

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**

spmh
SOCIEDADE PORTUGUESA
DE DOENÇAS METABÓLICAS





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**THE FUTURE OF INBORN ERRORS OF METABOLISM;
DECODING COMPLEXITY, DELIVERING INNOVATION**



SYMPOSIUM CHAIRPERSON

| João Durães, ULS Coimbra

ORGANIZING COMMITTEE

| João Durães, ULS Coimbra
| Sónia Moreira, ULS Coimbra

SCIENTIFIC COMMITTEE

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



<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
09:40	New therapy advancements in Metachromatic Leukodystrophy
10:00	Usefulness of blood tests in the diagnosis of GLUT1 deficiency syndrome
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
11:30	Management of pregnancy in lysosomal storage disorders
11:50	Hematopoietic Stem Cell Gene Therapy for Mucopolysaccharidosis Type I: clinical outcomes
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?
15:20	Recent advances in diagnosis and treatment of TK2 Deficiency



15:40	Small molecules as a therapeutic strategy in mitochondrial diseases
16:00	Discussion
16:15	Coffee Break & Posters
17:00	Session V - Masterclass PKU – Past, Present, and Future Perspectives

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
09:20	Bempedoic acid prolongs fasting time in patients with GSD type 1a
09:40	New avenues to treat Neutropenia in GSD type Ib and G6PC3-deficient patients
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
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
15:20	Metabolic Reprogramming in Inherited Metabolic Diseases
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