



Pipos 6 de Morçol (Ne 6 de Março) 125€ 150€ 150€ 100€ 125€

A inscrição no Simpósio Internacional inclui: Documentação, coffee breaks, almoços e jantar do Simpósio

#### Secretariat



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diet care

genzyme

Membro SPDM Não-Membro

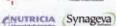






















19 - 20 March 2015 - Palácio do Freixo - Porto

# **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/



#### **THURSDAY. 19 MARCH**

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

## Session I - Lysosome and related structures: extending boundaries

- 09:00 Biogenesis, secretion and inter-cellular interactions of exosomes in health and disease
- 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-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 Lysossomal 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 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 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 Begining 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 Begining 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
  Lígia 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 Rochal 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\*\*

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#### **Session IX - Short Oral posters Presentations**

- 17:00 Begining of the Session
- 18:00 End of the Session
- 18:00 Awards / Closing Remarks

Luísa Diogo, SPDM President

18:15 **End** 

#### I Encontro Nacional de Nutrição

18:30 - 20:00 Doenças Hereditárias do Metabolismo