PROGRAMME

22ND INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

THE FUTURE OF INBORN ERRORS OF METABOLISM: DECODING COMPLEXITY, DELIVERING INNOVATION

HOTEL VILA GALÉ, COIMBRA

18th–20th March

18th-20th March

SPDM Sociedade Portuguesa de Doenças Metabólicas

ORGANIZING COMMITTEE'S PRESIDENT

João Durães, ULS Coimbra
Sónia Moreira, ULS Coimbra

ORGANIZING COMMITTEE

Helder Esperto
Joana Salgado
João Gomes
Luísa Diogo
Maria Carmo Macário
Maria Guedes Marques
Rui Diogo
Sara Ferreira
Tabita Maia

SCIENTIFIC COMMITTEE

Daniel Costa Gomes, SPDM Board
Dulce Quelhas, SPDM President
Esmeralda Martins, SPDM Board
Hugo Rocha, SPDM Board
Patrícia Janeiro, SPDM Vice-President

https://simposio.spdm.org.pt/

SCIENTIFIC PROGRAMME

WEDNESDAY, 18TH MARCH

14:00--18:00	**Pre-Congress Course: Emergencies in IEM: Practical Protocols and Interdisciplinary Management**
18:00--19:00	**SPDM Working groups meeting**
19:00--20.00	**SPDM Nutrition groups meeting**

THURSDAY, 19TH MARCH

09:00	**Symposium Opening** *Symposium Chairperson - João Durães and Sónia Moreira*
09:20	**Session I - New avenues for diagnosis and treatment of Neurometabolic diseases** *Chairpersons: Maria Carmo Macário (Coimbra) and Arlindo Guimas (Porto)*
09:20	**From Disease Discovery to Treatment in DEGS1 leukodystrophy** *Aurora Pujol, Barcelona, Spain*
09:40	**New therapy advancements in Metachromatic Leukodystrophy** *TBC*
10:00	**Usefulness of blood tests in the diagnosis of GLUT1 deficiency syndrome** *Hana Pavlú Pereira*
10:20	Discussion
10:40	Coffee Break & Posters
11:10	**Session II - Managing and treating lysosomal storage disorders** *Chairpersons: Ana Cristina Ferreira (Lisbon) and TBC*
11:10	**New therapeutic approaches in Fabry disease** *Patrício Aguiar, Lisbon, Portugal*
11:30	**Management of pregnancy in lysosomal storage disorders** *Derralynn A Hughes, London, UK*
11:50	**Hematopoietic Stem Cell Gene Therapy for Mucopolysaccharidosis Type I: clinical outcomes** *Matilde Cossutta, Milan, Italy*
12:10	Discussion
12:30	**IMMEDICA Symposium: Accumulated clinical experience in the management of patients with urea cycle disorders: Long-term impact on patients' lives. New therapeutic options**
13:15	Lunch
14:30	**Session III - Mitochondrial diseases: are we ready for innovative therapies?** *Chairpersons: Célia Nogueira (Porto) and Margarida Paiva Coelho (Porto)*
14:30	**Three decades of translational research in Leber's Hereditary Optic Neuropathy: what have we learned?** *Manuela Grazina, Coimbra, Portugal*
14:50	**Recent advances in diagnosis and treatment of TK2 Deficiency** *Cristina Dominguez-González, Madrid, Spain*

15:10	**Small molecules as a therapeutic strategy in mitochondrial diseases** *Paulo Oliveira, Coimbra, Portugal*
15:30	Discussion
16:00	Coffee Break & Posters
16:40	**Session IV - Masterclass PKU- Past, Present, and Future Perspectives** *Chairpersons: Luísa Diogo (Coimbra) and Esmeralda Martins (Porto)*
	Discussion *Rita Jotta (Lisbon, Portugal), Carla Carmona (Porto, Portugal), Nanci Baptista (Coimbra, Portugal), Elisabete Almeida (APOFEN, Portugal)*

FRIDAY, 20TH MARCH

09:00	**Session V - Oral Communications** *Chairpersons: TBC*
10:00	**Session VI - Recent therapeutic advances in Glycogen Storage Diseases**
10:00	**Hepatic outcomes in adult patients with glycogen storage disease type III** *Kevin Kuriakose, Manchester, UK*
10:20	**Bempedoic acid prolongs fasting time in patients with GSD type 1a** *Anibh Das, Hannover, Germany*
10:40	**New avenues to treat Neutropenia in GSD type Ib and G6PC3-deficient patients** *Maria Veiga da Cunha, Leuven, Belgium*
11:00	Discussion
11:20	Coffee Break & Posters
12:00	**Session VII - Advances and Challenges in Therapeutic Approaches for Inherited Amino Acid Catabolism Disorders** *Chairpersons: Ana Oliveira (Coimbra) and TBC*
12:00	**Therapy for Urea Cycle Disorders: Current Practice and Future Prospects** *Julien Baruteau, London, UK*
12:20	**Liver Transplantation in Aminoacidopathies and Organic Acidemias: The Portuguese Experience** *Sara Ferreira, Coimbra, Portugal*
12:40	Discussion
13:00	Lunch

14:00	**Session VIII - Oral communications & selected posters** *Chairpersons: Hugo Rocha and TBC*
15:00	**Session IX - Therapeutic and Technological Innovation in Inherited Metabolic Diseases** *Chairpersons: Joana Rosmaninho Salgado (coimbra) and Anabela Oliveira (Lisbon)*
15:00	**Targeted Nanomedicine in Inherited Metabolic Diseases** *Jose Alvarez Gonzalez, Santiago de Compostela, Spain*
15:20	**Metabolic Reprogramming in Metabolic Diseases** *Paulo Gameiro, Lisbon, Portugal*
15:40	**Gene therapy in Inherited Metabolic Diseases** *Rui Nobre, Coimbra, Portugal*
16:00	Discussion
16:20	Coffee Break
16:50	**Session X - SPDM grants**
17:30	**Closing Session & awards** *João Durães and Sónia Moreira*
18:00	**SPDM General Assembly**