

## “Linking metabolic signature to IEM treatment”

September 8<sup>th</sup>, 9<sup>th</sup>, 10<sup>th</sup>

Lux Fátima Park Hotel

Hybrid Symposium

### Organizing committee:

**Hugo Rocha - Symposium Chairperson**

*Instituto Nacional de Saúde Doutor Ricardo Jorge, Porto*

**Dulce Quelhas**

*Centro Hospitalar e Universitário do Porto, Porto*

**Esmeralda Martins**

*Centro Hospitalar e Universitário do Porto, Porto*

**Esmeralda Rodrigues**

*Centro Hospitalar Universitário de São João, Porto*

**Helena Santos**

*Centro Hospitalar de Vila Nova de Gaia/Espinho, Vila Nova Gaia*

**Manuela Ferreira de Almeida**

*Centro Hospitalar e Universitário do Porto, Porto*

**Paulo Castro Chaves**

*Centro Hospitalar Universitário de São João, Porto*

### Scientific committee:

**Daniel Gomes – SPDM President**

*Centro Hospitalar de Lisboa Norte, Lisboa*

**Patrícia Janeiro – SPDM Vice-president**

*Centro Hospitalar de Lisboa Norte, Lisboa*

**Dulce Quelhas – SPDM Board**

*Centro Hospitalar e Universitário do Porto, Porto*

**Esmeralda Martins – SPDM Board**

*Centro Hospitalar e Universitário do Porto, Porto*

**Hugo Rocha – SPDM Board**

*Instituto Nacional de Saúde Doutor Ricardo Jorge, Porto*

## September, 8th

**17:45 | 18:30**

SPDM Nutrition 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*

**10:00 | 10:30**

Revisiting IEM's classification.

*Carlos Ferreira*

**10:30 | 11:00**

**Coffee break**

**11:00 | 12:30**

**Session II - Newborn Screening for IEM**

**11:00 | 11:30**

Newborn Screening in Europe and initiatives to develop programmes in low - and middle-income countries.

*Jim Bonham*

**11:30 | 12:00**

New tests and future directions in the NBS for IEM.

*Giancarlo LaMarca*

**12:00 | 12:30**

Treatment follow-up of screened disorders.

*Paula Garcia*

**12:30 | 13:00**

**Oral Communications to the table**

**13:00 | 14:00**

**Lunch / Poster view**

**14:00 | 15:30**

**Session III - From CbL to Hcy**

**14:00 | 14:30**

Remethylation disorders: from cobalamin absorption, intracellular pathways to MTHFR deficiency.

*Matthias Baumgartner*

**14:30 | 15:00**

Inherited disorders of transsulfuration: clinical presentation, diagnosis, and recent advances in therapy.

*Viktor Kozich*

**15:00 | 15:30**

The transsulfuration pathway at the crossroads between homocysteine and hydrogen sulfide metabolism: (dys)regulation and (patho)physiological implications.

*João Vicente*

**15:30 | 16:00**

**Oral communications to the table**

**16:00 | 16:30**

**Coffee break**

**16:30 | 17:30**

**Session IV - IEM Nutritional and Pharmacological treatment**

**16:30 | 17:00**

Current and future approaches in PKU treatment.

*Cary Harding*

**17:00 | 17:30**

Micronutrients – role in IEM treatment.

*Júlio César Rocha*

**17:30 | 18:00**

**Session V - General Oral communications**

**20:30 | 21:45**

**Symposium dinner**

## September, 10<sup>th</sup>

**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*

**09:30 | 10:00**

Ketotic glycogen storage diseases (GSD): current knowledge, management, and monitoring.

*Ulrike Steuerwald*

**10:00 | 10:30**

How to find your way through metabolic disorder in patients with rhabdomyolysis.

*Pascal Laforêt*

**10:30 | 11:00**

**Coffe break**

**11:00 | 12:30**

**Session VII - Lysosomal Disorders**

**11:00 | 11:30**

A new face in the diagnosis and monitoring of lysosomal disorders.

*Johannes Aerts*

**11:30 | 12:00**

Gene therapy for lysosomal storage disorders: advances, challenges, and perspectives.

*Maria José Castro*

**12:00 | 12:30**

Immunomodulation and ERTs.

*Simon Jones*

**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*

**14:30 | 15:00**

Potential role of gene therapy for OTC deficiency.

*Alvaro Hermida*

**15:00 | 15:30**

A novel small molecule approach for the treatment of propionic and methylmalonic acidemias.

*Marshall Summar*

**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*

**16:30 | 17:00**

Renal involvement in Mitochondrial Disorders.

*Margarida Coelho*

**17:00 | 17:30**

The diagnosis of mitochondrial disease using multi-omic methodologies.

*Charlotte Alston*

**17:30 | 17:45**

**Bolsa SPDM Dr Aguinaldo Cabral 2019**

**17:45 | 18:00**

**Bolsa SPDM / Recordati Rare Diseases 2019**

**18:00**

**Awards / Closing remarks**