
Meriem Mediouni, La Rabta University Hospital, Tunisia

P-518 Mathematical Modeling of Mitochondrial Metabolite Overflow-Induced Neurovascular Uncoupling in Propionic Acidemia: An Agent-Based Spatiotemporal Simulation
Rifaldy Fajar, Mathematical and Computational BioMedicine Research Group, The Integrated Mathematical, Computational, and Data Science for BioMedicine Research Foundation, Indonesia

P-519 Generation and characterization of brain organoids from iPSC-based PCCA and PCCB knockout lines: A step towards novel propionic acidemia model system
Eva Richard, Centro de Biologia Molecular Severo Ochoa (UAM-CSIC), Spain

P-520 Aortopexy Prior to Liver Transplantation in an Infant with Propionic Acidemia and Tracheomalacia: A Case Report
Shogo Shioda, Department of Endocrinology and Metabolism, National Center for Child Health and Development (NCCHD)., Japan

P-521 Model-Informed Dose Selection for the Pivotal Study of mRNA-3705 in Methylmalonic Acidemia
Min Liang, Moderna, Inc., USA

P-522 Clinical Characteristics and Gene Mutation Analysis of 79 Patients with Methylmalonic Acidemia
Jianmei Yang, Department of Paediatric Endocrinology, Shandong Provincial Hospital affiliated to Shandong First Medical University, China

P-523 Bone Health in Patients with Isolated Methylmalonic Acidemia
Solaf Mohamed Elsayed, Medical Genetics Department, Egypt

P-524 Long term outcomes of Methylmalonic Acidemia in a pediatric Tunisian series
Samia Elouertani, Department of Pediatrics and Inherited Metabolic Diseases, La Rabta Hospital, Tunisia

P-525 Influence of cell type and disease-causing variant on mitochondrial energy production in methylmalonic aciduria
Miriam Alina Güra, University Children's Hospital Zurich, Switzerland / University of Zurich, Switzerland

P-526 Hidden Cases of Methylmalonic Aciduria: When You Don't See C3 on NBS
Annabel Oliveira, NSW Biochemical Genetics Service, Sydney Children's Hospitals Network, Australia

P-527 Mapping TCA Cycle Dysregulation in Methylmalonic Acidemia Using In Vivo Stable Isotope Tracing.
Florian Traversi, Division of Metabolism, University Children's Hospital Zurich, Switzerland

P-528 Retrospective Chart Review of Patients with Disorders of Intracellular Cobalamin Metabolism
Julia Eazer, University of South Florida Morsani College of Medicine, USA

P-529 Subcutaneous administration of hydroxocobalamin improves treatment adherence in patients with defects in the intracellular processing of vitamin B12
Ana Morais, Pediatric Nutrition and Metabolic Diseases Unit. Hospital Universitario La Paz, Spain

P-530 Development of High Strength 50mg/2ml Hydroxocobalamin Injections to Treat Cobalamin C Deficiency
Sam Whiting, Bristol Children's Hosptial, UK

P-531 Maternal UPD of chromosome 12 leading to a homozygous pathogenic variant in MMAB and cobalamin B deficiency
Erin Falsey, University of Illinois Chicago, USA

P-532 21 cases of cblC deficiency with renal damage in China
Yao Zhang, Peking University First Hospital, China

P-533 Successful biomarkers control and pharmacokinetics following hydroxocobalamin dose intensification treatment, in 5 patients with early-onset cblC deficiency: quantum-biochemistry hypothesis?
Emmanuel Scalais, Department of Pediatrics, Division of pediatric neurology, Centre Hospitalier de Luxembourg, Luxembourg

P-534 Cobalamin E defect and end stage renal disease requiring renal transplant A cause or a coincident
Rihab Salih, Prince Sultan military medical city, Saudi Arabia

P-535 Case Series: Perinatal-Onset Methylmalonic Acidemia with Homocystinuria
Xiaohong Shang, Shandong Provincial Hospital Affiliated to Shandong First Medical University, China

P-536 Clinical Application of Carglumic Acid in Chinese Pediatric Organic Acidemia Patients
Xiaohong Shang, Shandong Provincial Hospital Affiliated to Shandong First Medical University, China

P-537 Perioperative management of liver and combined liver-kidney transplantation for methylmalonic and propionic acidemia
Clothilde Marbach, Department of Gastroenterology, Hepatology, Nutrition, and inborn errors of metabolism, children's hospital , France

P-538 Oxidative and neurodegenerative biomarkers in patients with propionic and methylmalonic acidemias: effect of L-carnitine treatment
Bianca Gomes dos Reis, Medical Genetics Service of the Hospital de Clínicas de Porto Alegre, Brazil / Federal University of Rio Grande do Sul, Brazil

P-539 Determination of serum N-acetylglutamate concentrations in patients with methylmalonic acidemia and propionic acidemia
Yasuhiro Maeda, Open Facility Center, Fujita Health University, Japan

P-540 Variant-specific response to flavin adenine dinucleotide in glutaryl-CoA dehydrogenase: Implications for enzymatic function and thermal stability
Nicole Lewandowski, University Medical Center Hamburg, Germany

P-541 Toward the First Pharmacological Therapy for Glutaric Acidemia Type 1: Allosteric Chaperone UCR422 Restores GCDH Function
Charlotte Schultehinrichs, University Children's Research, UCR@Kinder-UKE, University Medical Center Hamburg-Eppendorf, Germany

P-542 Clinical and Biochemical Outcomes in Adults with Glutaric Aciduria Type 1: A Retrospective Descriptive Study.
Maria Arbelo Rodriguez, Hospital Universitario Nuestra Señora de la Candelaria, Spain

P-543 Clinico-radiological profile of Indian children with type 1 Glutaric aciduria at a Tertiary care referral centre in Southern India
Balamurugan Nagarajan, Pediatric Neurology Consultant , Rainbow Children's hospital, India

P-544 Classical Multiple Carboxylase Deficiency Presenting with Unusual Urine Organic Acid Findings: A Diagnostic Challenge
Chizuko Nakamura, Department of Endocrinology and Metabolism, Nagano Children's Hospital , Japan / Life Science Research Centre, Nagano Children's Hospital , Japan

P-545 Propionic Acid compromises cell metabolic viability, reduces glutathione content and changes proteomic profile in MO3.13 human oligodendrocyte cell line - Insights for Propionic Acidemia
Cesar A. J. Ribeiro, Universidade Federal do ABC, Brazil

P-546 Isovaleric acid compromises cell viability and alters respiratory parameters in an AMPK-related mechanisms in human SH-SY5Y neuroblastoma cells
Cesar A. J. Ribeiro, Universidade Federal do ABC, Brazil

P-547 Clinical characteristics, genotypes and follow-up analysis of 37 Chinese children with isovaleric acidemia
Yuning Sun, Department of Pediatric Endocrinology and Genetic Metabolism, Xinhua Hospital Affiliated to Shanghai Jiao Tong University School of Medicine, China

P-548 Urine Organic Acids Analysis as a Second-tier Test for Abnormal Newborn Screening for Organic Acidemia: Two Years of Experience at Ramathibodi Hospital, Thailand
Areeporn Sangcakul, Department of Pathology, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand

P-549 Protective effects of cannabidiol against energy and redox homeostasis disruption and neurodevelopment alterations induced by 3-hydroxy-3-methylglutaric acid in neonatal rats
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil / PPG Ciências Biológicas: Fisiologia, Universidade Federal do Rio Grande do Sul, Brazil / PPG Neurociências, Universidade Federal do Rio Grande do Sul, Brazil

P-550 Experimental evidence that L-carnitine prevents neuroinflammation and neurodevelopmental impairment caused by acute increase of D-2-hydroxyglutaric acid concentrations in brain of neonatal rats
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Departamento de Bioquímica, ICBS, UFRGS, Brazil

P-551 Bezafibrate prevents bioenergetic dysfunction, neuronal loss, astrocyte reactivity, and neuroinflammation induced in vivo by D-2 hydroxyglutaric acid in the cerebral cortex of young rats
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Departamento de Bioquímica, ICBS, UFRGS, Brazil

P-552 Evidence that methylmalonic acid induces oxidative stress, bioenergetic disruption and glial reactivity and disturbs amino acid profile in rat striatum
Guilhian Leipnitz, PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil / PPG Ciências Biológicas: Fisiologia, Universidade Federal do Rio Grande do Sul, Brazil / PPG Neurociências, Universidade Federal do Rio Grande do Sul, Brazil

P-553 Valine Restriction Extends Survival in a Drosophila Model of Short-Chain Enoyl-CoA Hydratase 1 (ECHS1) Deficiency
Sarah Mele, La Trobe University, Australia

P-554 Isobutyryl-coenzyme A dehydrogenase deficiency: Disease, or non-disease, that is the question
Joern Oliver Sass, Research Group Inborn Errors of Metabolism, Department of Natural Sciences & Institute for Functional Gene Analytics (IFGA), Bonn-Rhein-Sieg University of Applied Sciences, Germany

P-555 Untargeted Metabolomics Facilitates the Diagnosis of SLC13A5-Related Developmental Epileptic Encephalopathy with Amelogenesis Imperfecta in Consanguineous Siblings
Marwan Shinawi, Washington University School of Medicine, USA

P-556 Long-Term Follow-Up of Six Japanese Patients with Holocarboxylase Synthetase Deficiency Reveals Developmental Delay and Persistent Intractable Eczema
Natsuko Arai-Ichinoi, Tohoku University Hospital, Japan

P-557 Clinical and molecular spectrum of an Indian cohort with Methylmalonic acidemia
Mounika Endrakanti, All India Institute of Medical Sciences, India

BP-01 ~ BP-32 ➡ Exhibition & Poster 2 (Event Hall)
P-01 ~ P-557 ➡ Poster 1 (Annex Hall)
P-558 ~ P-668 ➡ Exhibition & Poster 2 (Event Hall)

22. Peroxisomal, Sterol, Bile Acid, Lipid and Lipoprotein Metabolism

P-558 Searching for plasma proteins as clinical type-specific markers for X-linked adrenoleukodystrophy (X-ALD)
Shigeo Takashima, iGCORE, Gifu University, Japan / Life Science Research Center, Gifu University, Japan / COMIT, Gifu University, Japan / United Graduate School of Drug Discovery and Medical Information Sciences, Gifu University, Japan

P-559 Hepatic phenotype of ELOVL1-related disorders
Anne Chun-Hui Tsai, university of Illinois, USA

P-560 A novel PEX13 variant causes Zellweger spectrum disorder with mild/intermediate phenotype and cystic leukoencephalopathy
Lara M Marten, Department of Pediatrics and Adolescent Medicine, University Medical Center Goettingen, Germany

P-561 Establishing Local Diagnostic Capabilities for Peroxisomal Disorders: Development of a GC-MS/MS Method for Biomarker Quantification in the Philippines
Grace Ann Gonzales Samson, University of the Philippines, Philippines

P-562 Mosaic ABCD1 in Two Males
Sebastian S Hanna, University of California, Irvine, Department of Neurology, USA

P-563 Does essential fatty acid deficiency alter plasmalogen levels? Implications for the laboratory evaluation of peroxisomal disorders
Irene De Biase, Department of Pathology, University of Utah School of Medicine, USA

P-564 The effect of plasmalogens replacement therapy on bone metabolism, psychomotor development, and GPCR21 activation in a case of RCDP type 2
Hiromi Nyuzuki, Niigata University Medical and Dental Hospital, Japan

P-565 Identification of an oxidized lipid in plasma from patients with X-ALD
Kotaro Hama, Faculty of Pharmaceutical Sciences, Teikyo University, Japan / Advanced Comprehensive Research Organization (ACRO) Teikyo University, Japan

P-566 Effect of combined therapy with N-acetylcysteine, coenzyme Q10 and beta-cyclodextrin nanoparticles on mitochondrial redox status, cholesterol levels and oxidative damage in Niemann-Pick type C1 patients
Franciele Fatima Lopes, Hospital de Clínicas de Porto Alegre, Brazil

P-567 Bile acid biosynthetic defect causing by HSD3B7 deficiency with G6PD deficiency in neonatal onset severe cholestatic liver disease
Pattima Pakhathirathien, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand / Department of Pediatrics, Faculty of Medicine, Prince of Songkla University, Thailand

P-568 Glycerol-3-phosphate dehydrogenase 1 (GPD1) deficiency - redefining the phenotype as a cause of persistent hepatic steatosis and hypertriglyceridaemia
Michael Gunn, Department of Paediatric Inherited Metabolic Diseases, Evelina London Children's Hospital, UK

P-569 Use of Cholic Acid in Smith-Lemli-Opitz Syndrome (SLOS): Real-world Patient Outcomes
Paul Hillman, Department of Pediatrics, McGovern Medical School at the University of Texas Health Science Center at Houston (UTHealth) and Children's Memorial Hermann Hospital, USA

P-570 A Case of Chylomicron Retention Disease: Utilization of Rapid Whole Genome Sequencing to Manage Intervention-Resistant Failure to Thrive
Jennifer Burton, University of Illinois College of Medicine, USA

P-571 Impact of Cholic Acid Therapy on Growth, Cognitive Abilities, and Psychosocial Development in Smith-Lemli-Opitz Syndrome (SLOS)
Paull Hillman, Department of Pediatrics, McGovern Medical School at the University of Texas Health Science Center at Houston (UTHealth) and Children's Memorial Hermann Hospital, USA

P-572 A case of chylomicron retention disease with neuromuscular manifestations. A new mutation.
M Teresa Cardoso, Inherited Metabolic Disorders Reference Centre, Internal Medicine Department, São João University Hospital Center, Portugal

P-573 Sitosterolemia: Different Forms of Presentation in Childhood
Mariela Mercedes De Los Santos Mercedes, Paediatric, Hepatology and Nutrition Service, Hospital Sant Joan de Deu, Spain

P-574 The Severity and Burden of Psychiatric Symptoms in Patients with Cerebrotendinous Xanthomatosis (CTX): A US-based survey
Rana Dutta, Mirum Pharmaceuticals, Inc., USA

