

19th International Symposium of the Portuguese Society for Metabolic Disorders

“The next step in inherited metabolic diseases:
from newborn screening to palliative care”

29th-31th March 2023

Eurostars Oasis Plaza - Figueira da Foz, Portugal

Wednesday 29 th March	
17:00	SPDM Nutrition Group Meeting
17:45	SPDM Groups Meeting
19:00	Welcome drink

Thursday 30 th March		
08:30	Registration opening	
09:00	Symposium Opening – Welcome address on behalf of the SPDM	
Session I – Inherited metabolic diseases in the lifetime		
<i>Chairpersons: Lelita Santos, Coimbra João Durães, Coimbra Maria João Guedes, Coimbra</i>		
09:10	Dietary management during the lifetime for IMD	Júlio Rocha (Lisbon)
09:30	Reproductive health and other challenges in adults with an IMD	Elaine Murphy (London)
09:50	Neurometabolic disorders in adults: IMD and beyond	Fanny Mochel (Paris)
10:10	Palliative care for IMD patients	Cândida Cancelinha (Coimbra)
10:30	Discussion	
10:50 11:20	Coffee break and Poster View	
Session II - Familial dyslipidaemias		
<i>Chairpersons: Ana Gaspar, Lisbon Teresa Cardoso, Porto Anália Carmo, Coimbra</i>		
11:20	Defects of lipoprotein metabolism: from the genes to the clinic	Adriaan Holleboom (Amsterdam)
11:40	Familial hypercholesterolaemia in children and adolescents - better safe than sorry	Albert Wiegman (Amsterdam)

12:00	The Portuguese screening programs for familial dyslipidaemias	Mafalda Bourbon (Lisbon)
12:20	Discussion	
12:40 14:30	Lunch	
Session III – Are we moving forward in lysosomal storage disorders?		
<i>Chairpersons: Esmeralda Rodrigues, Porto Sandra Alves, Porto Lúcia Lacerda, Porto</i>		
14:30	The future of enzyme replacement therapy	Eugen Mengel (Hochheim)
14:50	The path to gene therapy in lysosomal storage disorders	Francesca Tucci (Milan)
15:10	Registries in inherited metabolic diseases: how to move forward?	Nadia Belmatoug (Paris)
15:30	Discussion	
16:00 16:30	Coffee break and Poster View	
Session IV – Oral Communications I		
<i>Chairpersons: Sara Ferreira, Coimbra Isabel Rivera, Lisbon</i>		
17:30	SPDM General Assembly	
19:00	Departure to dinner	

Friday 31st March		
Session V – New approaches in porphyrias		
<i>Chairpersons: Arlindo Guimas, Porto Sónia Moreira, Coimbra Eulália Costa, Coimbra</i>		
09:00	Acute hepatic porphyria – The experience of a reference center in Brazil	Charles Marques Lourenco, (Ribeirão Preto)
09:20	Biochemical and Molecular Update on Porphyria Diagnosis	Filipa Ferreira (INSA Porto)
09:40	The RNAi therapeutics revolution: from bench to bedside and back again	Pedro Moreno (Porto)
10:00	Discussion	
10:30 11:00	Coffee break	
Session VI – Thinking big and bold in inherited metabolic diseases		
<i>Chairpersons: Laura Vilarinho, Porto Ana Cristina Ferreira, Lisbon Anabela Oliveira, Lisbon</i>		
11:00	Challenges and Opportunities for using of real world data in metabolic medicine	Mário Silva (Lisbon)
11:20	Newborn screening by WGS: opportunities and challenges	David Bick (London)
11:40	Aging in IMD: uncovering new phenotypes	Charles Lourenço (Ribeirão Preto)
12:00 12:20	Discussion	
Session VII – Oral Communications II		
<i>Chairpersons: Joana Salgado, Coimbra João Gomes, Coimbra</i>		
13:00 14:30	Lunch and Satellite Symposium II	
Session VIII – Unmet needs in inherited metabolic diseases		
<i>Chairpersons: Paulo Gonçalves, Lisbon Fátima Ventura, Lisbon Rui Tato Marinho, Lisbon</i>		
14:30	Unmet needs for the patient and the family	José Vilhena (Coimbra)
14:50	The pharmacist and the specificities of IMD	Sara Dias (Lisbon)
15:10	Reference Centres of IMD in Portugal - challenges and opportunities	Luisa Diogo (Coimbra)

15:30	Discussion	
15:50		
Session IX – Spotlight I		
Chairpersons: M Helena Santos, Vila Nova de Gaia / <i>Nanci Batista, Coimbra</i>		
15:50	Diet quality and saproterin dihydrochloride (BH4) use in children with phenylketonuria (PKU)	Maria Inês Gama (Lisbon)
16:10	Novel insights into treatment strategies for hyperammonemia-associated urea cycle disorders and organic acidurias	Margarida Silva (Lisbon)
16:30 17:00	Coffee break	
Session X – Spotlight II		
Chairpersons: Paulo Castro Chaves, Portugal / <i>Paula Leandro, Lisbon</i>		
17:00	Something stinks': impaired hydrogen sulfide and cysteine persulfide production by cystathionine β -synthase variants identified in classical homocystinuria patients	João Vicente (Lisbon)
17:20	Improve management of MADD patients: a curated database with clinical, molecular and cellular information	Bárbara Henriques (Lisbon)
17:40	Awards and Final remarks	
18:00	End of the Symposium	