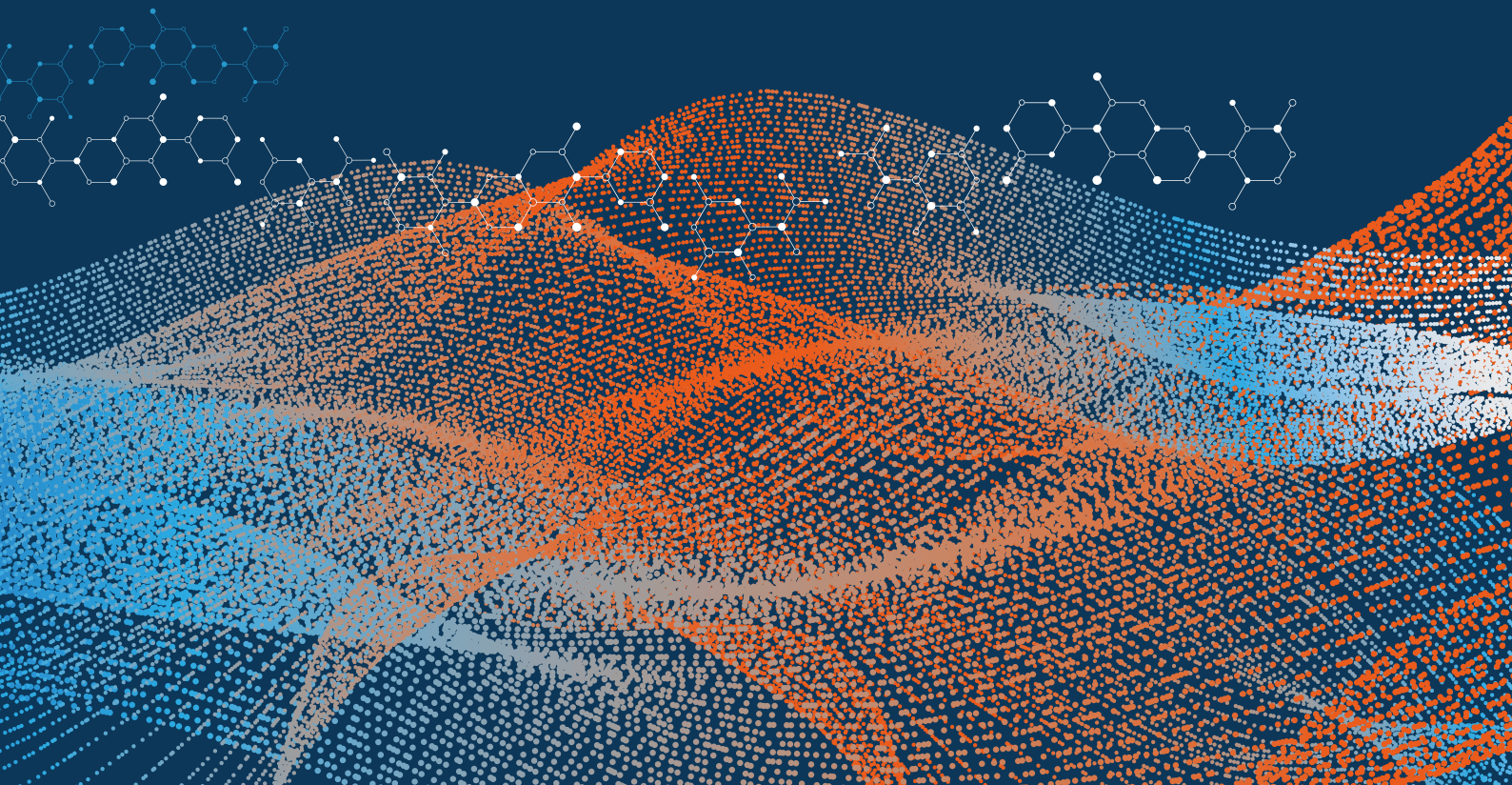
PROGRAM



SCIENTIFIC PROGRAM

MAY, 4TH

16:00	SPDM groups meeting
17:00	SPDM Nutrition Group Meeting

MAY, 5TH

09:00	Symposium Opening – Welcome Address on behalf of the SPDM *Patricia Janeiro – Symposium Chairperson*
	SESSION I – INBORN ERRORS OF METABOLITE REPAIR: A NEW CHALLENGE *Chairperson - Patrícia Janeiro, Lisboa, PT / Dulce Quelhas, Porto, PT*
09:15	Inborn errors of metabolite repair: insights in disease pathophysiology and treatment *Maria Veiga da Cunha, Brussels, Belgium*
09:40	Efficacy and safety of empagliflozin in Glycogen Storage Disease Type IB: Data from 112 Patients *Saskia Wortmann, Salzburg, Austria*
10:05	L-2-Hydroxyglutaric aciduria: an inborn error of metabolism repair *Eduard Struijs, Amsterdam, Netherlands*
10:30	Metabolic repair: a new approach to the treatment of abnormal glycosylation *Eva Morava, Minnesota, USA*
10:55	Coffee break
	SESSION II – FRONTIERS OF IEM: INTRACELLULAR CROSSTALK (PART A) *Chairperson – Hugo Rocha, Porto, PT / Luísa Diogo, Coimbra, PT*