P-575 Neutral Lipid Storage Disease Due to a Homozygous PNPLA2 Frameshift Variant: A Multisystemic Presentation with Cardiomyopathy and Pancreatitis
Merve Yoldas Celik, Adana City Training and Research Hospital, Department of Pediatric Metabolism, Turkey

P-576 Smith-Lemli-Opitz Syndrome: Results Of Clinical Observation Of 11 Russian Patients
Olga Borisovna Kondakova, National Medical Research Center for Children's Health, Ministry of Health of Russia, Russia

P-577 Sitosterolemia as a treatable cause of chronic thrombocytopenia: Beyond ITP-results of a targeted screening study
Tanyel Zubarioglu, İstanbul University-Cerrahpaşa, Cerrahpaşa Medical Faculty, Department of Pediatrics, Division of Pediatric Nutrition and Metabolism, Turkey

P-578 20 Years of Universal Cholesterol Screening in Children and Adolescents - Benefits for Early Detection and Treatment of Familial Hypercholesterolemia
Jana Saligova, Children's Faculty Hospital Kosice, Slovakia

P-579 DESCRIPTION OF THE X-LINKED ADRENOLEUKODYSTROPHY COHORT FROM THE CSUR UNIT OF ADULT METABOLIC DISORDERS AT VIRGEN DEL ROCIO UNIVERSITY HOSPITAL (SEVILLE)
Alfonso Manuel Soto-Moreno, Hospital Universitario Virgen del Rocio, Spain

P-580 Atypical Biochemical Presentation in Genetically Confirmed Cerebrotendious Xanthomatosis Presenting as Neonatal Jaundice
Maya Fowler, School of Medicine, OHSU, USA

P-581 Short-Term Outcomes Following Abnormal Newborn Screening for Adrenoleukodystrophy in Taiwanese Male
Yin-Hsiu Chien, Department of Medical Genetics, National Taiwan University Hospital, Taiwan

23. Phenylketonuria

P-582 ENZYME SUBSTITUTION THERAPY IN PATIENTS WITH CLASSICAL PHENYLKETONURIA: RUSSIAN EXPERIENCE
Tatiana V Bushueva, National Medical Research Center of Children's Health, Ministry of Health of the Russian Federation, Russia / Research Centre for Medical Genetics, Russia

P-583 Pegvaliase treatment in 18 adult patients with classical phenylketonuria, part 2: adverse event profile and management
Chika Takano, Department of Pediatrics and Child Health, Nihon University School of Medicine, Japan / Division of Microbiology, Department of Pathology and Microbiology, Nihon University School of Medicine, Japan

P-584 Real-world Safety and Tolerability of Pegvaliase: A Non-interventional Surveillance Study in Japan
Mika Ishige, Pediatrics and Child Health, Nihon University School of Medicine, Japan

P-585 Pegvaliase Therapy During Pregnancy in Patients with Phenylketonuria: Safety and Efficacy Outcomes in Two Pregnancies
Erika R Vucko, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

P-586 PALLADIUM: A Phase 4 Study to Evaluate a Rapid Drug Desensitization Protocol for Adults with Phenylketonuria Experiencing Hypersensitivity Reactions to Pegvaliase
Kristin Lindstrom, BioMarin Pharmaceutical Inc., USA

P-587 Effect of Long-Term Sepiapterin Treatment on Dietary Phenylalanine Tolerance in Patients with Phenylketonuria: Interim Results from the APHENITY Extension Study
Francjan van Spronsen, Division of Metabolic Diseases, Beatrix Children's Hospital, University Medical Center Groningen, University of Groningen, Netherlands

P-588 High-Dose Sapropterin Therapy in Phenylketonuria: Outcomes and Considerations
Kathryn Lynn Eckert, University of Utah, USA

P-589 Sepiapterin Treatment and PKU-QOL Outcomes in Children, Adolescents and Adults with PKU: Results From the Dietary Phenylalanine Tolerance Subgroup in the APHENITY Long-Term Extension Study
Heidi Peters, Department of Metabolic Medicine, The Royal Children's Hospital, Australia

P-590 Extended Evaluation of BH4 Responsiveness in Taiwanese Phenylketonuria Patients: Insights into Genotype-Phenotype Correlations and Novel Responsive Mutations
Yung-Hsiu Lu, Department of Pediatrics,Taipei Veterans General Hospital, Taiwan

P-591 GLytactin EfficiEncy in non treated adult PHENylketonuria patients: the GLEEPHEN randomised controlled Trial
Adrien Bigot, Internal Medicine Department, CHU Tours, Hopital Bretonneau, France

P-592 Effect of Long Neutral Amino acid supplementation in Paediatric PKU patients in India
Ketki V Kudalkar, NIRMAN, India

P-593 Lifetime Monitoring of Phe Levels in PKU from Birth to Adulthood in the Swedish Registry for Inherited Metabolic Diseases
Andreas Kindmark, Department of Medical Sciences, Uppsala University Hospital, Sweden

P-594 Work Ability in Adults with PKU in Sweden in 2020
Andreas Kindmark, Department of Medical Sciences, Uppsala University Hospital, Sweden

P-595 Neuropsychiatric Comorbidities in Adolescents with PKU in the United States
Karly S. Louie, BioMarin UK Ltd., UK

P-596 Agreement between the Amsterdam Neuropsychological Tasks and Cambridge Neuropsychological Test Automated Battery in the assessment of PKU-patients and controls: A study related to the FDPD^{+}-databank
Ellis Marleen van Steenis, University Medical Center Groningen, Netherlands

P-597 Safety of breastfeeding in PKU during the first 6 months of life: comparison of two different approaches
Federico Baronio, Inherited Metabolic Disease and Newborn screening Unit , Pediatric Unit , IRCCS AOUBO, Metab-ERN Center for Rare Metabolic Conditions, Italy

P-598 Growth Parameters and Prevalence of Obesity in PKU Patients and Peers: Is This the Right Comparison?
Albina Tummolo, Department of Metabolic Diseases and Clinical Genetics, Giovanni XXIII Children Hospital, Italy

P-599 Phase 2 Efficacy of JNT-517 in Reducing Plasma Phenylalanine (Phe) Concentration in Adults with Phenylketonuria (PKU): Subgroup Analyses by Baseline Characteristics
Nicola Longo, University of California Los Angeles, USA

P-600 Comorbid Conditions in Individuals with Phenylketonuria (PKU): A United States Electronic Health Records (US EHR) and Medical Notes Study
Nicola Longo, University of California Los Angeles, USA

P-601 Bone Helth in Adults with PKU - an audit of the Western Australian Cohort
Catherine Manolikos, Royal Perth Hospital, Australia

P-602 The impact of a lifelong dietary treatment on eating behavior and the social lives of adults with phenylketonuria
Sietske Haitjema, Division of Metabolic Diseases, Beatrix Children's Hospital, University Medical Center Groningen, University of Groningen, Netherlands

P-603 Factors determining the loss to follow-up in PKU patients
Arthur Minas Alberti, Medical School, Federal University of Health Sciences of Porto Alegre, Brazil

P-604 Early Motor Development of infants with (maternal) Phenylketonuria in relation to Phenylalanine levels: An Observational Pilot Study
Sidra Jendo, University Medical Centre in Groningen - Beatrix children's hospital, Netherlands

P-605 Management of hyperemesis and elevated phenylalanine levels using home-based nasogastric feeding in PKU pregnancy.
Kate Lefebure, Metabolic Diseases Unit, The Royal Melbourne Hospital, Australia

P-606 Genomic profiling and implications for genotype-based treatment of 131 patients with phenylketonuria: characterization of novel p.Pro416Leu PAH variant
Kristel Klaassen, Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Serbia

P-607 Sharing anonymous genotypes: How can my patient cohort be made visible?
Polina Gundorova, University Medical Center Hamburg-Eppendorf, Germany / German Center for Child and Adolescent Health (DZKJ), partner site Hamburg, Germany

P-608 Phenylketonuria and long non-coding RNA HULC as a phenotype modifier
Juliana Cristine Fontana, Postgraduate Program in Genetics and Molecular Biology, Department of Genetics, Federal University of Rio Grande do Sul, Brazil / BRAIN Laboratory, Experimental Research Center, Hospital de Clínicas de Porto Alegre, Brazil

P-609 Relating parental personality and stress with eating styles and metabolic control in early treated PKU patients
Agnese De Giorgi, Department of Human Neuroscience, Sapienza University of Rome, Italy

P-610 Stakeholder Perspectives: Identifying the Barriers, Facilitators, and Solutions to People with Phenylketonuria (PKU) to Performing Physical Activity and Exercise
Annabelle Grace Skidmore, Birmingham City University, UK

P-611 Evaluating the Quality and Reliability of You Tube Videos about Phenylketonuria
Bahar Kulu, Department of Inherited Metabolic Diseases, University of Health Sciences, Tepecik Education and Research Hospital, Turkey

P-612 Impact of protein substitutes used in the treatment of Phenylketonuria on gut nutrient absorption and bacteria growth - an in vitro study
Catarina Rodrigues, Nutrition & Metabolism, CHRC, NOVA Medical School, Faculdade de Ciências Médicas, NMS, FCM, Universidade NOVA de Lisboa, Portugal

P-613 Impact of Dietary Therapy on Gut Microbiome Composition and Immunological Profiles in Children with Phenylketonuria
Natsuki Ohmi-Shimizu, Department of Pediatrics and Child Health, Nihon University School of Medicine, Japan / Department of Pediatrics, Tokyo Metropolitan Hiroo Hospital, Japan

P-614 Characteristics of Dopamine drug control in PTPS deficiency
Yasutsugu Chinen, Department of Child Health and Welfare, Graduate School of Medicine, University of the Ryukyus, Japan

P-615 Clinical and molecular spectrum of 6-pyruvoyl-tetrahydropterin synthase deficiency
Rai-Hseng Hsu, Department of Medical Genetics, National Taiwan University Hospital, Taiwan / Department of Pediatrics, National Taiwan University Hospital, Taiwan / Department of Pediatrics, National Taiwan University College of Medicine, Taiwan

24. Translational Research/New Diseases

P-616 CRISPR/Cas versus Antisense Strategies for Correcting Aberrant Pseudoexon Inclusion in Inherited Metabolic Disorders
Mar Alvarez, Centro de Biologia Molecular Severo Ochoa (UAM-CSIC), Spain

P-617 Beyond Heme Catabolism: Dual Roles of Heme Oxygenase-1 in Mitochondrial Function and Genomic Stability
Julien H. Park, University Hospital Muenster, Department of General Pediatrics, Germany

P-618 Mitochondrial Dysfunction and mito-DAMPS Drive Liver Fibrosis in Alpha-1 Antitrypsin Deficiency
Rosa Ferriero, Telethon Institute of Genetics and Medicine, Italy

P-619 Favoring speed over precision: protein mistranslation during high translational demand
Eveline Frances Ilcken, Department of Metabolic Diseases, Wilhelmina Children's Hospital, University Medical Center Utrecht, Netherlands

P-620 RARE fNIRS: A Novel Approach to Identifying Brain Biomarkers in Neurometabolic Disorders
Andrea Lynne Gropman, Center for Experimental Neurotherapeutics, St Jude Children's Research Hospital, USA

P-621 Liver pathology in Lars1-knockin zebrafish and mouse models of infantile liver failure syndrome type 1
Masanori Inoue, Department of Pediatrics, Faculty of Medicine, Oita University, Japan

P-622 Witnessing the Genomic Revolution: A 15-Years Laboratory Experience in Diagnosing Rare Diseases in India
Mehul Mistri, Neuberg Center For Genomic Medicine, India / Sandip and Bhavini Research Institute (SBRI), India / Foundation For Research In Genetics and Endocrinology, Institute of Human Genetics, India

P-623 Single-Dose Malonate at Reperfusion Prevents Post-Infarction Heart Failure via Mitochondrial ROS Inhibition
Jiro Abe, National Hospital Organization Hokkaido Medical Center, Japan / MRC Mitochondria Biology Unit and Department of Medicine, University of Cambridge, UK / Hokkaido University Hospital, Japan

P-624 Leveraging Education in Inborn Errors of Metabolism to Support the Metabolic Workforce and Patient Care
Debra Sue Regier, Children's National Hospital, USA

P-625 Roadmap to Base and Prime Editing Therapies for Rare Metabolic Diseases: Regulatory, Ethical, and Costs Perspectives
Stijn Paul Eric van Breda Vriesman, Division of Metabolic Diseases, Wilhelmina Children's Hospital, University Medical Center Utrecht, Netherlands / Regenerative Medicine Center Utrecht, Netherlands

25. Urea Cycle Disorders

P-626 Systemic Biomarkers of Brain Injury Due to Hyperammonemia in Neonatal N-acetylglutamate Synthase Knockout Mice
Ljubica Caldovic, Center for Genetic Medicine Research, Childrens National Hospital, USA

P-627 Nasal Nitric Oxide Levels are Reduced in Patients with Argininosuccinate Lyase and Argininosuccinate Synthase Deficiency.
Mehdi Yeganeh, Division of Biochemical Genetics, Department of Pediatrics, BC Children's Hospital, University of British Columbia, Canada

P-628 Modification of the variant classification rules for ornithine transcarbamylase through pilot curation
Kara Simpson, Children's National Hospital, USA

P-629 A Four-year Prospective Pilot Study of Newborn Screening for Late-onset Proximal Urea Cycle Disorders in Hyogo Prefecture in Japan
Tomoko Lee, Department of Pediatrics, Hyogo Medical University, Japan

P-630 Liver ultrasound findings in Urea Cycle Disorders patients: a French-Brazilian study
Francois Maillot, Department of internal medicine, university hospital of Tours, France

P-631 A Novel LC-MS/MS Method to Differentiate Proximal Urea Cycle Disorders by Quantifying Serum N-Acetylglutamic Acid and Pyrimidine Metabolites
Yuta Sudo, Department of Pediatrics, Fujita Health University School of Medicine, Japan

