SCIENTIFIC PROGRAM

SEPTEMBER, 8TH

17:45 | 18:30	SPDM Nutrition Group Meeting
19:00 | 19:45	Welcome drink

SEPTEMBER, 9TH

08:30	Registration opening
09:15	Symposium Opening
09:30 | 10:30	**Session I - IEM's mapping**
09:30 | 10:00	MetabERN - where are we now? *Maurizio Scarpa, IT*
10:00 | 10:30	Broadening the concept of IMD: Novel disorders and international classification *Carlos Ferreira, USA*
10:30 | 11:00	Coffee break
11:00 | 13:00	**Session II - Newborn Screening for IEM**
11:00 | 11:30	Newborn Screening in Europe and initiatives tp develop programmes in low - and middle-income countries *Jim Bonham, UK*
11:30 | 12:00	New tests and future directions in the NBS for IEM *Giancarlo LaMarca, IT*
12:00 | 12:30	Treatment follow-up of screened disorders *Paula Garcia, PT*
12:30 | 13:00	Oral Communications to the table
13:00 | 14:00	Lunch / Poster view
14:00 | 15:50	**Session III - From CbL to Hcy**
14:00 | 14:30	Remethylation disorders: from cobalamin absorption, intracellular pathways to MTHFR deficiency *Matthias Baumgartner, CH*
14:30 | 15:00	Inherited disorders of transsulfuration: clinical presentation, diagnosis, and recent advances in therapy? *Viktor Kozich, CZ*
15:00 | 15:30	The transsulfuration pathway at the crossroads between homocysteine and hydrogen sulfide metabolism: (dys)regulation and (patho)physiological implications *João Vicente, PT*
15:30 | 15:50	Oral communications to the table
15:50 | 16:20	Coffee break
16:20 | 17:20	**Session IV - IEM Nutritional and Pharmacological treatment**
16:20 | 16:50	Current and future approaches in PKU treatment *Cary Harding, USA*
16:50 | 17:20	Micronutrients – role in IEM treatment *Júlio César Rocha, PT*
17:20 | 18:00	**Session V - General Oral communications**
18:00 | 19:30	Thematic Meeting - PKU Breaking the Dogma - (Programme page 11)
20:30 | 21:45	Symposium dinner

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09:00 | 10:30	**Session VI - The acetyl-CoA cross**
09:00 | 09:30	Pyruvate dehydrogenase complex deficiency in Portugal: the clinical, metabolic, and mutational landscapes and its peculiar characteristics *Hana Pavlu-Pereira, PT*
09:30 | 10:00	Ketotic glycogen storage diseases (GSD): current knowledge, management, and monitoring *Ulrike Steuerwald, DE*
10:00 | 10:30	How to find your way through metabolic disorder in patients with rhabdomyolysis *Pascal Laforêt, FR*
10:30 | 11:00	Coffe break
11:00 | 13:00	**Session VII - Lysosomal Disorders**
11:00 | 11:30	A new face in the diagnosis and monitoring of lisosomal disorders *Johannes Aerts, NL*
11:30 | 12:00	Gene therapy for lysosomal storage disorders: advances, challenges and perspectives. *Maria José Castro, ES*
12:00 | 12:30	Immunomodulation and ERTs *Simon Jones, UK*
12:30 | 13:00	Oral Communications to the table
13:00 | 14:00	Lunch / Poster view
14:00 | 15:30	**Session VIII - The ammonia roundabout**
14:00 | 14:30	New insights into the pathophysiology and molecular basis of UCD's *Johannes Haberle, CH*
14:30 | 15:00	Potential role of gene therapy for OTC deficiency *Alvaro Hermida, ES*
15:00 | 15:30	A novel small molecule approach for the treatment of propionic and methylmalonic acidemias *Marshall Summar, USA*
15:30 | 16:00	Coffee break
16:00 | 17:30	**Session IX - Down Town Mitochondria**
16:00 | 16:30	Mitochondrial plasticity - role in muscle pathology *Rita Ferreira, PT*
16:30 | 17:00	Renal involvemente in Mitochondrial Disorders *Margarida Coelho, PT*
17:00 | 17:30	The diagnosis of mitochondrial disease using multi-omic methodologies *Charlotte Alston, UK*
17:30 | 17:45	**Bolsa SPDM Dr Aguinaldo Cabral 2019** *Sandra Alves, PT*
17:45	**Awards / Closing remarks**

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ORAL COMMUNICATIONS

12h30 – 13h00 | Session II – **Newborn Screening for IEM**

OC 01	When the newborn screening prevents ketoacidosis, *J Tenete, Vila Nova de Gaia, PT*
OC 02	Trifunctional protein deficiency – case report of neonatal onset, *M Rodrigues, Porto, PT*
OC 03	Carnitine uptake deficiency in asymptomatic patients – the importance of 5' untranslated region (UTR) of SLC22A5, *H Santos, Vila Nova de Gaia, PT*

15h30 - 15h50 | Session III – **From CbL to Hcy**

OC 04	A Case Series of Paediatric Classical Homocystinuria – nutritional status, dietary intake and metabolic profile, *RS Loureiro, Lisboa, PT*
OC 05	New predictives for low vitamin B12 in newborns, *P Lipari Pinto, Lisboa, PT*

17h20 | 18h00 | Session V – **General Oral Communications**

OC 09	Long-term, sustained efficacy and safety results from a phase 1/2 clinical trial of an AAV8-mediated liver-directed gene therapy in adults with glycogen storage disease type Ia, *Maria-Luz C Pico, Santiago de Compostela, SP*
OC 10	AAV8 Gene Therapy as a Potential Treatment in Adults with Late-Onset Ornithine Transcarbamylase (OTC) Deficiency: Updated Results From a Phase 1/2 Clinical Trial, *Maria-Luz C Pico, Santiago de Compostela, SP*
OC 11	Genotypic and phenotypic features of the mothers of children with mtDNA-associated Leigh syndrome, *CA Soares, Porto, PT*
OC 12	New causes of persistent or recurrent 3-methylglutaconic aciduria – expanding the differential diagnosis of secondary 3-MGA-uria, *F Freitas, Porto, PT*

10TH SEPTEMBER 2021

ORAL COMMUNICATIONS

12h30 - 13h00 | Session VII – **Lysosomal Disorders**

OC 06	Targeted RNA-based therapies for Mucopolysaccharidosis, *JI Santos, Porto, PT*
OC 07	Functional characterization of five novel mutations found in patients with suspicion of Fabry disease, *P Varela-Calais, São Paulo, BR*
OC 08	NIEMANN PICK Type C – 26 years of diagnosis in a Portuguese Reference Center, *AM Capela, Porto, PT*