

## 19th International Symposium of the Portuguese Society for Metabolic Disorders

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

29<sup>th</sup>-31<sup>th</sup> March 2023

Eurostars Oasis Plaza - Figueira da Foz, Portugal

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

Thursday 30 <sup>th</sup> March		
08:30	<b>Registration opening</b>	
09:00	<b>Symposium Opening – Welcome address on behalf of the SPDM</b>	
<b>Session I – Inherited metabolic diseases in the lifetime</b>		
<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	<b>Coffee break and Poster View</b>	
<b>Session II - Familial dyslipidaemias</b>		
<i>Chairpersons: Ana Gaspar, Lisbon   Teresa Cardoso, Porto   TBC</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	<b>Lunch</b>	
<b>Session III – Are we moving forward in lysosomal storage disorders?</b>		
<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	<b>Coffee break and Poster View</b>	
<b>Session IV – Oral Communications I</b>		
<i>Chairpersons: Sara Ferreira, Coimbra   Isabel Rivera, Lisbon</i>		
17:30	<b>SPDM General Assembly</b>	
19:00	<b>Departure to dinner</b>	

<b>Friday 31st March</b>		
<b>Session V – New approaches in porphyrias</b>		
<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	
<b>10:30</b> <b>11:00</b>	<b>Coffee break</b>	
<b>Session VI – Thinking big and bold in inherited metabolic diseases</b>		
<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	
<b>Session VII – Oral Communications II</b>		
<i>Chairpersons: Joana Salgado, Coimbra   João Gomes, Coimbra</i>		
<b>13:00</b> <b>14:30</b>	<b>Lunch and Satellite Symposium II</b>	
<b>Session VIII – Unmet needs in inherited metabolic diseases</b>		
<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		
<b>Session IX – Spotlight I</b>		
<b>Chairpersons:</b> 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)
<b>16:30</b> <b>17:00</b>	<b>Coffee break</b>	
<b>Session X – Spotlight II</b>		
<b>Chairpersons:</b> Paulo Castro Chaves, Portugal / <i>Paula Leandro, Lisbon</i>		
17:00	Something stinks': impaired hydrogen sulfide and cysteine persulfide production by cystathionine $\beta$ -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	<b>Awards and Final remarks</b>	
18:00	<b>End of the Symposium</b>	