P-632 Plasma Glutamine and Ammonia Dynamics as Predictors of Hyperammonemic Crisis in Urea Cycle Disorders: A Retrospective Study Stratified by Onset Type
Yasuaki Yasuda, Department of Pediatrics, Fujita Health University School of Medicine, Japan

P-633 The standardized in-hospital protein loading test: a safe and effective tool for the evaluation of urea cycle disorders in adults
Margreet A. Wagenmakers, Center for Lysosomal and Metabolic Diseases, Department of Internal Medicine, Erasmus MC, Erasmus University Medical Center, Netherlands

P-634 Relevance of extracellular nutrients for ammonia detoxification in urea cycle disorders
Nathan Breuillard, University Children's Hospital Zurich, Switzerland

P-635 Comparative Efficacy, Safety, and Pharmacokinetics of Glycerol Phenylbutyrate Versus Sodium Phenylbutyrate in Japanese Patients with Urea Cycle Disorders: Results from a Phase 3 Study
Mahoko Furujo, Okayama Medical Center, Japan

P-636 Withdrawal

P-637 Birth prevalence of Urea Cycle Disorders in Argentina: A Nationwide Multicenter Study
Betiana Mabel Perez, Hospital Italiano De Buenos Aires, Argentina

P-638 Long-term Outcomes of Adult Urea Cycle Disorder (UCD) Patients in Japan: A Nationwide Study
Jun Kido, Department of Pediatrics, Faculty of Life Sciences, Kumamoto University, Japan

P-639 Symptom Profiles at Diagnosis and During Follow-up of Urea Cycle Disorders in a cohort of 73 patients from Argentina
Soledad Kleppe, Hospital Italiano de Buenos Aires, Argentina

P-640 The diagnosis, treatment, and outcome of 72 patients with ornithine carbamoyltransferase deficiency
Xue Ma, Children's Medical Center, Peking University First Hospital, China

P-641 Ornithine transcarbamylase deficiency in Vietnam during 20 years: Phenotype, genotype and outcome
Khanh Ngoc Nguyen, Vietnam National Children's Hospital, Vietnam

P-642 Under-recognized Neurological and Cognitive Symptoms of Late-onset Ornithine Transcarbamylase Deficiency (OTCD): A Narrative Review
Laura Konczal, University Hospitals Cleveland Medical Center, USA

P-643 Clinical, biochemical and genetic characteristics of Chinese patients with argininosuccinate lyase deficiency
kaichuang zhang, Xinhua Hospital Affiliated to Shanghai Jiao Tong University School of Medicine, China

P-644 A Patient with Hyperammonemia Due to a Novel Mutation in the CA5A Gene
Mustafa Kilic, University of Health Sciences, Ankara Etlik City Hospital, Department of Pediatrics, Metabolism Unit, Turkey

P-645 A boy with lysinuric protein intolerance and systemic lupus erythematosus
Jingtao Zhang, Children's Medical Center, Peking University First Hospital, China

P-646 Three Late-Diagnosed Cases of Citrullinemia Type 1
Sedef Alpdogan, Ege University Faculty of Medicine, Department of Pediatric Metabolic Diseases and Nutrition, Turkey

P-647 Marathon training in Ornithine Transcarbamylase deficiency- how far is too far?
Simon Tapley, UHBW, UK

P-648 A boy diagnosed with infantile-onset ornithine transcarbamylase deficiency presented with an acute onset hemiconvulsion-hemiplegia-epilepsy syndrome
Yoichiro Kai, Department of Pediatrics, Oita University Faculty of Medicine, Japan

P-649 Carrier Frequency and Prevalence of Citrin Deficiency in East Asians and Koreans Based on Comprehensive Analysis of SLC25A13 Pathogenic Variants
Hyung-Doo Park, Samsung Medical Center, Republic of Korea

P-650 Triheptanoin for Citrin Deficiency: A Pilot Clinical Study Evaluating Safety and Exploratory Efficacy
Kimihiko Oishi, Department of Pediatrics, The Jikei University School of Medicine, Japan

P-651 In Vivo Assessment of Acetaminophen Hepatotoxicity in Citrin Deficiency Mouse Model
Hikaru Nishida, Department of Pediatrics, The Jikei University School of Medicine, Japan

P-652 Pregnancy-Related Risks and Complications in Women with Citrin Deficiency: Low Risk of Perinatal Hyperammonemia and Caution with OGTT
Hikaru Nishida, Department of Pediatrics, The Jikei University School of Medicine, Japan

26. Late-Breaking Research

P-653 A human induced pluripotent stem cell derived neural progenitor gene therapy corrects a neuropathic lysosomal disease
Brian Bigger, University of Edinburgh, UK

P-654 TFE3 ASSOCIATED NEURODEVELOPMENTAL DISORDER A NEWLY RECOGNIZED LYSOSOMAL CONDITION
Eyby Leon, Rare Disease Institute Children's National Hospital, USA

P-655 Engineered Exosomes as mRNA Delivery Vehicles for Targeted Therapy in Fabry Disease
Cheng-Che Lin, Genetic Consultant Center Rare Disease Medical Research Center, Taipei Veterans General Hospital, Taiwan / Department of Pediatrics, Taipei Veterans General Hospital, Taiwan

P-656 Functional and Therapeutic Studies in Pyridoxine-Dependent Epilepsy Due to Antiquitin Deficiency
Hilal Al-Shekaili, Department of Biology, College of Science, Sultan Qaboos University, Oman / Changing Rare Disorders of Lysine Metabolism (CHARLIE) international consortium

P-657 Robust Statistical Framework for Targeted Metabolomics in Rare Disease Diagnostics
Martin Poms, University Children's Hospital Zurich, Division of Clinical Chemistry and Biochemistry, Switzerland

P-658 Point-of-Care Testing (POCT) for Blood Phenylalanine Monitoring: Preliminary Results
Alex Pinto, Birmingham Children's Hospital, UK

P-659 ACMSD Deficiency as a Novel Cause of Early-Onset Neurodegeneration via Quinolinic Acid Accumulation
Nika Schuermans, Center for Medical Genetics Ghent, Ghent University Hospital, Belgium / Department of Biomolecular Medicine, Ghent University Hospital, Ghent, Belgium

P-660 Cipaglucosidase alfa and alglucosidase alfa enzymes have similar stability at neutral pH and can be stabilized with miglustat
Filip Cosmanescu, Amicus Therapeutics, Inc., USA

P-661 Major Clinical Events and Healthcare Resource Use Among Individuals With Long-chain Fatty Acid Oxidation Disorders (LC-FAOD) Pre- and Post-triheptanoin Initiation: A Retrospective Claims Analysis
Erru Christy Yang, Ultragenyx Pharmaceutical Inc., USA

P-662 Isaralgagene Civaparvovec (ST-920) Shows Positive Mean Annualized eGFR Slope in Adults with Fabry Disease: Updated Results from the Registrational Phase 1/2 STAAR Gene Therapy Study
Robert J Hopkin, Cincinnati Children's Hospital Medical Center, USA and University of Cincinnati College of Medicine, USA / Emory University School of Medicine, USA

P-663 Clinical, Molecular Characteristics and Prognosis of 50 Children with Mitochondrial Leukoencephalopathy in China
Minhan Song, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, China

P-664 Expanding the phenotypic spectrum of Combined Saposin Deficiency with homozygous PSAP p.Lys227del variant presenting with Dystonia-like features
Priyanshu Mathur, Department of Medical Genetics, SMS Medical College, Jaipur, India

P-665 Resilience in patients with Fabry Disease and its association with disease course, psychosocial factors and quality of life: a multicentre cross-sectional study
Albina Nowak, MD, Switzerland

P-666 Novel brain-targeting gene editing strategies for treatment of neurometabolic disorders
Eveline Frances Ilcken, Metabolic Diseases, Division Pediatrics, Wilhemina Children's Hospital, University Medical Center Utrecht, Netherlands

P-667 Withdrawal

P-668 Longitudinal Changes in Left Ventricular Mass Index in Male Fabry Disease Patients Before and After Enzyme Replacement Therapy
Hsing-Yuan Li, Taipei Veterans General Hospital, Taiwan

ePosters

1. AI Based Research

EP-01 Novel MTR gene variations in a patient with cblG disorder: A case report and the treatment
Weimeng Ma, Children's Medical Center of Peking University First Hospital, China

EP-02 Withdrawal

EP-03 Deep Learning-Based Kinetic Parameter Prediction: Novel methods, Comparative Evaluation and Increased Accessibility
Saleh Alwer, Digital Metabolic Twin Centre, University of Galway, Ireland

EP-04 Human synonymous codon variation is constrained within multivalent coding regions
Rhys Dore, UK Dementia Research Institute at King's College London, UK

2. Amino Acid Disorders

EP-05 A Case of Maple Syrup Urine Disease with Infantile Epileptic Spasms Syndrome: Diagnostic and Therapeutic Challenges in a Resource-Limited Setting
Kenji Iwai, Pediatric department, Sunrise Japan Hospital Phnom Penh, Cambodia

EP-06 Withdrawal

EP-07 Accessing Maple Syrup Urine Disease genetic variants in Brazil: a literature review
Rafael Hencke Tresbach, BRAIN Laboratory (Basic Research and Advanced Investigations in Neurosciences), CPE, Hospital de Clínicas de Porto Alegre, Brazil

EP-08 Two siblings with branched chain ketoacid dehydrogenase Kinase deficiency and the outcome after nutritional treatment
Huiting Zhang, Peking University First Hospital, China

EP-09 From Symptoms to Solutions: A case study on Alkaptonuria and its management
Stephanie Brown, Queensland Lifespan Metabolic Service, Mater Hospital, Australia / University of Queensland, Australia

EP-10 Glutaminase Hyperactivity Associated with Interferonopathy
Laura Andrea Castaneda-Correa, Division of Genetics, Genomics and Metabolism, Department of Pediatrics, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

EP-11 METHYLMALONIC ACIDEMIA AND DESQUAMATION: A SURPRISING RESPONSE TO ZINC THERAPY
Hadeel a alrabee, Prince Sultan Military Hospital , Saudi Arabia

EP-12 Diagnostic Yield of Metabolic Tests Covered by Thailand's Universal Health Coverage for Rare Diseases
Nithiwat Vatanavicharn, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj Hospital, Mahidol University, Thailand

EP-13 Atypical nonketotic hyperglycinemia with long-term epileptic seizure remission: A case with a novel AMT gene variant
Tomoyo Itonaga, Department of Pediatrics, Oita University Faculty of Medicine, Japan

EP-14 5-Oxoprolinase Deficiency with Coexisting COL4A1-Related Cerebral Vasculopathy in a Child with Neurodevelopmental Delay
Merve Yoldas Celik, Adana City Training and Research Hospital, Department of Pediatric Metabolism, Turkey

EP-15 Challenges in Diagnosing and Managing Maple Syrup Urine Disease in Limited Facilities
Rini Andriani, Department of Child Health, Kharitas Bhakti Hospital, Indonesia / Department of Child Health, Antonius General Hospital, Indonesia / Department of Child Health, Tanjungpura University, Indonesia

EP-16 Lysinuric protein intolerance presenting as pancytopenia and splenomegaly mimicking acute leukaemia: a case report
Hashan Kavinga Pathiraja, Department of Paediatrics, Faculty of Medicine, University of Kelaniya, Sri Lanka

EP-17 A CASE OF GLUTATHIONE SYNTHETASE DEFICIENCY PRESENTING WITH HEMOLYTIC ANEMIA AND METABOLIC ACIDOSIS DUE TO MUTATIONS IN THE GSS GENE
Phuong Thi Hong Chu, Department of Neonatology 2 - Metabolism & Genetics, Children's Hospital 1, Vietnam

EP-18 Alkaptonuria diagnosed in a 4-year-old boy in Indonesia: A rare case of metabolic disorder
Mislina Munir, Pediatric Nutrition and Metabolic Disease Division, Department of Child Health, Faculty of Medicine, Universitas Indonesia, Cipto Mangunkusumo General Hospital, Indonesia

EP-19 Methylmalonic aciduria and pyridoxine responsive seizure in a child: A management challenge
Rihab Mohammed Salih, Prince Sultan military medical city, Saudi Arabia

EP-20 Estimation of cerebral amino acid influx in patients with propionic acidemia
Sinziana Stanescu, CSUR Enfermedades Metabolicas, Hospital Ramon y Cajal, , Spain

3. Carbohydrate Disorders

EP-21 Expanding the Genotype-Phenotype Correlation in Pyruvate Carboxylase Deficiency: Homozygous p.Arg631Gln Causes a Mild Intermittent Phenotype
Elis Vanessa de Lima Silva, Hospital Infantil Dr Juvencio Mattos, Brazil / Hospital Universitário da Universidade Federal do Maranhão, Brazil

EP-22 Severe lactic acidosis induced by COVID-19 infection in GSD type Ia
Yusuke Noda, Department of Pediatrics, Kumamoto University Hospital, Japan

EP-23 Suspecting Hereditary Fructose Intolerance (HFI): can we rely on urinary fructose for the diagnosis or should other diagnostic handles guide us?
Agata Maria Capodiferro, Foundation IRCCS Ca' Granda Ospedale Maggiore Policlinico, Clinical Metabolic Center, Italy / University of Milan, Italy

EP-24 Cerebral venous thrombosis as the first presentation in a neonate with GSD type1a
Liumei Liu, Department of Pediatrics and Child Health, Canada

EP-25 Clinical, paraclinical data and genetic analysis of 2 cases affected by Fanconi-Bickel syndrome in Iran
Talieh Zaman, Clinical & Research unit of Iranian National Society of SSIEM, Canada

EP-26 A Case of Continuous Glucose Monitoring in Children with Glutamate Dehydrogenase Hyperinsulinism
Hae Soon Kim, Ewha Womans Uinversity Seoul Hospital , Republic of Korea / Ewha Womans Uinversity Mokdong Hospital , Republic of Korea

