SOCIEDADE PORTUGUESA DE DOENÇAS METABÓLICAS

"Unlimited boundaries in inborn errors of Metabolism"

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

May, 4th

16:00	SPDM groups meeting
17:00	SPDM Nutrition Group Meeting
18:00	Departure to Visit Óbidos

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 *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	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)
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**

SOCIEDADE PORTUGUESA DE DOENÇAS METABÓLICAS

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

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
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