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Local Organizing Committee

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**General information & online registration:
Symposium and/or Satellite Meeting**
(Registration in the Satellite Meeting is free, but required)
www.asic.pt - congressos@asic.pt

Oral Communications & Posters
deadline for submission: 26th February

Hotel reservation can be done through direct contact (www.asic.pt). Let them know that you are attending the SPDM Symposium and/ or the Satellite Meeting.

Delegate Registration
SPDM Members 125€
Non-SPDM Members 150€
Students 100€
Symposium dinner included;
- registration required -

Secretariat

asic

associação de saúde infantil de coimbra

Online registration in:
www.asic.pt
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12th International Symposium SPDM

Sociedade Portuguesa de Doenças Metabólicas

17th - 18th March 2016

COIMBRA, HOTEL VILA GALÉ

Satellite Meeting 19th March

MPS: from paediatric to adult care



Thursday, 17 March

- 08:00 Registration Opening
- 09:00 Symposium Opening - Welcome Address on behalf of the SPDM
Local organization representatives
- Session I ❖ Unraveling disease mechanisms in Inborn Errors of Metabolism (IEM)**
- 09:10 Mechanisms of cell quality control in inherited metabolic diseases, *Henrique Girão, Coimbra, PT*
- 09:30 iPSc disease models of Gaucher Disease and Tay-Sachs, *Gustavo Tiscornia, Faro, PT*
- 09:50 LSD - Implications of glycosphingolipid storage in immunity, *Fátima Macedo, Porto, PT*
- 10:10 Dissecting the metabolic crosstalk between peroxisomes and mitochondria: a CRISPR-Cas9 approach, *Sara Violante, New York, USA*
- 10:30 Noninvasive methods for quantifying hepatic intermediary metabolic fluxes in humans, *John Jones, Coimbra, PT*
- 10:50 Panel discussion
- 11:10 Coffee Break / Poster view
- Session II ❖ Glucose homeostasis and IEM**
- 11:40 Glucose homeostasis: are children different from adults? *Hidde Huidekoper, Amsterdam, NL*
- 12:00 GSD are a cause of ketotic hypoglycaemia, *David Weinstein, Gainesville - FL, USA*
- 12:20 Impaired amino acid metabolism contributes to fasting-induced hypoglycemia in fatty acid oxidation defects, *Sander Housten, New York, USA*
- 12:40 Panel discussion
- 13:00 Lunch/ Poster view
- Session III ❖ Obesity in IEM**
- 14:00 Obesity - an overview of pathogenic mechanisms, *Paula Freitas, Porto, PT*
- 14:20 Prevention of hypoglycaemia and obesity in GSD type 1, *David Weinstein, Gainesville - FL, USA*
- 14:40 Dietary management of FAOD patients - carbohydrates: friends or foes? *Marjorie Dixon, London, UK*
- 15:00 Protein restricted diets and obesity, *Júlio Rocha, Porto, PT*
- 15:20 Panel discussion
- 15:40 Coffee Break / Poster view
- Session IV ❖ Oral communications I**
- 16:10 Selected oral communications
- 17:15 **Assembleia Geral da SPDM**
- 19:45 Departure from the hotel
- 20:00 Symposium Dinner
- 22:30 Return to the Hotel

Friday, 18 March

- Session V ❖ Short oral poster presentations**
- 08:15 Selected oral poster presentations
- Session VI ❖ IEM - treatment advances and setbacks**
- 09:20 Advances in the treatment of branched chain organic acidurias *Alberto Burlina, Padova, IT*
- 09:40 Fabry disease treatment -«state of the art», *João Paulo Oliveira, Porto, PT*
- 10:00 Treatment strategies for mitochondrial disorders, *Julien Beyrath, Nijmegen, NL*
- 10:20 Complementary and alternative medicine use in IEM, *Saskia Wortmann, Salzburg, Austria*
- 10:40 Lessons from the use of sapropterin in PKU , *Anabela Bandeira, Porto, Pt*
- 11:00 Panel discussion
- 11:20 Coffee Break / Poster view
- Session VII ❖ From Garrod to whole exome sequencing era – the boundaries of IEM**
- 11:50 The evolving concept of IEM - place in the International Classification of Diseases, *Shamima Rahman, London, UK*
- 12:10 Enzyme complexity in intermediary metabolism, *Emile van Schaftingen, Louvain, Belgium*
- 12:30 Brain lipids - much more than a structural role, *Catarina Oliveira, Coimbra, PT*
- 12:50 Panel discussion
- 13:00 Lunch / Poster view
- Session VIII ❖ IEM of brain membrane lipids and proteins**
- 14:00 Treatable IEM due to membrane vitamin transporters deficiency – clinical and laboratorial approach, *Belén Pérez Dueno, Barcelona, SP*
- 14:20 Neurological presentations of IEM involving synthesis and remodeling of complex lipids, *Àngels Garcia-Carzola, Barcelona, SP*
- 14:40 Clinical and laboratorial approach to synaptic communication, *Sofia Duarte, Lisboa, PT*
- 15:00 Neuroinflammation in IEM , *Filipe Palavra, Coimbra, PT*
- 15:20 Panel discussion
- 15:40 Coffee Break / Poster view
- Session IX ❖ Oral communications II**
- 16:10 Selected oral communications
- 17:40 Awards / Closing Remarks, *Luísa Diogo, SPDM President*
- 18:00 **Assembleia Eleitoral da SPDM**