

“Unlimited boundaries in inborn errors of Metabolism”

May 4th, 5th, 6th

MH Peniche Hotel, Peniche, Portugal

Hybrid Symposium

May, 4th

- 17:00 SPDM groups meeting
- 18:00 SPDM Nutrition Group Meeting
- 19:00 Welcome drink

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, Vurzburg, 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 Lysosomal 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 Nutritional 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 mucopolisacaridoses: 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