

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 Lelita Santos (coimbra); João Durães (Coimbra)		
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	Coffee break	
Session II - Familial dyslipidaemias Ana Gaspar (Lisboa); Teresa Cardoso (Porto); Eulália Costa (coimbra)		
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	Lunch /Poster View	
Session III – Are we moving forward in lysosomal storage disorders? Esmeralda Rodrigues (Porto); Sandra Alves (Porto); Lucia Lacerda (Porto)		
14:30	The future of enzyme replacement therapy	Eugen Mengel
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	Coffee break	
Session IV – Oral communications I Sara Ferreira (Coimbra); Isabel Riviera (lisboa)		
16:30		
17:30	SPDM General Assembly	
19:00	Departure to dinner	

Friday 31st March

Session V – New approaches in porphyrias

Arlindo Guimas (Porto); Sónia Moreira (Coimbra);

09:00	Sporadic and latent acute hepatic porphyria	Por confirmar:
09:20	Mechanisms of neuronal damage in acute hepatic porphyrias	Por confirmar:
09:40	The RNAi therapeutics revolution: from bench to bedside and back again	Pedro Moreno (I3S Porto)
10:00	Discussion	

10:30	Coffee break	
Session VI – Thinking big and bold in inherited metabolic diseases Laura Vilarinho (INSA); Fabiana Ramos (Coimbra); Anabela Oliveira (Lisboa)		
11:00	Big data and artificial intelligence 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	Discussion	
	Session VII – Oral communications II Joana Salgado (Coimbra); Paulo Chaves (Porto)	
12:20		
13:00	Lunch /Poster View	
Session VIII – Unmet needs in inherited metabolic diseases Chairpersons: Paulo Gonçalves (RD-Portugal); Fátima Ventura (Infarmed); Rui Tato Marinho (Presidente Comissão CR)		
14:30	Unmet needs for the patient and the family	José Vilhena (Coimbra)
14:50	The pharmacist and the specificities of IMD	Sara Dias (Lisboa)
15:10	Reference Centres of IMD in Portugal - challenges and opportunities	Luisa Diogo
15:30	Discussion	
Session IX – Spotlight I Ana Cristina Ferreira (Lisboa); Nanci Batista (Coimbra)		
15:50	Diet quality and saproterin dihydrochloride (BH4) use in children with phenylketonuria (PKU)	Maria Inês Gama
16:10	Novel insights into treatment strategies for hyperammonemia-associated urea cycle disorders and organic acidurias.	Margarida Silva
16:30	Coffee break	
Session X – Spotlight II Helder Esperto (Coimbra); Paula Leandro (Lisboa)		
17:00	Something stinks': impaired hydrogen sulfide and cysteine persulfide production by cystathionine β -	João Vicente

	synthase variants identified in classical homocystinuria patients	
17:20	Improve management of MADD patients: a curated database with clinical, molecular and cellular information	Bárbara Henriques
17:40	Awards and Final remarks	
18:00	End of the Symposium	