



Sociedade Portuguesa de Doenças Metabólicas

Annual Symposium 2019

15th International SPDM Symposium

14th – 16th March 2019

Hotel Vila Galé, Coimbra

“Old diseases, new treatments”

Programme

Thursday, 14th March

08:00 - **Registration Opening**

08:45 - **Symposium Opening** - Welcome Address on behalf of the SPDM

Local organization representatives

09:00 – 10:15 - Session I – The moving targets of expanded newborn screening for inborn errors of metabolism

Chairpersons: Helena Santos, Vila Nova de Gaia, PT / Laura Vilarinho, Porto, PT

09:00 - **The experience in the United Kingdom**

Rachel Carling | London, UK

09:20 - **Newborn Screening of Lysosomal Storage Disorders**

Alberto Burlina | Padova, IT

09:40 - **Expanded newborn screening in Portugal**

Ana Marcão | Porto, PT

10:00 – Discussion

10:15 – 10:30 - Coffee Break

10:30 – 11:45 - Session II – Politics and economics in the treatment of inborn errors of metabolism

Chairpersons: José Manuel Silva, Coimbra, PT / António Freire Gonçalves, Coimbra, PT

10:30 - **Economic perspective**

Nadim Habib | Lisboa, PT



10:50 - **Regulatory perspective**

Carlos Fontes Ribeiro | Coimbra, PT

11:10 - **Clinical perspective**

Teresa Cardoso | Porto, PT

11:30 - Discussion

11:45 – 13:30 - Session III – Inborn errors of metabolism I: an update

Chairpersons: Silvia Sequeira, Lisboa, PT / Isabel Rivera, Lisboa, PT

11: 45 - **Therapeutic goals in Gaucher Disease: the best definition of treatment response**

Deborah Elstein | Jerusalem, IL

12: 05 - **Neuronal Ceroid Lipofuscinoses: assessment and treatment**

Paul Gissen | London, UK

12:25 - **Expanding treatment options for Methylmalonic aciduria**

Stephanie Grünwald | London, UK

12: 45 - **CYP46A1 as a new therapeutic target in Niemann-Pick type C disease (2018 SPDM research grant winner)**

Elsa Rodrigues | Lisboa, PT

13:05 - Discussion

13:30 – 14:45 - Poster View / Lunch

14:45 – 16:00 - Session IV – Metabolic pathways and subcellular organelles: new connections

Chairpersons: Catarina R. Oliveira, Coimbra, PT / Isabel Santana, Coimbra, PT

14:45 - **Endoplasmic reticulum stress: new insights in inborn errors of metabolism**

Cláudia Pereira | Coimbra, PT

15:05 - **Hypothalamic dysfunction and neurodegeneration**

Cláudia Cavadas | Coimbra, PT

15:25 - **Intercellular communication in inborn errors of metabolism**

Henrique Girão | Coimbra, PT

15:45 - Discussion

16:00 – 16:30 - Coffee Break



16:30 – 18:00 - Session V – Selected Free Communications

Chairpersons: Esmeralda Rodrigues, Porto, PT / Patrícia Janeiro, Lisboa, PT

18:10 First SPDM National Treatment Protocol Group Meeting

18:10 Fifth SPDM Nutrition meeting (SPDM-GN)

Chairperson: Manuela Ferreira de Almeida, Porto, PT

19:30 Departure from the hotel

20:00 Symposium Dinner

Friday, 15th March

08:00 – 09:00 - Session VI – Selected Free Communications

Chairpersons: Anabela Oliveira, Lisboa, PT / Hugo Rocha, Porto, PT

09:00 – 10:15 - Session VII – Nutritional paradigms in inborn errors of metabolism

Chairpersons: Ana Faria, Coimbra, PT / Júlio Rocha, Porto, PT

09:00 - Micronutrients: speculations in inborn errors of metabolism

Peter Clayton | London, UK

09:20 - Nutritional Intervention after hepatic transplant in inborn errors of protein metabolism

Anne Daly | Birmingham, UK

09:40 - The effect of the nitrogen source on metabolism in Phenylketonuria (2018 SPDM/Orphan Europe research grant winner)

Maria João Pena | Porto, PT

10:00 - Discussion

10:20 – 10:40 - Coffee Break

10:40 – 11:45 - Session VIII – Mitochondrial diseases: from clinics to basic research and back

Chairpersons: Célia Noqueira, Porto, PT / Sandra Jacinto, Lisboa, PT

10:40 - The challenges in the clinical evaluation of mitochondrial diseases

Shamima Rahman | London, UK



11:00 - **Current role of functional mitochondrial study in the genomic era**

Manuela Grazina | Coimbra, PT

11:20 - **What is new in the treatment of mitochondrial disorders?**

Mirian Janssen | Nijmegen, NL

11:40 - Discussion

12:00 – 13:15 - Session IX – Selected Free Communications

Chairpersons: Ana Cristina Ferreira, Lisboa, PT / Hélder Esperto, Coimbra, PT

13:15 – 14:15 - Poster View / Lunch

14:15 – 15:10 - Session X – Inborn errors of metabolism II: an update

Chairpersons: Dulce Quelhas, Porto, PT / Anabela Bandeira, Porto, PT

14:15 - **Urea Cycle Disorders: what is old, what is new, what can we change?**

Amaya Bélanger-Quintana | Madrid, ES

14:35 - **Riboflavin-Responsive Disorders**

Manuel Schiff | Paris, FR

14:55 - Discussion

15:10 – 16:00 - Session XI – Treating inborn errors of metabolism: future directions

Chairpersons: Elisa Leão Teles, Porto, PT / Isabel Tavares de Almeida, Lisboa, PT

15:10 - **The development of new enzyme replacement therapies: the example of Hypophosphatasia**

Sérgio Sousa | Coimbra, PT

15:30 - **The possibilities of gene therapy for inborn errors of metabolism**

Paul Gissen | London, UK

15:50 - **Oral treatment for Fabry Disease: the evolution in chaperone therapies**

Patrício Aguiar | Lisboa, PT

16:10 - Discussion

16:30 – 16:50 Coffee Break



16:50 – 17:50 - Session XII – Selected Poster Communications

Chairpersons: Paula Leandro, Lisboa, PT / João Durães, Coimbra, PT

18:00 Awards / Closing Remarks

Teresa Cardoso, SPDM President

18:30 SPDM General Assembly