

PROGRAMME

22

ND

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

spm
SOCIEDADE PORTUGUESA
DE DOENÇAS METABÓLICAS





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



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<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 <i>Symposium Chairperson - João Durães</i>
09:20	Session I – New avenues for diagnosis and treatment of Neurometabolic diseases
09:20	From Disease Discovery to Treatment in DEGS1 leukodystrophy <i>Aurora Pujol</i>
09:40	New therapy advancements in Metachromatic Leukodystrophy <i>Caroline Bergner</i>
10:00	Usefulness of blood tests in the diagnosis of GLUT1 deficiency syndrome <i>Hana Pavlú Pereira</i>
10:20	Discussion
10:40	Coffee Break & Posters
11:10	Session II - Managing and treating lysosomal storage disorders
11:10	New therapeutic approaches in Fabry disease <i>Patrício Aguiar</i>
11:30	Management of pregnancy in lysosomal storage disorders <i>Derralynn Hughes</i>
11:50	Hematopoietic Stem Cell Gene Therapy for Mucopolysaccharidosis Type I: clinical outcomes <i>Matilde Cossuta</i>
12:10	Discussion
12:30	Sessão III - Oral Communications
13:30	Lunch
14:00	Industry session
15:00	Session IV - Mitochondrial diseases: are we ready for innovative therapies?
15:00	Three decades of translational research in Leber's Hereditary Optic Neuropathy: what have we learned? <i>Manuela Grazina</i>
15:20	Recent advances in diagnosis and treatment of TK2 Deficiency <i>Cristina Dominguez-González</i>
15:40	Small molecules as a therapeutic strategy in mitochondrial diseases <i>Paulo Oliveira</i>

16:00	Discussion
16:15	Coffee Break & Posters
17:00	Session V - From Deep Phenotyping to Innovative Therapies in IEM <i>Ana Gaspar (25min)</i>
	Discussion <i>Carla Carmona, Elisabete Almeida e Nanci Baptista</i>

FRIDAY, 20TH MARCH

09:00	Session VI – Recent therapeutic advances in Glycogen Storage Diseases
09:00	Hepatic outcomes in adult patients with glycogen storage disease type III <i>Kevin Kuriakose</i>
09:20	Bempedoic acid prolongs fasting time in patients with GSD type 1a <i>Anibh Das</i>
09:40	New avenues to treat Neutropenia in GSD type 1b and G6PC3-deficient patients <i>Maria Veiga da Cunha</i>
10:00	Discussion
10:20	Coffee Break & Posters
11:00	Session VII – Advances and Challenges in Therapeutic Approaches for Inherited Amino Acid Catabolism Disorders
11:00	Therapy for Urea Cycle Disorders: Current Practice and Future Prospects <i>Julien Baruteau</i>
11:20	Liver Transplantation in Aminoacidopathies and Organic Acidemias: The Portuguese Experience
11:40	Discussion
12:00	Industry symposium
13:00	Lunch
14:00	Session VIII - Oral communications & selected posters
15:00	Session IX - Therapeutic and Technological Innovation in Inherited Metabolic Diseases
15:00	Targeted Nanomedicine in Inherited Metabolic Diseases <i>Jose Alvarez Gonzalez</i>
15:20	Metabolic Reprogramming in Metabolic Diseases <i>Paulo Gameiro</i>
15:40	Gene therapy in Inherited Metabolic Diseases
16:00	Discussion
16:20	Coffee Break
16:50	Session X – SPDM grants communication
17:30	Closing Session & awards
18:00	SPDM General Assembly