

SAVE THE DATE

17TH INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

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

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HYBRID EVENT



LINKING METABOLIC SIGNATURE
TO IEM TREATMENT



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SOCIEDADE PORTUGUESA
DE DOENÇAS METABÓLICAS

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

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CIENTIFIC 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

Chairpersons: Luísa Diogo, Coimbra, PT | Dulce Quelhas, Porto, PT

09:30 - MetabERN - where are we now?

Maurizio Scarpa, IT

10:00 - Revisiting IEM's classification

Carlos Ferreira, USA

10:30 - Coffee Break

Session II - Newborn Screening

Chairpersons: Hugo Rocha, Porto, PT | Ana Gaspar, Lisboa, PT

11:00 - NBS for IEM - current status and efforts towards dissemination

Jim Bonham, UK

11:30 - New tests and future directions in the NBS for IEM

Giancarlo LaMarca, IT

12:00 - Treatment follow-up of screened disorders

Paula Garcia, PT

12:30 - Oral Communications to the table

13:00 - Lunch / poster view

Session III - "From CbL to Hcy"

Chairpersons: Isabel Tavares de Almeida, Lisboa, PT | Patrícia Janeiro, Lisboa, PT

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

Matthias Baumgartner, CN

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

Viktor Kozich, CZ

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

João Vicente, PT

15:30 - Oral Communications to the table

16:00 - Coffee Break

Session IV - IEM Nutritional and Pharmacological treatment

Chairpersons: Paula Leandro, Lisboa, PT | Sandra Mexia, Lisboa, PT

16:30 - Current and future approaches in PKU treatment

Cary Harding, USA

17:00 - Micronutrients – role in IEM treatment

Julio Rocha, PT

Session V

Chairpersons: Helena Santos, V.N.Gaia, PT | Ana Cristina Ferreira, Lisboa, PT

17:30 - General Oral Communications

20:30 - Symposium Dinner

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September 10th

Session VI - The acetyl-CoA cross

Chairpersons: Domingo Lamuño, SP | M. Teresa Cardoso, Porto, PT

- 09:00** - Pyruvate dehydrogenase complex deficiency in Portugal: the clinical, metabolic and mutational landscapes and its peculiar characteristics
Hana Pavlu-Pereira, PT
- 09:30** - Ketotic glycogen storage diseases (GSD): current knowledge, management and monitoring
Ulrike Steuerwald, DE
- 10:00** - How to find your way through metabolic disorder in patients with rhabdomyolysis
Pascal Laforêt, FR
- 10:30** - Coffee Break

Session VII- Lysosomal Disorders

Chairpersons: Elisa Leão Teles, Porto, PT | Paulo Gaspar, Porto, PT

- 11:00**- A new face in the diagnosis and monitoring of lysosomal disorders
Johannes Aerts, NL
- 11:30** - Gene therapy for lysosomal storage disorders: advances, challenges and perspectives
Maria José Castro, SP
- 12:00** - Immunomodulation and ERTs
Simon Jones, UK
- 12:30** - **Oral Communications to the table**
- 13:00** - Lunch / poster view

Session VIII - The ammonia roundabout

Chairpersons: Margarida Silva, Lisboa, PT | Teresa Almeida Campos, Porto, PT

- 14:00** - New insights into the pathophysiology and molecular basis of UCD's
Johannes Haberle, CN
- 14:30** - Potential role of gene therapy for OTC deficiency
Alvaro Hermida, SP
- 15:00** - A novel small molecule approach for the treatment of propionic and methylmalonic acidemias
Marshall Summar, USA
- 15:30** - Coffee Break

Session IX - Down Town Mitochondria

Chairpersons: Esmeralda Martins, Porto, PT | Célia Nogueira, Porto, PT

- 16:00** - Mitochondrial plasticity - role in muscle pathology, Rita Ferreira, PT
- 16:30** - Renal involvement in Mitochondrial Disorders
Margarida Coelho, PT
- 17:00** - The diagnosis of mitochondrial disease using multi-omic methodologies, Charlotte Alston, UK
- 17:30** - "Bolsa SPDM Dr Aguinaldo Cabral 2019"
- 17:45** - "Bolsa SPDM / Recordati Rare Diseases 2019"
- 18:00** - Awards / closing remarks