11:30	Peroxisomal and fatty acid metabolism: intracellular interactions *Sander Houten, New York, USA*
11:55	Inborn Errors of the malate-aspartate shuttle *Nanda Verhoeven-Duif, Utrecht, Netherlands*
12:20	Different lipid signature in fibroblasts of long-chain fatty acid oxidation disorders *Sara Tucci, Freiburg, Germany*
12:45	Lunch
	SESSION II – FRONTIERS OF IEM: INTRACELLULAR CROSSTALK (PART B) *Chairperson – Margarida Silva, Lisboa PT / Daniel Gomes, Lisboa, PT*
14:00	Chemical individuality in T cells: a Garrodian view in immunometabolism *Peter J. McGuire, Bethesda, USA*
14:25	Mitochondrial redox regulation in hereditary cardiomyopathies *Christoph MaacK, Vurzburgo, Germany*
14:50	Mitochondrial dysfunction and 3-merthylglutaconic aciduria: a heterogeneous group of syndromes *Frederic Tort, Barcelona, Spain*
15:15	Mitochondrial disease in adults: recent advances and future promise *Yi Ng, Newcastle upon Tyne, UK*
15:40	Coffee break
16:00	**SESSION III - ORAL COMMUNICATIONS** *Chairperson – Anabela Oliveira, Lisboa PT / Manuela Grazina, Coimbra, PT*
17:00	End of the Session
	Poster View
18:30	Departure to Dinner

