11th International Symposium
19 - 20 March 2015 - Palácio do Freixo - Porto

KEY DATES
Abstract Submission Opens: 20 January
Abstract Submission Deadline (close at 24:00): 27 February
Notification of Abstract Acceptance: 6th March
Full Information on: www.spdm.org.pt/events/simposio-anual-spdm/
### THURSDAY, 19 MARCH

**08:00** Registration Opening  
**08:50** Symposium Opening - Welcome Address on behalf of the SPDM  
- Local organization representatives

**Session I – Lysosome and related structures: extending boundaries**

- **09:00** Biogenesis, secretion and inter-cellular interactions of exosomes in health and disease  
  Graça Raposo | Paris, FR

- **09:30** Lysosome revisited – Connecting rare and common illnesses  
  Francisco Coutinho | Porto, PT

- **10:00** NGS in paediatric metabolic and neurodegenerative diseases expands the horizon  
  Ana Fernández-Marmiesse | Santiago de Compostela, ES

**10:30** Coffee Break

**Session II – Lysosomal Storage Disorders – new insights into treatment approaches**

- **11:00** Late breaking news on Lysosomal Storage Disorders treatment  
  Begoña Cachon Gonzalez | Cambridge, UK

- **11:30** Development of a U1 snRNA-adapted gene therapeutic strategy to correct 5' splicing defects in Lysosomal Storage Disorders  
  Liliana Mateu | Porto, PT

- **11:50** MPS VII trial treatment  
  Antonio González Monasterio | Sevilha, ES

- **12:20** Eliglustat: oral substrate reduction therapy for Gaucher disease type 1 – First results from a Portuguese cohort  
  Cristina Fraga | Ponta Delgada, PT

**12:50** Poster View / Lunch

**Session III – Inborn errors of metabolism and the adult**

- **13:50** Challenges of transferring patients from paediatric to adult care: A bridge too far?  
  Christine Lawry | London, UK

- **14:10** Planning and managing pregnancy in women with inherited metabolic disease  
  Elena Murphy | London, UK

- **14:40** Differential diagnosis in adult IMDs  
  Rafael Enrique de Salamanca | Madrid, ES

**15:10** Porphyria – rare disorder or under diagnosed?  
  To be announced

**15:40** Coffee Break

**Session IV – Oral Communications**

**16:00** Beginning of the Session  
**17:30** End of the Session

**17:45** SPDM Assembleia Geral

**19:30** Departure from the hotel

**20:00** Symposium Dinner

**22:30** Return to the Hotel

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### FRIDAY, 20 MARCH

**Session V – Oral Communications**

- **09:00** Beginning of the Session

- **10:30** End of the Session

- **10:30** Coffee Break

**Session VI – Mitochondria and fatty-acid oxidation**

- **10:50** New insights into the diagnosis of mitochondrial disorders - from clinic to bench and back again  
  Eva Morava | New Orleans, USA

- **11:20** Biosynthesis and remodeling of phospholipids: role in mitochondrial fusion and fission or signal transduction  
  Luisa Almeida | Porto, PT

- **11:40** Krebs cycle dysfunction and energy failure: novel insights into mitochondrial regulation  
  Manuela Grosso | Coimbra, PT

- **12:00** Proteomics approach to fatty-acid oxidation defects  
  Hugo Rocha | Porto, PT

- **12:20** Management of fatty-acid oxidation defects: data from a base daily experience  
  Patricia Jimeno | Lisboa, PT

- **12:40** Treatment of mitochondrial long chain fatty acid: what we know and where to go  
  Sara Tucci | Freiburg, DE

**13:00** Poster View / Lunch

**Session VII – Inborn errors of metabolism: nutritional and pharmacological treatment – Present and future**

- **14:00** Micronutrients status - too much is less  
  Sharon Evans | Birmingham, UK

- **14:30** Glycomacropeptide - Advances in the nutritional treatment of phenylketonuria  
  Diny Niy | Madison, USA

- **15:00** Sapropterin now and again – lessons from nutritional experience in PKU  
  João César Rocha | Porto, PT

**15:30** Coffee Break

**Session VIII – Congenital disorders of glycosylation**

- **15:50** CDG – Can we still be surprised?  
  Eva Morava | New Orleans, USA

- **16:20** Glycosylation and immunological defects in CDG  
  Ana Fernández-Liébana | Lisboa, PT

- **16:40** How metabolomics can contribute to our understanding of CDG pathophysiological mechanisms  
  Antonio Pineda-Lucena | Valencia, ES

**16:50** Coffee Break

**Session IX – Short Oral posters Presentations**

- **17:00** Beginning of the Session

- **18:00** End of the Session

**18:00** Awards / Closing Remarks  
- Luísa Diogo, SPDM President

**18:15** End

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**I Encontro Nacional de Nutrição**

- **18:30** - 20:00 Doenças Hereditárias do Metabolismo