EP-27 Clinical variability of persistent congenital hyperinsulinism experienced in a single tertiary care center
Jeesuk Yu, Department of Pediatrics, Dankook University Hospital, Republic of Korea

EP-28 A locus-specific database of G6PC1 gene variants associated with Glycogen Storage Disease type Ia
Kimimasa Tobita, Ultragenyx Japan K. K., Japan

EP-29 Generalized Galactose Epimerase (GALE) Deficiency in a dysmorphic infant with failure to thrive and hypotonia
Christina L. Grant, Children's National Hospital, Genetics and Metabolism, USA

EP-30 Glycogen storage disease type IX in Vietnam National Children's Hospital
Ngoc Thi Bich Can, Vietnam National Children's Hospital, Vietnam

EP-31 Co existence of Gaucher type 1 and GSD type VI in a patient
Rihab Mohammed Salih, Prince Sultan military medical city, Saudi Arabia

EP-32 Glycerol Kinase Deficiency Presenting as Pseudohypertriglyceridemia: A Case Series
Engin Kose, Department of Pediatrics, Ankara University Faculty of Medicine, Turkey

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Agnieszka Konopka, Polish Mothers Memorial Hospital, Research Institute, Regional Centre for Rare Diseases, Poland

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Greg Woodhead, Royal Children's Hospital, Australia

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Yuan Ding, Beijing Children's Hospital, China

EP-36 Methylmalonic Aciduria is Found the Cause of Persistent Proteinuria in a Girl After Ten Years diagnosis of Atypical Nephropathy. Case report and Literature Review
Min Yang, Shengjing Hospital of China Medical University, China

EP-37 Epidemiological characteristics of diabetes in children before and after the COVID-19 pandemic in Henan,China, during 2017 to 2024
Yifan Lin, Henan Children's Hospital, China

EP-38 A Novel Mutation in SALL1 Gene Causes Townes Brocks Syndrome: A Case Report
zhang jing, Chengdu Women's and Children's Center Hospital. School of Medicine, University of Electronic Science and Technology of China, China

EP-39 Genetic Insights into Infantile Interstitial Lung Disease: A Case of SMDP2 with SFTPC Mutation and Complex Clinical Features
Prajnya Paramitha Narendraswari, Harapan Kita Women and Children Hospital, Indonesia

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shuxian Yuan, Children's Hospital Affiliated to Zhengzhou University, Henan Provincial Children's Hospital, China

EP-41 Clinical Characteristics and Follow-up Observations of 12 Cases of Graves' Disease in Chinese Infants
Jia jia Wang, Children's Hospital Affiliated to Zhenzhou University , China

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Zi ying Wu, Department of Genetics and Endocrinology, Women and Children's Medical Center, Guangzhou Medical University, China

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Yan-fei LUO Yan-fei LUO, First Affiliated Hospital of Xinjiang Medical University, Department of Pediatrics, China

EP-44 Phenotypic and genotypic characteristics of neurofibromatosis type 1 in children: A single-center cohort study
zhiying li, Shanghai Children's Medical Center, Shanghai Jiaotong University School of Medicine, China

EP-45 Clinical follow up of a patient with Smith Lemli Opitz treated with Cholic acid
Alvaro Serrano, ETSU Health Quillen College of Medicine, USA

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Hannah Roberts, Make-A-Wish International, Netherlands

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Novitria Dwinanda, Harapan Kita Women and Children Hospital, Indonesia

EP-48 A Case Report of Neonatal Adrenal Crisis
Fang Sun, China Japan Friendship Hospital, China

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Bei bei Zhang, Beijing Children's Hospital, China

EP-50 Clinical and genetic analysis and literature review of 23 cases of hypogonadotropic hypogonadism in men due to FGFR1 variants
Zixia Zhang, Affiliated Children's Hospital of Zhengzhou University, China

EP-51 Type 1 Dent Disease with Bartter-like Syndrome in a Child: Case Report and Literature Review
Maimaiti M, The First Affiliated Hospital of Xinjiang Medical University, China

EP-52 Insight into Gut Microbiota of Normal Body Mass Index Children with Precocious Puberty
Yifan Lin, Henan Children's Hospital, China

EP-53 Assessment of cross-linking of donor corneas following therapeutic keratoplasty in cases of Infectious Keratitis
Sushma Nandyala, All India Institute of Medical Sciences, India

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Aree Rattanathongkom, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Siriraj hospital, Mahidol University, Thailand / Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine, Khon Kaen University, Thailand

EP-55 A boy with treatable autism spectrum disorder due to carnitine deficiency caused by TMLHE gene mutation
Jingtao Zhang, Children's Medical Center, Peking University First Hospital, China

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Thomas Lundqvist, Karolinska University Hospital, Sweden

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Phuc Nguyen, Children's Hospital 1, Vietnam

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Farid Zare, School of Medicine, University of Galway, Ireland

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Suzan Hilhorst, BNNVARA, Netherlands

EP-60 Trends of Sex Hormone-Binding Globulin in Obese Children during Puberty and Its Association with Clinical Outcomes
Ziyu Zhao, Children's Hospital Affiliated to Zhengzhou University, China

EP-61 Characteristics of metabolic complications related to childhood obesity and construction of prediction models:A single-center retrospective study
Yi Wei, Department of Endocrinology and Inherited Metabolic, Children's Hospital Affiliated to Zhengzhou University, China

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Chloe Miller, Evelina London Childrens Hospital, UK

EP-63 RELEVANT ASPECTS TO CONSIDER IN ADDITION TO THE NUTRITIONAL MEDICAL TREATMENT IN CHILDREN WITH MAPLE SYRUP URINE DISEASE FOR A GOOD METABOLIC CONTROL
Gabriela Noemi Nicola Orejas, Son Espases University Hospital, Endocrinology Department, Spain / Son Espases University Hospital, Endocrinology Department., Spain

EP-64 Dietary Micronutrient Intake and Contribution of Metabolic Formula Among Patients with Aminoacidopathies(AAs) and Organic Acidurias(OADs) in Malaysia
Maslina Mohamad, Dietetics and Food Services Department,National Cancer Institute, Malaysia / Dietetics and Food Services Department,Hospital Kuala Lumpur(HKL), Malaysia

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Gabriele Skacel, Division of Pediatric Pulmonology, Allergology and Endocrinology, Department of Pediatrics and Adolescent Medicine, Medical University of Vienna, Austria

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Joana Maria Palhotas, 1 Reference Center for Inherited Metabolic Diseases, Dietetic and Nutrition Department, Santa Maria´s Health Local Unit, Portugal

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Dayaana Vasileva, Yakut Scientific Center, Siberian Branch, Russian Academy of Sciences, Russia / North-Eastern Federal University, Russia

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Camille Newby, Bristol Royal Hospital for Children , UK

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Clare Dale, Queen Elizabeth Hospital, UK

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Zaridah Zainuri, Dietetic and Food Service Department, Hospital Pulau Pinang, Malaysia

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Xue Ma, Children's Medical Center, Peking University First Hospital, China

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Mireguli Maimaiti, The First Affiliated Hospital of Xinjiang Medical University, China

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Fahad J Alharbi, Prince Sultan Military Medical City, Saudi Arabia

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Miki Matsui, Department of Pediatrics, Hyogo Medical University, Japan

EP-75 A CASE OF SEVERE ACIDOSIS IN A 15-MONTH-OLD CHILD: SUCCINYL-COA:3-KETOACID-COA TRANSFERASE DEFICIENCY DUE TO OXCT1 GENE MUTATIONS
Phuong Thi Hong Chu, Department of Neonatology 2 - Metabolism & Genetics, Children's Hospital 1, Vietnam

EP-76 Placental histopathological findings facilitate prognostication and help guide management of perinatal, lethal LCHADD.
Beena Devanapalli, NSW Biochemical Genetics Service, Australia

EP-77 Two sisters with a mild phenotype of Carnitine palmitoyltransferase 1A (CPT1A) deficiency
elham saeed bagrayn, Prince Sultan Military Medical City.Riyadh,Saudi Arabia , Saudi Arabia

EP-78 3-hydroxybutyrate supplementation in decompensation with prominent liver failure in an adult with multiple acyl-Coa dehydrogenase deficiency
Dinusha Pandithan, Department of Genetic Medicine, Westmead Hospital, Australia

EP-79 Disorder or distraction: Considering the significance of an ACADSB gene variant in a young person with developmental delay
James Nurse, Southampton General Hospital, UK

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Kamel Monastiri, Department of Intensive Care and Neonatal Medicine, Teaching Hospital of Monastir, Tunisia

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Aparna Ananthanarayan, Department of Clinical Chemistry, Inselspital, University Hospital Bern, University of Bern , Switzerland

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Mustafa Kilic, University of Health Sciences, Ankara Etlik City Hospital, Department of Pediatrics, Metabolism Unit, Turkey

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Walaa Ali Elkhalil, pediatric department /Prince Sultan Military Hospital, Saudi Arabia

EP-84 Persistent beetroot colored urine in a barely three-year-old child: Early-Onset Porphyria Cutanea Tarda in a Pediatric Patient
Eliane Sardh, Centre for Inherited Metabolic Diseases, Porphyria Centre Sweden, Karolinska University Hospital, Department of Molecular Medicine and Surgery, Karolinska Institutet, Sweden

EP-85 Detection of hyperuricemia in children with simple obesity and its factors affecting the detection of hyperuricemia in children with simple obesity
Yixuan Zhao, Henan Children's Hospital, China

EP-86 Clinical and genetic analysis of MPS IVA caused by GALNS mutation in a Chinese boy and literature review
Jianmei Yang, Department of Paediatric Endocrinology, Shandong Provincial Hospital affiliated to Shandong First Medical University, China

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Wenjing Li, Capital Medical University, Beijing Children's Hospital, China

EP-88 A Rare Cause of Recurrent Apnea in Infancy: MTHFR Deficiency
Elham Saeed Bagaryn, Genetics and Metabolic Medicine Division,Department of Pediatrics,Prince Sultan Military Medical City, Saudi Arabia

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Saja Baheer Abdulwahhab, Department of Genetics, Sidra Medicine, Qatar

EP-90 Pulmonary Emphysema and Fatal Tension Pneumothorax in Menkes Disease
Yasuko Mikami Saito, Tohoku University Hospital, Japan

EP-91 The metabolic landscape of tetrahydrobiopterin metabolism disorders in the Republic of Ireland
Arie Fisher, National Centre for Inherited Metabolic Disorders,Children's Health Ireland at Temple Street, Ireland

EP-92 Calcium channelopathy due to CACNA1E mutation resulting in Developmental Epileptic Encephalopathy with response to Acetazolamide - A case report
Hashan Kavinga Pathiraja, Department of Paediatrics, Faculty of Medicine, University of Kelaniya, Sri Lanka

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Katarzyna Olszewska-Durkacz, Department of Pediatrics, Nutrition and Metabolic Disorders, the Children's Memorial Health Institute, Poland

EP-94 Treatment of metabolic decompensation with vitamin B6 and intravenous thiamine of a patient with biotin-and thiamine-responsive basal ganglia disease
Dorota Wesol-Kucharska, Department of Pediatrics, Nutrition and Metabolic Disorders, Children's Memorial Health Institute, Poland

EP-95 A Preliminary Study on the Levels of Serum Melatonin and 25-Hydroxyvitamin D in Girls with Precocious Puberty
Yi Wei, Department of Endocrinology and Inherited Metabolic, Children's Hospital Affiliated to Zhengzhou University, China

EP-96 Wilson disease in a toddler presented with mild elevation of liver enzymes
Manal Abdelrahim Wadatalla Abdelrahim, prince sultan military medical city, Saudi Arabia

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Kamel Monastiri, Teaching Hospital of Monastir, Tunisia / Department of Medical Genetics, Teaching Hospital of Monastir, Tunisia

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Goksu Demirbas, Basaksehir Cam and Sakura City Hospital, Turkey

EP-99 Growth profile and outcome of X-linked hypophosphatemia in Chinese population
Chunhua Zeng, Guangzhou Women and Children's Medical Center, Guangzhou Medical University, China

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Chunhua Zeng, Guangzhou Women and Children's Medical Center, Guangzhou Medical University, China

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Ozge Kamer Karalar Pekuz, Division of Pediatric Metabolic Diseases, Department of Pediatrics, Faculty of Medicine Dokuz Eylul University, Turkey

EP-102 Long-term galactose supplementation in a patient with SLC35A2-CDG: a 7-year clinical study
Yupeng Liu, Children's Hospital of Philadelphia, USA

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Ezgi Burgac, Adana City Training and Research Hospital, Turkey

EP-104 A case report: Maternal uniparental disomy of chromosome 17 in a patient with Pompe disease?
Trang Thuy Nguyen, Department of Human Genetic, Vietnam

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Nadia Moosa Alhashmi, Royal hospital, Oman

EP-106 THE FIRST CASE OF CONGENITAL DISORDERS OF GLYCOSYLATION TYPE 2G (CDG 2G) IN FATMAWATI HOSPITAL: DIFFICULTY IN ESTABLISHING DIAGNOSIS
Lanny Christine Gultom, Fatmawati Hospital, Indonesia

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Rose Marino, Glycomine Inc, USA

10. Inborn Errors of Metabolism in Adults

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Mathanky Rajalingam, Department of Chemical Pathology, Teaching Hospital Jaffna, Sri Lanka

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Pelin Teke Kisa, Dokuz Eylul University, Turkey

EP-110 Expanding Knowledge of Sexual and Reproductive Health in Fatty Acid Oxidation Disorders Using Mixed Methodology
Jessica I Gold, Northwell Health, USA / Cohen Children's Medical Center, USA / Feinstein Research Institute, USA

EP-111 Adult-Onset Pompe Disease Presenting with Low Back Pain
Bengu Arslan, Department of Pediatric Metabolism, Sakarya University Training and Research Hospital, Turkey

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Rebekah Barrick, Division of Metabolic Disorders, Children's Hospital of Orange County, USA