SCIENTIFIC PROGRAM

MAY, 6TH

09:00	**SESSION IV – SHORT ORAL COMMUNICATIONS** *Chairperson – Esmeralda Rodrigues, Porto, PT / Francisca Coutinho, Porto, PT*
09:50	**SESSION V - THE CLINICAL SPECTRUM OF IEM: ORGAN AS A CLUE TO THE DIAGNOSIS** *Chairperson – João Durães, Coimbra, PT / Anabela Bandeira, Porto, PT*
	Disorders of cellular trafficking: mechanisms and symptoms *Angels Garcia-Cazorla, Barcelona, Spain*
10:15	Neuronal migration defects related to IEM *Stephanie Grunewald, London, UK*
10:40	Coffee break
11:10	The skin as a clue for the diagnosis of inherited metabolic disorders *Carlo Dionisi-Vici, Rome, Italy*
11:35	Muscle manifestations of IEM *Mirjam Langeveld, Amsterdam, Netherlands*
12:00	Kidney manifestations of Lisossomal diseases *Patrício Aguiar, Lisboa, Portugal*
12:25	Eye manifestations of IEM: clues to diagnosis *Ana Cláudia Fonseca, Lisboa, Portugal*
12:50	Lunch /Poster View
	SESSION VI – TREATMENT MANAGEMENT IN IEM: DIETETIC AND PHARMACOLOGIC *Chairperson – Maurizio Scarpa, Udine, IT / Ana Paula Leandro, Lisboa, PT*
14:15	Novel RNAi therapeutics for Primary Hyperoxaluria *Sander Garrelfs, Amsterdam, Netherlands*
14:40	Nutricional care in lysosomal storage disorders *Francesca Carubbi, Modena, Italy*
15:05	Metabolic approaches for the treatment and prevention of epilepsy *Devle Boison, New Jersey, USA*
15:30	Intravenous enzyme replacement therapy in mucopolisacharidoses: clinical effectiveness and limitations *Rossella Parini, Milan, Italy*
15:55	Coffee Break
16:20	**SESSION VII – ORAL COMMUNICATIONS** *Chairperson – Ana Cristina Ferreira, Lisboa, PT / Teresa Campos, Porto, PT*
17:30	Awards and Final Remarks
17:45	End of the Symposium

ORAL COMMUNICATIONS

THURSDAY, 5TH MAY

16h00 – 17h00 | Session III

OC 01	Mitochondria dysfunction and potential riboflavin correction in mild MADD patients-derived fibroblasts
OC 02	Impaired FAD incorporation: an additional factor contributing to the pathogenesis of medium-chain acyl CoA dehydrogenase deficiency
OC 03	Genetic testing in LHON: classic approaches vs. a novel 24h test for top-3 variants
OC 04	Response to sapropterin dihydrochloride (Kuvan®) in a series of 17 patients with phenylketonuria
OC 05	Successful thioacetylation of human phenylalanine hydroxylase: towards enzymosome formulation
OC 06	Full mtDNA variation in a cohort of mitochondrial disorders suspected patients

FRIDAY, 6TH MAY

16h20 – 17h30 | Session VII

OC 07	Classic Galactosemia: how genotype affects biochemical phenotype and long-term outcome
OC 08	Congenital disorders of glycosylation and their impact on the endocrine system: a retrospective review
OC 09	3-OH quinoline derivatives modulate the aggregation of the p.G46S phenylalanine hydroxylase variant and rescues enzyme function
OC 10	Maple Syrup Urine Disease. The metabolic effect of BCAA and BCKA in vivo and in vitro studies
OC 11	Three patients with alpha-mannosidosis: clinical aspects and therapeutic options
OC 12	An Underestimated cause of Myopathy: A case series of Mcardle disease
OC 13	Can the X tell how much disease you can get?

SHORT ORAL COMMUNICATIONS

FRIDAY, 6TH MAY

09h00 – 09h50 - Session IV

SOC 01	Partial ornithine transcarbamylase deficiency in three generations with variable phenotype in males
SOC 02	Effect of the COVID-19 pandemic on body weight and body composition of adults with phenylketonuria
SOC 03	Biochemical and biophysical tools to study mitochondrial aminoacyl-tRNA synthetase-related neurological diseases
SOC 04	ODIMET®: Pionering online Tool for dietary management of Inborn Errors of Metabolism and its usefulness for Tele-Health
SOC 05	Look and take care for gangliosidosis
SOC 06	Sapropterin dihydrochloride (BH4) loading test: genotype-phenotype prediction of BH4 responsiveness in a cohort of phenylketonuric patients
SOC 07	Determination of orotic acid using stable isotope dilution GC-MS to the elucidation of urea cycle function and mechanisms of hyperammonemia
SOC 08	Evaluation of endoplasmic reticulum (ER) stress-mediated apoptosis in LHON patient primary cells
SOC 09	Clinical, Biochemical and Molecular Features of a Cohort of 10 Patients with Congenital Disorders of Glycosylation in a Metabolic Reference Center