



11th International Symposium

19 - 20 March 2015 - Palácio do Freixo - Porto

KEY DATES

Abstract Submission Opens 28 January

Abstract Submission Deadline (close at 24:00) 27 February

Notification of Abstract Acceptance 04 March

Full information on: www.spdm.org.pt/eventos/simposio-anual-spdm/



	19e 6 de Março	19de 6 de Março
Membro SPDM	125€	150€
Não-Membro	150€	175€
Estudantes	100€	125€

A inscrição no Simpósio Internacional inclui: Documentação, coffee-breaks, almoços e jantar do Simpósio
Presença no jantar do Simpósio (a convite do organizador)



Secretariat



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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**
Francisca Coutinho | Porto, PT
10:00 **NGS in paediatric metabolic and neurodegenerative diseases expands the horizon**
Ana Fernández-Marmisse | 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 Matos | Porto, PT
11:50 **MPS VII trial treatment**
Antonio Gonzalez Menezes | Sevilha, ES
12:20 **Egluostat: 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 Lavery | London, UK
14:10 **Planning and managing pregnancy in women with inherited metabolic disease**
Elaine Murphy | London, UK
14:40 **Diferential diagnosis in adult IMDs**
To be announced
15:10 **Porphyria – rare disorder or under diagnosed?**
Rafael Enriquez de Salamanca | Madrid, ES
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**

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**
Ligia Almeida | Porto, PT
11:40 **Krebs cycle dysfunction and energy failure: novel insights into mitochondrial regulation**
Manuela Grazina | 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 Janeiro | 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**
Denise Ney | Madison, USA
15:00 **Sapropterin now and again – lessons from nutritional experience in PKU**
Júlio 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**
Paula Videira | Lisboa, PT
16:40 **How metabolomics can contribute to our understanding of CDG pathophysiological mechanisms**
Antonio Pineda-Lucena | Valencia, ES

Session IX – Short Oral posters Presentations

- 17:00 **Beginning of the Session**
18:00 **End of the Session**
18:00 **Awards / Closing Remarks**
Luisa Diogo, SPDM President
18:15 **End**

I Encontro Nacional de Nutrição

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