EP-113 Transition experience in rare metabolic diseases in Poland
Ewa Beata Ehmke vel Emczynska-Seliga, Department of Paediatrics, Nutrition and Metabolic Diseases (Children's Memorial Health Institute), Poland

EP-114 Bilateral Renal Calcification and End-Stage Renal Disease in a Young Woman with Primary Hyperoxaluria Type 1: A Case Report
Cynthia Rucinski, Colsanitas, Colombia

EP-115 Pseudo-Bartter Syndrome as an Initial Manifestation of Cystic Fibrosis: A Case Report
Feng-Jung Yang, Department of Medical Genetics, National Taiwan University Hospital, Taiwan

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Tai Chaiamarit, Department of Physiology, Faculty of Science, Mahidol University, Thailand

EP-117 Is there any change in clinical practice after the approval of gene therapy?
Cecilia Marinova, Medasol sro, Czech Republic

EP-118 Evaluation of AAV vectors with tissue-specific or ubiquitous promoters in a mouse model of mucopolysaccharidosis type IVA
Shunji Tomatsu, Nemours Children's Health, USA

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Soyoung Kim, Asan Institute for Life Sciences, Asan Medical Center, University of Ulsan College of Medicine, Republic of Korea

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Rayyan Albarakati, BC Children's Hospital, Canada

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Yoshikatsu Eto, Advanced Clinical Research Center, Southern Tohoku Institute for Neuroscience, Japan

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Boudewijn A.W. van Binsbergen, University Medical Center Utrecht, Department of Plastic, Reconstructive, and Hand Surgery, Netherlands / Wilhelmina Children's Hospital Utrecht, Department of Plastic, Reconstructive, and Hand Surgery, Netherlands / Wilhelmina Children's Hospital Utrecht, Department of Metabolic Diseases, Netherlands

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Yuko Fujiwara, Teikyo University , Japan

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Bohdan Mahlovanyi, Insitute of Physics, University of Rzeszow, Poland

EP-126 PROPEL Japan subpopulation: efficacy and safety of cipaglucosidase alfa plus miglustat versus alglucosidase alfa in patients with late-onset Pompe disease
Hiroshi Kobayashi, Division of Gene Therapy, Research Center for Medical Sciences, The Jikei University School of Medicine, Japan

EP-127 Diagnostic Journey For Patients With Infantile Krabbe Disease And Late-Onset Krabbe Disease
Merve Bilen, Depertment of Pediatric Metabolism and Nutrition, Dokuz Eylul University Faculty of Medicine, Turkey

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Mariia Haidei, National Specialized Children's Hospital Ohmatdyt, Ukraine

EP-129 Navigating the real-world challenges of alpha-mannosidosis patients and caregivers: Understanding their journeys during, and after, diagnosis
Nato V. Vashakmadze, Pirogov Russian National Research Medical University, Federation Research Institute of Pediatrics and Children's Health in Petrovsky National Research Centre of Surgery, Russia

EP-130 Discordance of TPP1 Enzyme Activity and Genetic Findings in Two Siblings with Neuronal Ceroid Lipofuscinosis Type 2: A Case Report
Ronald Rompies, Pediatric Nutrition and Metabolic Division, Department of Child Health, Faculty of Medicine, Sam Ratulangi University, Kandou Hospital, Indonesia

EP-131 Respiratory manifestations in neuronopathic Gaucher disease
Solaf Mohamed Elsayed, Medical Genetics Department, Ain Shams University, Egypt

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Shungo Okamoto, Department of Pediatrics, Osaka Metropolitan University Graduate School of Medicine, Japan

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Thouraya Ben Younes, LR18SP04 and Department of Pediatric Neurology, National Institute Mongi Ben Hmida of Neurology of Tunis, Tunisia

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Merve Yoldas Celik, Adana City Training and Research Hospital, Department of Pediatric Metabolism, Turkey

EP-135 Avalglucosidase Alfa in Early-Onset Pompe disease:One-Year Safety and Efficacy Outcomes
Ana Clara Roa, Hospital Italiano de Buenos Aires, Argentina

EP-136 A confirmation of complex rearrangements of intron 3 and intron 7 of IDS-IDS2 in two Hunter syndrome cases in Indonesia
Bobby Pambudi, Department of Child Health, Kandou General Hospital-Faculty of Medicine, Sam Ratulangi University, Indonesia

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Wararat Chiangjong, Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University , Thailand

EP-138 In Silico Prediction of Pharmacological Chaperone Responsiveness in Fabry Disease Variants
Akif Altun, Department of Rare Diseases, Institute of Graduate Studies in Health Sciences, Istanbul University, Turkey

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Vincenza Gragnaniello, University Hospital of Padua, Italy

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Takanori Onuki, Department of Pediatrics, Niigata University Graduate School of Medical and Dental Sciences, Japan

EP-141 Breaking barriers in Farber disease: how Tocilizumab and HSCT altered the disease course
Chern Yan Tan, The Willink metabolic unit, UK

EP-142 Efficacy of Hydroxychloroquine in Managing Respiratory Complications in Niemann-Pick Disease Type C
Faizah Hashimy, Department of Pediatrics, Tawam Hospital, UAE / Department of Genetic & Genomics, College of Medicine and Health Sciences, United Arab Emirates University, UAE / AlJalila Children's hospital, UAE

EP-143 Comparative case series demonstrating disease modifying outcome with early ambroxol therapy in acute neuronopathic Gaucher disease
Katherine Li, Genetic Metabolic Disorders Service, The Children's Hospital at Westmead, Australia

EP-144 From Childhood to Adulthood: A Descriptive Analysis of Clinical Features in Patients with Acid Sphingomyelinase Deficiency (ASMD)
Maria Camprodon Gomez, Hospital Vall d'Hebron, Spain / Internal Medicine Service, Spain / Rare Diseases and Metabolic Unit, Spain

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John A. Bernat, University of Iowa Health Care, USA

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Myeongjin Kim, Samsung Medical Center, Republic of Korea

EP-147 Biochemical and enzymatic evaluation on the clinical efficacy of migalastat in Japanese patients with Fabry disease
Masahisa Kobayashi, Department of Pediatrics, The Jikei University School of Medicine, Japan

EP-148 Mortality and health inequalities in Late Onset Pompe Disease; a single tertiary centre experience
Andrew Oldham, Adult Inherited Metabolic Diseases, UK

EP-149 Global Insights into Parental Perception of Treatment Options for Mucopolysaccharidosis: Development of a Collaborative International Survey for Advancing Clinical Trials
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

EP-150 Should enzyme therapy be offered to mild cases of Nieman Pick type B
elham saeed bagrayn, Genetics and Metabolic Medicine Division,Department of Pediatrics, Prince Sultan Military Medical City, Saudi Arabia

EP-151 Dermatological Manifestations in Lysosomal Storage Diseases: Review and Genotype-Phenotype Correlation at the Adult National Centre for Inherited Metabolic Disorders, Mater Misericordiae University Hospital
Madalina Lefter, Adult National Centre for Inherited Metabolic Disorders, Mater Misericordiae University Hospital, Ireland

EP-152 Cardiovascular Structure and Function in MPS VII Subjects
Adriana Montano, Saint Louis University, USA

EP-153 Outcomes of 2 patients with neuronal ceroid lipofuscinosis type II (CLN2) on enzyme replacement therapy
Nadia Moosa Alhashmi, Royal Hospital, Oman

EP-154 Creation Manuals of Home Infusion Enzyme Replacement Therapy for Lysosomal Disease in Japan
Hiroyuki Yamakawa, Department of Cardiology, School of Medicine, Keio University, Japan / Center for Preventive Medicine, School of Medicine, Keio University, Japan

EP-155 Tailored desensitization and omalizumab-induced immunotolerance protocol in a child affected by chronic neurovisceral ASMD with previous olipudase alfa-related anaphylaxis
Laura Fiori, Department of Pediatrics, Vittore Buzzi Children's Hospital, Italy

EP-156 A case of infantile form of Tay-Sachs disease with recurrent acute pancreatitis in adolescence.
Keiko Ichimoto, Department of Metabolism, Chiba Children's Hospital, Japan

EP-157 Recognizing Social Stigma in Fabry Disease: First Step to Better Patient Outcomes
Yin-Hsiu Chien, National Taiwan University Hospital, Taiwan

EP-158 Cardiac characteristics of Fabry disease from baseline enrolment data in a nationwide prospective Japanese registry
Toru Kubo, Department of Cardiology and Geriatrics, Kochi Medical School, Kochi University, Japan

EP-159 Immunometabolic Background of COVID-19 Resistance in Gaucher Disease: Clinical Observations and Pathophysiological Hypotheses.
Nikola Maria Krol, Faculty of Medicine, Collegium Medicum, University of Rzeszow, Poland

EP-160 Lysosomal Storage Disease Diagnosed in Adults with Hypoxic Ischaemic Encephalopathy: Fucosidosis Case Report
Emine Didem Demirdoken, Department of Pediatric Metabolic Diseases, Dokuz Eylul University Faculty of Medicine, Turkey

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Wataru Oboshi, AnGes Clinical Research Laboratory, Japan / Department of Clinical Genomics, Saitama Medical University, Japan

EP-162 Evaluation of Ten Individuals with Non-Pathogenic GLA Variants
Funda Bostanci, Gazi University, Turkey

EP-163 Gangliosidosis: Clinical and paraclinical profile in a Pediatric Tunisian Series
Zouhour Miladi, Research unit LR18SP04 and Department of Child and Adolescent Neurology, National Institute Mongi Ben Hmida of Neurology of Tunis, Tunisia

EP-164 Thirty months of enzyme replacement therapy with Olipudase A in a paediatric patient with acid sphingomyelinase deficiency.Real world data from Cyprus.
Emilia Athanasiou, Genetics Clinic, Archbishop Makarios Hospital III, Cyprus

EP-165 Niemann - Pick disease type A/B : diagnosis from childhood to adulthood in a health system with limited resources - clinical geneticist's perspective
Vasilica Plaiasu, INSMC Alessandrescu-Rusescu, Regional Center of Medical Genetics, Romania

EP-166 A Novel Mutation in the CTNS Gene Causes Intermediate Cystinosis
Yukiko Shimizu, Department of Laboratory Animal Medicine, National Institute of Global Health and Medicine, Japan Institute for Health Security (JIHS), Japan / Department of Pediatrics and Adolescent Medicine, School of Medicine, Juntendo University, Japan

EP-167 Olipudase alfa enzyme replacement therapy. One year outcomes in an adult patient with Acid Sphingomyelinase Deficiency Type B
M Teresa Cardoso, Inherited Metabolic Disorders Reference Centre, Internal Medicine Department, São João University Hospital Center, Portugal

EP-168 Characteristics of South Korean patients with late onset Pompe disease: Data from the Pompe Registry
Beom Hee Lee, Department of Pediatrics, Asan Medical Center, University of Ulsan College of Medicine, Republic of Korea

EP-169 Sebelipase Alfa Six Months Treatment. Clinical Outcomes in Two Adult Siblings with Lysosomal Acid Lipase Deficiency and Long Time Evolution Disease
M Teresa Cardoso, Inherited Metabolic Disorders Reference Centre, Internal Medicine Department, São João University Hospital Center, Portugal

EP-170 Clinical, Epidemiological, Diagnostic and Therapeutic Profile of Fabry Disease Patients: a study based on the Brazilian Network for Rare Diseases
Vinícius Lima Ferraz, Hospital de Clínicas de Porto Alegre, Brazil

EP-171 Genetic insights into late-infantile Galactosialidosis: A Novel CTSA variant in a Thai family
Lukana Ngiwsara, Chulabhorn Research Institute, Thailand

EP-172 Evaluating the Quality of Life in Mucopolysaccharidosis
Aliye Gulbahce, Kocaeli City Hospital, Turkey

EP-173 Two Rare Cases of Saposin B Deficiency Mimicking Metachromatic Leukodystrophy
Merve Yoldas Celik, Adana City Training and Research Hospital, Department of Pediatric Metabolism, Turkey

EP-174 Long-term impact of pabinafusp alfa on disease burden in hunter syndrome: a 6-year follow-up of patient-reported outcomes
Marco A Curiati, Centro de Referencia em Erros Inatos do Metabolismo (CREIM-UNIFESP), Brazil

EP-175 Efficacy of early intracerebroventricular enzyme replacement therapy in a Japanese infantile patient with neuronopathic mucopolysaccharidosis type II
Sumito Dateki, Department of Pediatrics, Nagasaki University Graduate School of Biomedical Sciences, Japan

EP-176 Pompe physiotherapy service improvement - Facilitating monitoring in a specialist metabolic centre
Emma Louise Sarrecchia, Adult Inherited Metabolic Diseases, UK

EP-177 A Quarter Century with MPS I: Clinical Manifestations and Long-Term Outcomes from a Single Center
Sakina Mammadova, Ege University Medical Faculty Department of Pediatrics Division of Inborn Error of Metabolism and Nutrition, Turkey

EP-178 SaposinB deficiency/A Case of Variyant-Metachromatic Leukodystrophy with PSAP Mutation
Ayse Yuksel Yanbolu, Ege University Faculty of Medicine Children's Hospital, Department of Pediatric Metabolism and Nutrition, Turkey

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Vinícius Lima Ferraz, Hospital de Clinicas de Porto Alegre, Brazil

EP-180 Successful Reintroduction of Enzyme Replacement Therapy in a Patient with Pompe Disease Using Rapid Drug Desensitization
Takehiro Homma, Department of Metabolism, Genetic Medical Center, Chiba Children's Hospital, Japan

EP-181 TRIGGER FINGER IN CHILDREN WITH MUCOPOLYSACCHARIDOSIS TYPE I
Boudewijn A.W. van Binsbergen, University Medical Center Utrecht, Department of Plastic, Reconstructive, and Hand Surgery, Netherlands / Wilhelmina Children's Hospital Utrecht, Department of Plastic, Reconstructive, and Hand Surgery, Netherlands / Wilhelmina Children's Hospital Utrecht, Department of Metabolic Diseases, Netherlands

