

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

18<sup>th</sup>-20<sup>th</sup>  
March

sph  
SOCIEDADE PORTUGUESA  
DE DOENÇAS METABÓLICAS





# 22<sup>ND</sup> | INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

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



## 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, 18<sup>TH</sup> MARCH

14:00- -18:00	<b>Pre-Congress Course: Emergencies in IEM: Practical Protocols and Interdisciplinary Management</b>
18:00- -19:00	<b>SPDM Working groups meeting</b>
19:00- -20:00	<b>SPDM Nutrition groups meeting</b>

### THURSDAY, 19<sup>TH</sup> MARCH

09:00	<b>Symposium Opening</b> <i>Symposium Chairperson - João Durães</i>
09:20	<b>Session I – New avenues for diagnosis and treatment of Neurometabolic diseases</b>
09:20	<b>From Disease Discovery to Treatment in DEGS1 leukodystrophy</b>
09:40	<b>New therapy advancements in Metachromatic Leukodystrophy</b>
10:00	<b>Usefulness of blood tests in the diagnosis of GLUT1 deficiency syndrome</b>
10:20	Discussion
10:40	Coffee Break & Posters
11:10	<b>Session II - Managing and treating lysosomal storage disorders</b>
11:10	<b>New therapeutic approaches in Fabry disease</b>
11:30	<b>Management of pregnancy in lysosomal storage disorders</b>
11:50	<b>Hematopoietic Stem Cell Gene Therapy for Mucopolysaccharidosis Type I: clinical outcomes</b>
12:10	Discussion
12:30	<b>Sessão III - Oral Communications</b>
13:30	Lunch
14:00	<b>Industry session</b>
15:00	<b>Session IV - Mitochondrial diseases: are we ready for innovative therapies?</b>
15:00	<b>Three decades of translational research in Leber's Hereditary Optic Neuropathy: what have we learned?</b>
15:20	<b>Recent advances in diagnosis and treatment of TK2 Deficiency</b>

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

## FRIDAY, 20<sup>TH</sup> MARCH

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