PROGRAMME _

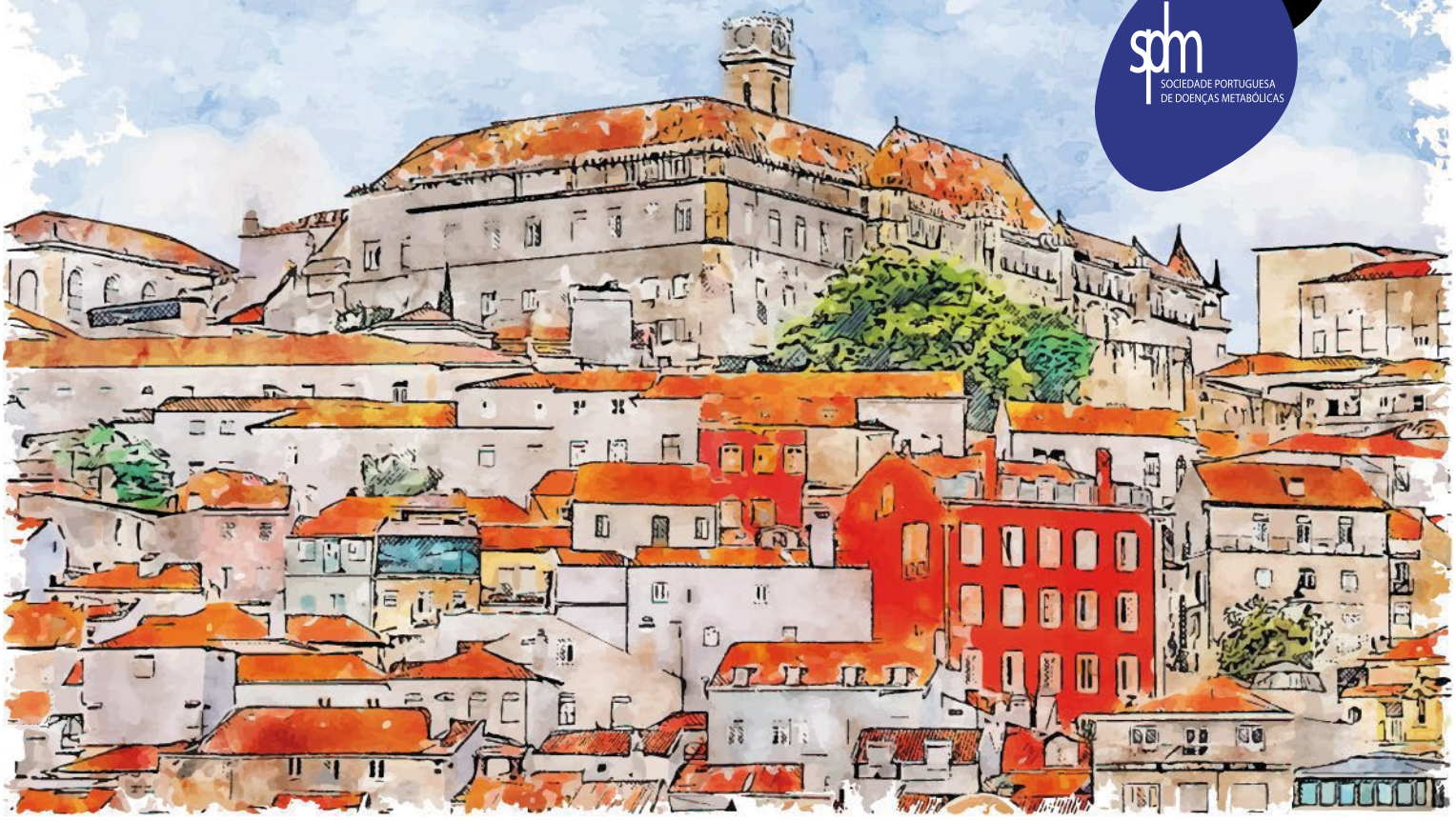
22ND INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

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

HOTEL VILA GALÉ, COIMBRA

18th–20th March

spdm SOCIEDADE PORTUGUESA DE DOENÇAS METABÓLICAS



18th-20th March

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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

08:30	**Secretariat Opening**
09:00	**Symposium Opening** *João Durães (Coimbra) and Sónia Moreira (Coimbra)*
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** *Cutillo Gianni, Milan, Italy*
10:00	**Usefulness of blood tests in the diagnosis of GLUT1 deficiency syndrome** *Hana Pavlú Pereira, Lisbon, Portugal*
10:20	Discussion
10:40	Coffee Break
11:10	**Session II - Managing and treating lysosomal storage disorders** *Chairpersons: Ana Cristina Ferreira (Lisbon) and João Gomes (Coimbra)*
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** *Elisa Leão Teles, Porto, Portugal* *Esmeralda Rodrigues, Porto, Portugal*
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 communications
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 (Porto, Portugal)*
17:45	**SPDM General Assembly**

FRIDAY, 20TH MARCH

09:00	**Session V - Oral Communications** *Chairpersons: Ana Paula Leandro (Lisboa) and Teresa Almeida Campos (Porto)*
10:00	**Session VI - Recent therapeutic advances in Glycogen Storage Diseases** *Chairpersons: Helder Esperto (Coimbra) and Anabela Bandeira (Porto)*
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 communications
12:00	**Session VII - Advances and Challenges in Therapeutic Approaches for Inherited Amino Acid Catabolism Disorders** *Chairpersons: Ana Oliveira (Coimbra) and Manuela Ferreira De Almeida (Porto)*
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** *Chairpersons: Hugo Rocha (Porto) and Mariana Pintalhão (Porto)*
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 & Posters communications
16:50	**Session X - SPDM grants** *Chairpersons: Dulce Quelhas (Porto) and Isabel Rivera (Lisbon)*
16:50	**Validation of a questionnaire to assess eating disorders in inherited metabolic disease patients requiring dietary treatment** *Inês Curvelo Mendes*
17:30	**Closing Session & awards** *João Durães (Coimbra) and Sónia Moreira (Coimbra)*