EP-182 Hunter Syndrome in One of Dichorionic Twins: A Case Report Emphasizing Diagnostic Discrepancy and Therapeutic Challenges
Hasriza Eka Putra, Division of Nutrition and Metabolic Disease, Departement of Child Health, Faculty of Medicine, Universitas Indonesia, Dr. Cipto Mangunkusumo Hospital, Indonesia

EP-183 Clinical Evaluation First: MPS Type 2 diagnosis confirmed by RNA sequencing analysis
Asli Durmus, Trabzon Kanuni Training And Research Hospital, Turkey

EP-184 A case of Infantile-Onset Pompe Disease treated with High-Dose Enzyme Replacement Therapy starting on day 31
Yusuke Miyagi, Dept. Pediatrics and Child Health, Kurume University, Japan

EP-185 Pycnodysostosis two Tunisian new cases
Kamel Monastiri, Teaching Hospital of Monastir, Tunisia

EP-186 Safety of Accelerated Infusion of Next-Generation Enzyme Replacement Therapy (pabinafusp alfa) in Mucopolysaccharidosis Type II
Tetsumin So, National Center for Child Health and Development, Japan

EP-187 Sandhoff Disease: An Infantile Case and Family Study
Kamel Monastiri, Department of Intensive Care and Neonatal Medicine, Teaching Hospital of Monastir, Tunisia

EP-188 Mucopolysaccharidosis type VII: a case report
Ngoc Thi Bich Can, Vietnam National Children's Hospital, Vietnam

EP-189 15 Year Experience in MPS type 2 Disease; Diagnosis and Optimal Care at the Iranian Reference Center
Fatemeh Hadipour, Medical Genetic Department, Atieh Hospital, Iran

EP-190 Alpha Mannosidosis in Siblings: A Clinical Continuum?
Hui Bein Chew, Department of Genetics, Kuala Lumpur Hospital, Malaysia

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Harun Yildiz, Ankara Etlik City Hospital, Turkey

EP-192 Unraveling the Gut Puzzle: Lessons from Irritable Bowel Syndrome in Managing Gastrointestinal Symptoms in Paediatric Fabry Disease
Anna-Maria Wiesinger, Institute of Inherited Metabolic Diseases, Paracelsus Medical University, Austria / European Reference Network for Hereditary Metabolic Diseases, MetabERN, Udine, Italy

EP-193 Stroke in a child treated as Methylene tetrahydrofolate reductase deficiency and proved to be Fabry disease
Doaa Ali Alsultan, Genetics and Metabolic Medicine Division, Department of Pediatrics, Prince Sultan Military Medical City, Saudi Arabia

EP-194 Comparative study of participants with Hunter syndrome receiving enzyme replacement therapy and hematopoietic stem cell therapy: impact on growth and overall performance
Aadhira Anandkumar Nair, FRIGE's Institute of Human Genetics, India

EP-195 Title: Evaluation of Circadian Rhythm Profile in Patients with Lysosomal Storage Disorders Receiving Enzyme Replacement Therapy
Hanim Seyma Topuz, Basaksehir Cam and Sakura City Hospital, Turkey

EP-196 Niemann-Pick Disease (NPD) type AB presented with ataxia while on enzyme therapy
Doaa Ali Alsultan, Genetics and Metabolic Medicine Division, Department of Pediatrics, Prince Sultan Military Medical City, Saudi Arabia

EP-197 Pathological and biochemical studies of an autopsy case with Gaucher disease treated with enzyme replacement therapy
Hayato Naruse, Department of Pediatrics, The Jikei University School of Medicine, Japan

EP-198 Marked Improvements in Airway Abnormalities and Multifaceted Outcomes after Two Years Switching to Avalglucosidase Alfa: Evaluation of A 19-Year-Old Male Diagnosed with Late-Onset Pompe Disease
Li-Zhen Chen, Taipei Veterans General Hospital, Taiwan

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Kamel Monastiri, Teaching Hospital of Monastir, Tunisia

EP-200 The importance of Holistic management in the progression of Late Onset Pompe Disease
Sarah Steeds, University Hospitals Birmingham, UK

EP-201 Early enzyme replacement therapy guided by HyperCKemia in asymptomatic late-onset Pompe disease.
Yoshimi Fujita, Department of Pediatrics, The Jikei University School of Medicine, Japan

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Sibel Burcak Sahin, İzmir City Hospital , Turkey

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Meta Herdiana Hanindita, Dr. Soetomo General Hospital/Medical Faculty, Airlangga University, Indonesia

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Assel Tulebayeva, Scientific Center of Pediatrics and Pediatric Surgery, Kazakhstan / Asfendiyarov Kazakh National Medical University, Kazakhstan

EP-205 5-year experience in familial cases with classic Fabry disease switching from ERT to pharmacological chaperon
YoungBae Sohn, Ajou University Hospital, Republic of Korea

EP-206 Northern Irish Experience of Adult Niemann-Pick Patients
Tabib Dabir, Belfast Health & Social Care Trust, UK

EP-207 Clinical manifestations and molecular genetics of seven patients with Niemann-Pick type-C: a case series with a novel variant
Engin Kose, Department of Pediatric Metabolism, Ankara University Faculty of Medicine, Turkey

EP-208 Efficacy of combination therapy with laronidase and hematopoietic stem cell transplantation for a case of mucopolysaccharidosis type 1
Chida Rie, Tokyo Medical University Hachioji Medical Center, Department of Pediatrics, Japan

EP-209 Mucolipidosis type III gamma in a boy with joint pain: a case report
Natalia V. Zhurkova, Research Institute of Pediatrics and Children's Health in Petrovsky National Research Centre of Surgery, Russia

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Esra Er, University of Health Sciences Dr. Behcet Uz Child Disease and Pediatric Surgery Training and Research Hospital, Turkey

EP-211 Comparison of Testing Methods in Newborn Screening for Lysosomal Storage Diseases
Shinichiro Yoshida, KM Biologics Co., Ltd., Japan

EP-212 Hand disorders in mucopolysaccharidosis type I: A review
Boudewijn A.W. van Binsbergen, University Medical Center Utrecht, Department of Plastic, Reconstructive, and Hand Surgery, Netherlands / Wilhelmina Children's Hospital Utrecht, Department of Plastic, Reconstructive, and Hand Surgery, Netherlands / Wilhelmina Children's Hospital Utrecht, Department of Metabolic Diseases, Netherlands

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Kei Hirayama, Department of Pediatrics, Fukuchiyama City Hospital, Japan

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Jingtao Zhang, Children's Medical Center, Peking University First Hospital, China

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Arzu Selamioglu, Department of Rare Diseases, Institute of Graduate Studies in Health Sciences, Istanbul University, Turkey

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Esra Sayar, Ankara Etlik City Hospital, Department of Pediatrics, Metabolism Unit, Turkey

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Ana Felipe-Rucian, University Hosital Vall d'Hebron, Spain

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Mathanky Rajalingam, Department of Chemical Pathology, Lady Ridgeway Hospital for Children, Sri Lanka

EP-220 A Rare Presentation of Duchenne Muscular Dystrophy: Acute Rhabdomyolysis
Esra Sayar, Ankara Etlik City Hospital, Department of Pediatrics, Metabolism Unit, Turkey

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Kamel Monastiri, Department of ICU and Neonatal Medicine, Teaching Hospital of Monastir, Tunisia

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Dan Ross Brooks, Department of Molecular and Human Genetics, Department of Pediatrics, Baylor College of Medicine, USA / Department of Molecular and Human Genetics, Texas Children's Hospital, USA

EP-223 Evaluation of amino acid ratio combinations as a diagnostic tool for patients with pyruvate dehydrogenase complex deficiency: A single center experience
Xiaowei Fu, Department of pathology, University of Tennessee Health Science Center, USA

EP-224 Pediatric Mitochondrial Disease: Clinical and Genetic Insights From a Single-Center Cohort
Halil Tuna Akar, Ankara Etlik City Hospital Department of Pediatric Metabolism, Turkey

EP-225 MT-ATP6 related mitochondrial disease with polyneuropathy and basal ganglia lesion, case report.
Sabine Laktina, Childrens Clinikal University Hospital, Latvia / Riga Stradins University, Latvia

EP-226 Contribution of Expanded Newborn Screening in the diagnosis of not included in the screening disorder.
Kochkina Diana, North-Eastern Federal University, Russia / Republic Hospital No. 1, Russia

EP-227 Improvement in myopathy, lactate and physical function using taurine supplementation in a child with MELAS
Sarah Donoghue, Metabolic Department, Women's and Children's Hospital, Australia

EP-228 Lack of response to L-lysine supplement in a patient with KARS1-related congenital deafness and adult-onset progressive leukoencephalopathy
Intisar Talib Al Fahdi, Oman medical specialty board, Oman

EP-229 Modeling the Crosstalk of Peripheral Neuropathy in Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency: Implications for Mitochondrial Function and Therapy
Chen Zhang, Research Unit for Molecular Medicine, Department of Clinical Medicine, Aarhus University, Denmark

EP-230 Neonatal hypoglycemia and proximal tubulopathy: a case report of two novel BCS1L variants
Laura Rubert, Department of Pediatrics, Regional Centre for Newborn Screening, Diagnosis and Treatment of Inherited Metabolic Diseases and Congenital Endocrine Dise Azienda Ospedaliera Universitaria Integrata of Verona, Italy

EP-231 Acyl-CoA dehydrogenases family number 9 (ACAD9) deficiency with renal tubular dysfunction
Yoshimitsu Osawa, Department of Pediatrics, Gunma University Graduate School of Medicine, Japan / Department of Endocrinology and Metabolism, Gunma Children's Medical Center, Japan

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bengu arslan, Sakarya University Faculty of Medicine, Department of Pediatric Metabolism, Turkey

EP-233 A Leigh Syndrome Case with Dramatic Improvement Following Alkaline Ionized Water Supplementation
Hanim Seyma Topuz, Basaksehie Cam and Sakura City Hospital, Turkey

EP-234 Extended follow-up of a case of Leigh Syndrome caused by NDUFAF6 compound heterozygous variants
Maria Novelli, Sapienza University, Italy

EP-235 Sildenafil Prolongs Survival and Reduces Decompensation in Leigh Syndrome: A Case Series of Two Siblings
Irma Sri Hidayati, Division of Nutrition and Metabolic Disease, Department of Child Health, Faculty of Medicine, Universitas Indonesia/ Dr. Cipto Mangunkusumo National Central Hospital, Indonesia / Division of Nutrition and Metabolic Disease, Department of Child Health, Faculty of Medicine, Public Health and Nursing, Universitas Gadjah Mada/ Dr. Sardjito Hospital, Indonesia

EP-236 Response to L-tryptophan treatment in a patient with WARS2 deficiency
Agnese De Giorgi, Department of Human Neuroscience, Sapienza University of Rome, Italy

EP-237 Amino acid ratios as biomarkers for pyruvate dehydrogenase complex deficiency and for evaluating effectiveness of response to ketogenic diet in a UPMC cohort
Bianca Seminotti, UPMC Children's Hospital of Pittsburgh, USA / Division of Genetic and Genomic Medicine, Department of Pediatrics, University of Pittsburgh School of Medicine, USA

EP-238 A novel mutation FBXL4 gene with an unusual clinical sign
Ayse Nur Altun, Gazi Yaşargil Training and Research Hospital, Department of Pediatrics, Division of Pediatric Metabolic Diseases, Turkey

EP-239 PRIMARY COENZYME Q10 DEFECT: A TUNISIAN CASE
Kamel Monastiri, Teaching Hospital of Monastir, Tunisia

EP-240 Mitochondrial neurogastrointestinal encephalopathy and Mitochondrial DNA 4977-bp deletion
maria carmo macario, Neurology, Coimbra University Hospital, Coimbra Local Healt Unit, Portugal

EP-241 MELAS Syndrome in Siblings with Discordant Clinical Presentation
Hadeel a alrabee, Prince Sultan Military Hospital , Saudi Arabia

EP-242 Case Report: A Rare Variant in the Mitochondrial MT-ND1 Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON)
Martin Engvall, Karolinska Institutet, Sweden / Centre for Inherited Metabolic Diseases, Karolinska University Hospital, Sweden

EP-243 Ketogenic diet for pyruvate dehydrogenase complex deficiency in children: a case series of 7 patients
Irina Artamonova, Almazov National Medical Research Centre, Russia

EP-244 Long-Read Genome Sequencing identifies Biallelic ACAD9 Variants in two Infants with Severe Lactic Acidosis and Hypertrophic Cardiomyopathy
Rungroj Thangpong, Center of Excellence for Medical Genomics, Department of Pediatrics, Faculty of Medicine, Chulalongkorn University, Thailand

EP-245 Endocrinologic Manifestations in Mitochondrial DNA Depletion Syndrome Associated with Biallelic LIG3 Variants
Sukdong Yoo, Pusan National University Children's Hospital, Republic of Korea

EP-246 Deep intronic mutation leads to case series of pyruvate dehydrogenase E3 binding protein deficiency in Saudi Arabia.
Hanem Sayed Ahmed Abdelraouf, PSMMC, Saudi Arabia

EP-247 10 years follow up of a patient with Pearson syndrome
Jana Saligova, Children's Faculty Hospital, Slovakia

EP-248 Comparison of Plasma Acylcarnitine and Urinary Organic Acid analysis in Combined Oxidative Phosphorylation Deficiency 23: Two Cases from China
Hao Liu, Chongqing Health Center for Women and Children/Women and Children's Hospital of Chongqing Medical University, China

EP-249 A Rare Clinical Presentation of TWNK Gene Mutation: A Case Presenting with Renal Tubular Acidosis
Burcu Koseci, Adana City Hospital, Department of Pediatric Nutrition and Metabolism, Turkey

EP-250 Cardiomyopathy as a novel phenotypic feature in NSUN3-related mitochondrial disease
Ayse Senol Ersak, Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Metabolism and Nutrition, Turkey

