SOCIEDADE PORTUGUESA
DE DOENÇAS METABÓLICAS

"Unlimited boundaries in inborn errors of Metabolism"

May 4th, 5th, 6th
MH Peniche Hotel, Peniche, Portugal
Hybrid Symposium

Organizing committee:

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Scientific committee:

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Instituto Nacional de Saúde Doutor Ricardo Jorge, Porto

Patrícia Janeiro – *SPDM Vice-president*

Centro Hospitalar de Lisboa Norte, Lisboa

May, 4th

16:00 SPDM groups meeting

17:00 SPDM Nutrition Group Meeting

May, 5th

9:00 Symposium Opening – Welcome Address on behalf of the SPDM

Patricia Janeiro – Symposium Chairperson

Session I – Inborn Errors of Metabolite Repair: a new challenge

09:15 Inborn errors of metabolite repair: insights in disease pathophysiology and treatment

Maria Veiga da Cunha, Brussels, Belgium

09:40 A story of metabolite-repair in glycolysis: neutropenia and neutrophil dysfunction in glycogen storage disease type Ib/a

Saskia Wortmann, Salzburg, Austria

10:05 L-2-Hydroxyglutaric aciduria: an inborn error of metabolism repair

Edward A. Struys, Amsterdam, Netherlands

10:30 Metabolite repair enzymes as targets for new therapies in CDG

Eva Morava, Minnesota, USA

10:55 **Coffee Break**

Session II – Frontiers of IEM: intracellular crosstalk (Part A)

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)
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**
17:00	**End of the Session**
	Poster View
18:30	**Departure to Dinner**

May, 6th

09:00	**Session IV – Short Oral Communications**
	Session V - The clinical spectrum of IEM: organ as a clue to the diagnosis
09:50	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
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**
17:30	Awards and Final Remarks
17:45	End of the Symposium