

SAVE THE DATE

# 17<sup>TH</sup> INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

SEPTEMBER, 8 - 10 | HOTEL LUX - FÁTIMA, PORTUGAL

[WWW.SIMPOSIO.SPDM.ORG.PT](http://WWW.SIMPOSIO.SPDM.ORG.PT)

HYBRID EVENT



LINKING METABOLIC SIGNATURE  
TO IEM TREATMENT

## 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*

Registration and Abstracts Submission opened, please check it on  
[www.simposio.spdm.org.pt](http://www.simposio.spdm.org.pt)

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## SCIENTIFIC PROGRAM

### September 8th

17:45 - SPDM Nutrition meeting

19:00 - Welcome drink

### September 9th

08:30 - Registration Opening

09:15 - Symposium Opening

#### Session I - IEM's mapping

09:30- MetabERN - where are we now?, **Maurizio Scarpa**

10:00 - Revisiting IEM's classification, **Carlos Ferreira**

10:30- Coffee Break

#### Session II - Newborn Screening

11:00 - Newborn Screening in Europe and initiatives to develop programmes in low- and middle-income countries, **Jim Bonham**

11:30 - New tests and future directions in the NBS for IEM, **Giancarlo LaMarca**

12:00 - Treatment follow-up of screened disorders, **Paula Garcia**

12:30 - Oral Communications to the table

13:00 - Lunch / poster view

#### Session III - From CbL to Hcy

14:00 - Remethylation disorders: from cobalamin absorption, intracellular pathways to MTHFR deficiency, **Matthias Baumgartner**

14:30 - Inherited disorders of transsulfuration: clinical presentation, diagnosis and recent advances in therapy?, **Viktor Kozich**

15:00- The transsulfuration pathway at the crossroads between homocysteine and hydrogen sulfide metabolism: (dys)regulation and (patho)physiological implications, **João Vicente**

15:30 - Oral Communications to the table

16:00 - Coffee Break

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

16:30 - Current and future approaches in PKU treatment, **Cary Harding**

17:00 - Micronutrients – role in IEM treatment, **Julio Rocha**

17:30 - Session V - General Oral Communications

20h:30 - Symposium dinner

### September 10th

#### Session VI - The acetyl-CoA cross

09:00 - Pyruvate dehydrogenase complex deficiency in Portugal: the clinical, metabolic and mutational landscapes and its peculiar characteristics, **Hana Pavlu-Pereira**

09:30 - Ketotic glycogen storage diseases (GSD): current knowledge, management and monitoring, **Ulrike Steuerwald**

10:00 - How to find your way through metabolic disorder in patients with rhabdomyolysis, **Pascal Laforêt**

10:30 - Coffee Break

#### Session VII - Lysosomal Disorders

11:00 - A new face in the diagnosis and monitoring of lysosomal disorders, **Johannes Aert**

11:30 - Gene therapy for lysosomal storage disorders: advances, challenges and perspectives, **Maria José Castro**

12:00 - Immunomodulation and ERTs, **Simon Jones**

12:30 - Oral Communications to the table

13:00 - Lunch / poster view

#### Session VIII - The ammonia roundabout

14:00 - New insights into the pathophysiology and molecular basis of UCD's, **Johannes Haberle**

14:30 - Potential role of gene therapy for OTC deficiency, **Alvaro Hermida**

15:00 - A novel small molecule approach for the treatment of propionic and methylmalonic acidemias, **Marshall Summar**

15:30 - Coffee Break

#### Session IX - Down Town Mitochondria

16:00 - Mitochondrial plasticity - role in muscle pathology, **Rita Ferreira**

16:30 - Renal involvement in Mitochondrial Disorders **Margarida Coelho**

17:00 - The diagnosis of mitochondrial disease using multi-omic methodologies, **Charlotte Alston**

17:30 - Bolsa SPDM Dr Aguinaldo Cabral 2019

17:45 - Bolsa SPDM / Recordati Rare Diseases 2019

18:00 - Awards / closing remarks