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Sherry Fang, Department of Paediatric Inherited Metabolic Disease Great Ormond Street Hospital NHS Foundation Trust, UK

EP-252 Aromatic L-amino acid decarboxylase deficiency: first case in Bali, Indonesia
I Gusti Lanang Sidiartha, Division of Nutrition and Metabolic Disease, Department of Child Health, Faculty of Medicine, Universitas Udayana - Ngoerah Hospital, Indonesia

EP-253 Dopa responsive dystonia caused by a novel variant in TH gene in a Sudanese child
Esraa abdelaziz Elhassan, Prince Sultan Military Medical City, Saudi Arabia

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Azusa Tamada, Research Institute of Medical Mass Spectrometry, Kurume University School of Medicine, Japan

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Caiqi Du, Department of Pediatrics, Tongji Children's Hospital, Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, China

EP-256 Resistant Hypertension in a Patient with Carbonic Anhydrase II Deficiency
bengu arslan, Sakarya University Faculty of Medicine, Department of Pediatric Metabolism, Turkey

EP-257 Persistent central cyanosis of unknown cause in adolescents: A rare case of congenital methemoglobinemia due to CYB5R3 gene mutation - Diagnostic difficulties and challenges
Tuan Minh Tran, Department of Pediatric, Faculty of Medicine, University of Medicine and Pharmacy at Ho Chi Minh City, Vietnam

EP-258 Hypophosphatasia-diagnostic and therapeutic challenge based on long-term follow-up of patients at the Polish Mother's Memorial Health Institute in Lodz Poland
Izabela Michalus, Department od Endocrinology and Metabolic Diseases, Polish Mother's Memorial Health Institute in Lodz, Poland / Outpatient Clinic For Inborn Metabolic Disorders, Poland

EP-259 HYALINE FIBROMATOSIS SYNDROME A RARE DISEASE
Kamel Monastiri, Department of Intensive Care and Neonatal Medicine, Teaching Hospital of Monastir, Tunisia

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Maria Camila Leon Sanabria, Institute of Human Genetics, Pontificia Universidad Javeriana, Colombia / Hospital La Victoria, Colombia

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Silvia Radenkovic, Department of Genetics, Section Metabolic Diagnostics, University Medical Centre Utrecht, Netherlands

EP-262 Gaucher disease eradicated by a change in newborn screening method? Oregon's experience with screening by tandem mass spectrometry
Sarah Viall, Oregon Health & Science University, USA

EP-263 Genotypic and Phenotypic Comparison in Patients with Biallelic DUOX2 Mutations: Differentiating Transient and Permanent Congenital Hypothyroidism
Chih-Ya Cheng, Department of Pediatrics, Taipei Veterans General Hospital, Taiwan

EP-264 Five-Year Analysis of Fabry Disease Detected by Newborn Screening and Family Screening in Miyazaki, Japan
Satoru Meiri, Division of Pediatrics, Department of Developmental and Urological Reproductive Medicine, Faculty of Medicine, University of Miyazaki, Japan

EP-265 Evaluation of the ImmunoIVD Spot-it kit for Newborn Screening for Spinal Muscular Atrophy in New Zealand
Sandra Divanisova, LabPLUS, Auckland City Hospital, Health New Zealand

EP-266 Newborn Screening for Biotinidase Deficiency: An Initial Experience
Neerja Gupta, Division of Genetics, Department of Pediatrics, All India Institute of Medical Sciences, India

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Michael H. Gelb, University of Washington, USA / Institute for Protein Design, University of Washington, USA

EP-268 Expanded Newborn Screening for Inborn Errors of Metabolism in Greece: One Year Experience of the National Newborn Screening Program
Dimitrios Platis, Institute of Child Health, Greece

EP-269 Neonatal Jaundice and Its Effect on Amino Acid and Acylcarnitine Profiles: Findings from Expanded Newborn Screening
Preawwalee Wintachai, Clinical Laboratory Section, Srinagarind Hospital, Faculty of Medicine, Khon Kaen University, Thailand / Srinagarind Excellent Laboratory, Faculty of medicine, Khon Kaen University, Thailand

EP-270 Inherited Metabolic Disorders Among Neonates and Infants in Critical Condition Admitted to a Tertiary Hospital
Hector Alejandro Zambrano-Herrera, Department of Genetics, Facultad de Medicina y Hospital Universitario "Dr. José Eleuterio González", Universidad Autónoma de Nuevo León, Mexico

EP-271 Necessity of second blood spot newborn screening by tandem mass spectrometry in maternal COVID-19 infection
Khunton Wichajarn, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine, Khon Kaen University, Thailand / Center of Excellence in Precision Medicine, Srinagarind Hospital, Khon Kaen University, Thailand

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Muhammad Wasim, Maternal and Children's Health Research Institute, Shunde Women and Children's Hospital, Guangdong Medical University, China

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Sakura Morishima, Department of Pediatrics, Oita University School of Medicine, Japan

EP-274 Carrier-Screening for Spinal Muscular Atrophy in Peri-Urban Setting in Karachi, Pakistan
Bushra Afroze, Department of Paediatrics and child health. Aga Khan University Hospital, Pakistan

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Hisato Aihara, Department of Pediatrics, Juntendo University , Japan / Department of Pediatrics, Tokyo Metropolitan Toshima Hospital, Japan

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Jennifer Burton, University of Illinois College of Medicine, USA

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Maria Lucia Tommolini, University G. d'Annunzio of Chieti-Pescara, Italy

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Sook Za Kim, KSZ Children's Hospital, Republic of Korea

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Brenna Downey, University of Illinois at Chicago, USA

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Sharmila Kiss, The Royal Children's Hospital, Australia

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Martina Zandl-Lang, Department of Pediatrics and Adolescent Medicine, Joint Facilities,Medical University of Graz, Austria / Department of Pediatrics, Division of General Pediatrics, Medical University of Graz, Austria

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Sabine Scholl-Buergi, Department of Pediatrics I, Medical University of Innsbruck, Austria

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Yuqin Wang, Swansea University, UK

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Eunice Ya Ping Lim, Nursing Clinical Services, KK Women's and Children's Hospital, Singapore

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Sofia Panato Ribeiro, Post-graduate Program in Medical Sciences, Brazil

EP-286 BREASTFEEDING PRACTICES IN CHILDREN WITH MAPLE SYRUP URINE DISEASE
Sofia Panato Ribeiro, Post-graduate Program in Medical Sciences, Brazil

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Sarah Steeds, Department for Inherited Metabolic Disorders, University Hospitals Birmingham NHS Foundation Trust, UK

EP-288 The enzyme replacement therapy in pediatric patients with lysosomal diseases: the nurses´ experience at the Children Hospital Giovanni XXIII in Bari (ITALY)
Annamaria Pagano, Ospedale pediatrico Giovanni XXIII - Policlinico, Italy

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Noura AlDhaheri, College of Medicine and Health Sciences- UAE University, UAE / Tawam Hospital, UAE

EP-290 Maintenance therapy with carglumic acid reduces metabolic decompensation and hospital admission in patients with organic acidemia: A single center experience
Soojin Hwang, Department of Pediatrics, Medical Genetics Center, Asan Medical Center, University of Ulsan College of Medicine, Republic of Korea

EP-291 Congenital Neutropenia and global developmental delay in CLPB deficiency : a Case report
Hyunwoo Bae, Department of Pediatrics, School of Medicine, Kyungpook National University, Republic of Korea

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elham saeed bagrayn, Genetics and Metabolic Medicine Division, Department of Pediatrics, Prince Sultan Military Medical City, Saudi Arabia

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Ying Zhang, Fuzhou First General Hospital Affiliated with Fujian Medical University, Fuzhou Children's Hospital of Fujian Medical University, China

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Tumelo M Satekge, Division of Chemical Pathology, University of Limpopo, South Africa

EP-295 CLINICAL-BIOLOGICAL HETEROGENEITY PRESENTATION OF METHYLMALONIC ACIDEMIA TYPE MMUT
Kamel Monastiri, Department of Intensive Care and Neonatal Medicine, Teaching Hospital of Monastir, Tunisia

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Kaori Fukui, Department of Pediatrics and Child Health, Kurume University School of Medicine, Japan

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Pedro Louro, Local Health Unit of São João, Portugal / Faculty of Health Sciences, University of Beira Interior, Covilhã, Portugal

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Yoo-Mi Kim, Chungnam National University Sejong Hospital, Republic of Korea

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Amanda Caro, Institute for the Study of Inborn Errors of Metabolism. Pontificia Universidad Javeriana, Colombia

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Neti Nurani, Child Health Department, Faculty of Medicine, Universitas Gadjah Mada, Dr Sardjito General Hiospital, Indonesia

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Silvia Maria Meavilla Olivas, Paediatric Gastroenterology, Hepatology and Nutrition Department, Metabolic Diseases Unit, Sant Joan de Déu Hospital, Spain

EP-302 Fanconi Renotubular Syndrome Due to a Novel De Novo EHHADH Variant: Case Report and Therapeutic Implications
Cassandra Afseth, Northwestern University Feinberg School of Medicine, USA

EP-303 Combination Therapy with Metreleptin and GLP-1R Antagonist in a Patient with Generalized Lipodystrophy-Associated Progeroid Syndrome, Case Report
Antti Saari, Chief Physician, Pediatric endocrinologist, Rare Disease Unit, Kuopio University Hospital, Finland

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Ee Shien Tan, Genetics Service,KK Women's and Children's Hospital, Singapore

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Hiroki KAWAI, Department of Pediatrics, Gifu University Graduate School of Medicine, Japan / Division of Genomics Research, Life Science Research Center, Gifu University, Japan

EP-306 A novel variant in ATP8B1 gene associated with Byler syndrome.
Viktoriia Sofronova, North-Eastern Federal University, Russia

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Elaine Jayadiwangsa, Metabolic Department, Women's and Children's Hospital, Australia

EP-308 A 24-year-old woman with Smith-Lemli-Opitz syndrome: an atypical silent case
Hadeel a alrabee, Prince Sultan Military Hospital , Saudi Arabia

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Yanqin Ying, Department of Pediatrics, Tongji Hospital, Tongji Medical College, Huazhong University of Science and Technology, China

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William J Griffiths, Swansea University, UK

22. Phenylketonuria

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Tatiana V Bushueva, National Medical Research Center of Children's Health, Ministry of Health of the Russian Federation, Russia / Research Centre for Medical Genetics, Russia

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Melissa Dawn Lah, Indiana University, USA

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Vito Di Tullio, UOC Metabolic and Genetic Diseases, Giovanni XXIII Paediatric Hospital, Italy

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Anita MacDonald, Department of Dietetics, Birmingham Women's and Children's Hospital, UK

EP-315 Late-diagnosed PKU patients - a forgotten population?
Kirsten Kiaer Ahring, Clinic for PKU, Copenhagen University Hospital, Denmark

EP-316 Quality of life in adolescents with phenylketonuria
Solaf Mohamed Elsayed, Medical Genetics Department , Ain Shams University, Egypt

EP-317 Bowel Preparation in Phenylketonuria: Navigating Dietary Challenges. A Case Report
Anita MacDonald, Department of Dietetics, Birmingham Women's and Children's Hospital, UK

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SALWA Abdullah Alharbi, Prince Sultan Military Medical City, Saudi Arabia

EP-319 Dynamic associations of mood states and lifestyle behaviors in youth affected by phenylketonuria
Jennifer Glaus, Division of Child and Adolescent Psychiatry, Department of Psychiatry, Lausanne University Hospital and Lausanne University, Switzerland

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Olga Yaneth Echeverri, Instituto de Errores Innatos del Metabolismo. Pontificia Universidad Javeriana, Colombia

EP-322 Alpha Cell Hyperplasia and Tumor Risk in Glucagon Receptor Deficient Mice Is Ameliorated by a Low-Protein Diet
Hong Li, Department of Human Genetics, Emory University School of Medicine, USA

EP-323 Health literacy interventions for inborn errors of metabolism: a scoping review
Bruna Bento dos Santos, Health Technology Assessment Group in Clinical Genetics, Hospital de Clínicas de Porto Alegre, Brazil / Graduate Program in Collective Health, University of Brasília, Brazil

EP-324 Diagnostic Utility of Biochemical and Molecular Genetic Testing in Patients with Unexplained Wide Anion Gap Metabolic Acidosis
Hathaipat Vaseenon, Division of Medical Genetics, Department of Pediatrics, Faculty of Medicine Ramathibodi Hospital, Mahidol University, Thailand / Department of Pediatrics, Faculty of Medicine Vajira Hospital, Navamindradhiraj University, Thailand

EP-325 Acute Hepatocellular Failure in Children
khouloud ben mansour, Rabta Hospital, Tunisia

EP-326 Inborn Errors of metabolism and the definition of ultra-rare diseases: a scoping review
Bruna Bento dos santos, Health Technology Assessment Group in Clinical Genetics, Hospital de Clínicas de Porto Alegre, Brazil / Graduate Program in Collective Health, University of Brasília, Brazil

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EP-327 Hidden in Plain Sight: Adult Onset CPS1 Deficiency in Two Patients with Hyperammonemic Encephalopathy
Rana Aljaberi, Emory University, USA

EP-328 Identification of a Novel ASS1 Mutation in neonatal-onset Citrullinemia Type 1
Hiroaki Sugiyama, NHO Okayama Medical Center Neonatology Department, Japan / NHO Okayama Medical Center Pediatric Department, Japan

EP-329 Clinical Spectrum and Outcome in 5 patients with CA5A Deficiecny in India
Ketki V Kudalkar, NIRMAN, India

EP-330 Genetic, Biochemical, and Phenotypic Characterization of a False-Negative Newborn Screening Case of HHH Syndrome in Southeastern Mexico.
Felix Julian Campos-Garcia, Hospital General "Dr. Agustín O'Horán", Mexico / Universidad Marista Merida, Mexico

EP-331 A Treatable Mimic: Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome Presenting as Complex Hereditary Spastic Paraparesis
Isabella Lince-Rivera, Universidad Militar Nueva Granada, Colombia / Universidad Militar Nueva Granada, Colombia / Instituto Roosevelt, Colombia / Instituto Roosevelt, Colombia / Instituto Roosevelt, Colombia

