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## Local Organizing Committee

Paula Garcia  
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Letícia Ribeiro  
Luísa Diogo

## Scientific Committee

Luísa Diogo  
Isabel T. Almeida  
M. Teresa Cardoso  
Dulce Quelhas  
Patrícia Janeiro

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

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](http://www.asic.pt)  
+351 239 482 000 +351 961 466 788  
E-mail: [congressos@asic.pt](mailto:congressos@asic.pt)

Oral Communications & Posters  
deadline for submission: 28<sup>th</sup> February

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

# 12<sup>th</sup> International Symposium SPDM

Sociedade Portuguesa de Doenças Metabólicas

**17<sup>th</sup> - 18<sup>th</sup> March 2016**  
**COIMBRA, HOTEL VILA GALÉ**

**Satellite Meeting 19<sup>th</sup> 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)**  
**Chairpersons:** Manuela Grazina, Margarida Silva  
 09:10 Mechanisms of cell quality control in inherited metabolic diseases, Henrique Girão, Coimbra, PT  
 09:30 Neuroinflammation in IEM, Filipe Palavra, Coimbra, 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**  
**Chairpersons:** Paula Garcia, Hugo Rocha  
 11:40 Glucose homeostasis during fasting: lessons learned in and from IEMs, Hidde Huidekoper, Rotterdam, 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 hypoglycaemia in fatty acid oxidation defects, Sander Houten, New York, USA  
 12:40 Panel discussion  
 13:00 Lunch / Poster view  
**Session III ❖ Obesity in IEM**  
**Chairpersons:** Manuela Almeida, Hugo Clemente  
 14:00 Dietary management of FAOD patients - carbohydrates: friends or foes? Marjorie Dixon, London, UK  
 14:20 Obesity - an overview of pathogenic mechanisms, Paula Freitas, Porto, PT  
 14:40 Prevention of hypoglycaemia and obesity in GSD type 1, David Weinstein, Gainesville - FL, USA  
 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**  
**Chairpersons:** Cláudia Costa, Francisco Laranjeira  
 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**  
**Chairpersons:** Ana Marcão, Sónia Moreira  
 08:15 Selected oral poster presentations  
**Session VI ❖ IEM - treatment advances and setbacks**  
**Chairpersons:** Esmeralda Martins, Paula Leandro  
 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, AT  
 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**  
**Chairpersons:** Laura Vilarinho, Sérgio Sousa  
 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, BE  
 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**  
**Chairpersons:** Lígia Almeida, M. Carmo Macário  
 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, Àngeles Garcia-Cazorla, Barcelona, SP  
 14:40 Clinical and laboratorial approach to synaptic communication, Sofia Duarte, Lisboa, PT  
 15:00 Panel discussion  
 15:20 Coffee Break / Poster view  
**Session IX ❖ Oral communications II**  
**Chairpersons:** Ana Cristina Ferreira, Rita Castro  
 15:50 Selected oral communications  
 17:10 Awards / Closing Remarks, Luísa Diogo, SPDM President  
 17:30 Assembleia Eleitoral da SPDM