EP-332 Unraveling the pathomechanisms of a specific OTC-variant by applying induced pluripotent stem cell technology
Alexander Laemmle, Division of Pediatric Endocrinology, Diabetology and Metabolism, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland / University Institute of Clinical Chemistry, Inselspital, Bern University Hospital, University of Bern, Switzerland / Department of Biomedical Research, University of Bern, Switzerland / CureOTCD, Canada

EP-333 Hyperammonemia in a child with short bowel syndrome: urea cycle disorder or complication of altered gastrointestinal function?
Rachel VanCoillie, University Hospitals, USA

EP-334 When the Carrier Is the Patient: A Case Series of Symptomatic Women with OTC Deficiency
Jenniffer Andrea Romero Morales, Clinical Genetics Unit, Hospital Universitario San Ignacio, Colombia / Institute of Human Genetics, Pontificia Universidad Javeriana, Colombia

EP-335 Case report: Challenges in Diagnosis and Treatment of a Newborn with Carbamoyl Phosphate Synthase 1 Deficiency
Maria Galuh Kamenyangan Sari, Division of Nutrition and Metabolic Disease, Departement of Child Health, Faculty of Medicine, Universitas Indonesia, Dr. Cipto Mangunkusumo Hospital, Indonesia

EP-336 Severe neonatal OTCD in one of dichorionic twins due to a novel OTC gene deletion: clinical divergence and genetic insights from a family case
Francesca Cappozzo, Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DINOGMI), University of Genoa, Italy

EP-337 An Unusual Presentation of Ornithine Transcarbamylase Deficiency Starting from Hyperthyroidism
Zhenran Xu, Children's Hospital of Fudan University, China

EP-338 Clinical analysis of burosumab in the treatment of X-linked hypophosphatemic rickets
fan yang, Clinical Research Center, Shanghai Children's Medical Center, School of Medicine, Shanghai Jiao Tong University, China

EP-339 Rarely seen cause of hyperammonemia
Mariela Mercedes De Los Santos Mercedes, Paediatric Gastroenterology, Hepatology and Nutrition, Spain

EP-340 A Neonatal Case of CPS1 Deficiency Complicated by Acute Heart Failure: Successful Management Without ECMO
Noritaka Yamamoto, Children's Medical Center, Osaka City General Hospital, , Japan

EP-341 Long-term Sodium Benzoate Oral Administration for Neonate with Urea Cycle Disorder (UCD) CPS1 in Limited Resource Setting
Nur Aisiyah Widjaja, Universitas Airlangga Faculty of Medicine, Indonesia / Dr. Soetomo General Academic Hospital, Indonesia

25. Late-Breaking Research

EP-342 Biostatistics Collaboration Team, Research Core Center, National Cancer Center, Goyang, Republic of Korea
Aram Yang, Kangbuk Samsung Hospital, Sungkyunkwan University School of Medicine, Republic of Korea

EP-343 Identification and functional characterization of a novel SEMA3A exon deletion variant in Kallmann Syndrome
Shaolian Zang, Shanghai Children's Hospital, China

EP-344 Withdrawal

EP-345 Emergency Algorithm for the Investigation and Management of Pediatric Patients with Suspected Inborn Errors of Metabolism
Daniel Zamanfar, Mazandaran University of Medical Sciences, Iran

EP-346 Glycogen Storage Disease Type I in Brazilian Patients: A Metabolomic Approach
Fernanda Bertao Scalco, LABEIM IQ Universidade Federal do Rio de Janeiro, Brazil

EP-347 Broadening the clinical spectrum of NDUFAF3–related disorder:functional characterization as a key diagnostic tool
Alessandra Verde, University of Brescia, Italy

EP-348 Oral trehalose improves histological and behavior symptoms of mucopolysaccharidosis type II in iduronate 2-sulfatase deficiency mice
Jaeeun Chung, Busan St.Mary's hospital, Republic of Korea

EP-349 A Very Rare Case of Metabolic Myopathy:Tarui Disease
Aliye Gulbahce, Kocaeli City Hospital, Pediatric Metabolism, Turkey

EP-350 Diagnostic and treatment Mucopolysaccharidosis type IVA in Indonesia: case study from West Java, Indonesia
Viramitha Kusnandi Rusmil, Nutrition and Metabolic Disease Division, Department of Pediatrics, Faculty of Medicine Universitas Padjadjarn, Indonesia / Dr Hasan Sadikin Hospital, Indonesia

EP-351 Hypoinsulinemic Hypoglycemia with Hemihypertrophy from AKT2 Gain-of-function Mutation
Elizabeth Grace Martinez Perez, University of the Philippines-Philippine General Hospital, Philippines

EP-352 Clinical characteristics, genetic spectrum and therapeutic effects of 51 male patients with idiopathic hypogonadotropic hypogonadism from southern China
Yunting Lin, Guangzhou Women and Children's Medical Center, China

EP-353 Virtual Screening of AASS (Aminoadipate Semialdehyde Synthase) Inhibitors from the FDA approved drugs for Glutaric Acidemia Type I Substrate Reduction Therapy
Akif Altun, Department of Rare Diseases, Institute of Graduate Studies in Health Sciences, Istanbul University, Turkey

EP-354 CONTIGUOUS GENE DELETION SYNDROME IN Xp21: ASSOCIATED WITH GLYCEROL KINASE DEFICIENCY, AN UNUSUAL
Andrea Salgado Bustos, Universidad Militar Nueva Granada, Colombia

EP-355 A STUDY TO FIND THE OCCURENCE OF ORGANIC ACIDEMIAS AND AMINO ACIDEMIAS IN CRITICALLY ILL NEWBORNS ADMITTED IN NEONATAL INTENSIVE CARE UNIT
Priyanshu Mathur, SMS medical college, India

EP-356 Whole genome sequencing for the PKU diagnostic
Olga Shchagina, Research centre for medical genetics, Russia

EP-357 Characterization of Primary Macrophages from Fabry Disease Patients
Veronica Lentini, Department of Haematology, Royal Free Hospital, University College London, UK / University of Sassari, Italy

EP-358 The mechanism of 46, XY DSD caused by four NR5A1 gene variations
Qingxu Liu, Shanghai Children's Hospital, School of medicine, Shanghai Jiao Tong University, China

EP-359 Efficacy of Zoledronic Acid in a Pediatric Hajdu-Cheney Syndrome: A Four-Year Therapeutic Observation
Rong Du, Guangzhou women and children's medical center, China

EP-360 Population-Specific Diagnostic Algorithm for Biotinidase Deficiency: An Essential Step Towards Optimized Newborn Screening
Hafsa Majid, Newborn Screening Lab, Section of Chemical Pathology, Department of Pathology and Laboratory Medicine, Aga Khan University, Pakistan

EP-361 A Fatal Case of Encephalopathy Due to Ornithine Transcarbamylase Deficiency in a Newborn Male Infant
Hyunwoo Bae, Department of Pediatrics, School of Medicine, Kyungpook National University, Republic of Korea / Department of Pediatrics, Kyungpook National University Hospital, Republic of Korea

EP-362 EARS2 and Other Nuclear Genes as Critical Modifiers in Mitochondrial Disorders: Insights into RIRCD Characteristics and Pathogenesis
Tongyue Li, Department of Neurology, Beijing Children's Hospital, Capital Medical University, National Center for Children's Health, China

EP-363 Recognizing the Unseen: A Clinical and Genetic Profile of Metabolic Disorders in Newborns from Southern Vietnam
PHUONG THI HONG CHU, Department of Neonatology 2 - Metabolism & Genetics, Children's Hospital 1, Vietnam

EP-364 The Brain Can Recover: Cognitive Improvements After 4 Months of Dietary Re-Initiation in Adults with Phenylketonuria – #backtoclinic Study
Maximilian Pichler, University Clinic for Internal medicine III, Division of Endocrinology and Metabolism, Medical University of Vienna, Austria

EP-365 The Brain Doesn't Forget: Cognitive Consequences of Treatment Gaps in Phenylketonuria – #backtoclinic Study
Maximilian Pichler, University Clinic for Internal medicine III, Division of Endocrinology and Metabolism, Medical University of Vienna, Austria

EP-366 Progress Toward Including Wilson Disease in Washington State: Wilson Disease Soon to Join Washington's NBS panel
Sihoun Hahn, University of Washington School of Medicine, USA / Seattle Children's Hospital, USA / Key Proteo, Inc., USA

EP-367 Evaluation of Immune Dysregulation in Niemann-Pick Disease Type C
Hanim Aghakishili, Medical Faculty of Cerrahpasa, İstanbul University-Cerrahpasa, Department of Pediatrics, Division of Nutrition and Metabolism, Turkey

EP-368 Applicability of N-palmitoyl-O-phosphocholineserine as diagnostic biomarker for Niemann Pick Type C2 patients
Steffen Fischer, Centogene GmbH, Germany

EP-369 Restoration of Urea Cycle Function in OTC Deficiency: Glutamine Reduction Following ARCT-810 mRNA Therapy Across Two Clinical Studies
Benjamin Greener, Arcturus Therapeutics, USA

EP-370 The Impact of Breastfeeding on Nutritional and Clinical Outcomes in Organic Acidemias: Single Centre Experience
Esma Uygur, Acibadem Mehmet Ali Aydinlar University, Institute of Health Sciences, Department of Nutrition and Dietetics, Turkey / Istanbul University-Cerrahpasa, Cerrahpasa Faculty of Medicine, Division of Nutrition and Metabolism, Turkey

EP-371 Modification of the NBS diagnostic algorithm in response to an increasing number of elevated isovalerylcarnitine results in newborns
Ewa Glab-Jablonska, Institute of Mother and Child, Poland

EP-372 Implementation of a Health Care Transition Pathway for Adolescents with Inborn Errors of Metabolism: A Pilot Project
Brittany Marie Murray, Boston Children's Hospital, USA

EP-373 Metformin therapy in adults with classic maple syrup urine disease
Grace Loudon Meier, Clinic for Special Children, USA

EP-374 Genomics Prenatal Care in the Highly Inbred Society of Saudi Arabia.
Yara Zahi Alqahtani, King Faisal Specialist Hospital & Research Center, Saudi Arabia

EP-375 Clinical Tolerability and Biomarker Monitoring under Pegunigalsidase Treatment A Real-World Insight
Jasia Bokhari, University Hospital Zurich, Switzerland

EP-376 Hidden Burden: Frequency Of Disorders Of The Creatine Pathway In Children With Autism
Hanim Aghakishili, Department of Pediatrics, Division of Nutrition and Metabolism, Medical Faculty of Cerrahpasa, İstanbul University-Cerrahpasa, Turkey

EP-377 From Genes to Stones: Emerging Frontiers in Cystinuria, Integrating Diagnostics and Novel Adjunct Therapies
Romana Vulturar, Department of Cell and Molecular Biology, Iuliu Hatieganu University of Medicine and Pharmacy, Cluj-Napoca, Romania / Association for Innovation in Rare Inflammatory, Metabolic, Genetic Diseases INNOROG, Cluj-Napoca, Romania

EP-378 Newborn Screening and Genetic Analysis of Benign Hypermethioninemia in China
Fei Wang, Department of Endocrinology,Children's Hospital of Shanghai, Shanghai Jiaotong University, China

EP-379 Clinical and genetic characterization of a patient with vitamin D-dependent rickets type IA
Pin Li, Department of Endocrinology, Children's Hospital of Shanghai, Shanghai Jiaotong University, China

EP-380 Clinical characteristics and molecular genetic analysis of a pedigree with late-onset ornithine transcarbamylase deficiency
Mingyu Jiang, Shanghai Children's Hospital, School of medicine, Shanghai Jiao Tong University, China

EP-381 A TALE OF PARACETAMOL, PYROGLUTAMATE, AND THE MIMICRY OF METABOLIC DISEASE
Riona Singh-Gansan, Division of Chemical Pathology, Department of Pathology, University of Cape Town, South Africa / C17 Chemical Pathology Laboratory, National Health Laboratory Service, Groote Schuur Hospital, South Africa / Inherited Metabolic Disease Laboratory, National Health Laboratory Service, Red Cross War Memorial Children's Hospital, South Africa

EP-382 Fibroblasts from methylmalonic acidemia patients present alterations in cell respiration and glutathione-related redox homeostasis - Insights for Methylmalonic Acidemia
Cesar Ribeiro, Universidade Federal do ABC, Brazil / Programa de Pós-Graduação em Biossistemas, UFABC, Brazil

EP-383 RARE PRESENTATION OF EARLY-ONSET ORNITHINE TRANSCARBAMYLASE DEFICIENCY IN A FEMALE WITH SKEWED X-INACTIVATION
Riona Singh-Gansan, Division of Chemical Pathology, Department of Pathology, University of Cape Town, South Africa / C17 Chemical Pathology Laboratory, National Health Laboratory Service, Groote Schuur Hospital, South Africa / Inherited Metabolic Disease Laboratory, National Health Laboratory Service, Red Cross War Memorial Children's Hospital, South Africa

EP-384 Case Report: Identification of a treatment responsive variant of Propionic Acidaemia in South African infants
Malishca Devani Perumal, Inherited Metabolic Disease Laboratory, National Health Laboratory Service, Red Cross War Memorial Children's Hospital, South Africa

EP-385 Reye Syndrome in a South African Infant: A Fatal Case Unmasked by Metabolic Profiling
Malishca Devani Perumal, Inherited Metabolic Disease Laboratory, National Health Laboratory Service, Red Cross War Memorial Children's Hospital, South Africa

EP-386 Structural logic enables prediction of epistasis across proteins associated with inherited metabolic diseases
Aimee M. Dudley, Pacific Northwest Research Institute, USA

EP-387 Argininemia
A CASE report
Tisnasari Hafsah, Hasan Sadikin Hospital, Indonesia / Universitas Padjadjaran Bandung, Indonesia