

Thursday, March 16th

08:45	Welcome Address Isabel Tavares de Almeida, SPDM President
9:00 -11:00	Session I – The future of diagnostics for IEM Chairpersons: Ronald Wanders, Amsterdam, NL and Sérgio B. de Sousa , Coimbra, PT
9:00 - 9:20	The role of genomics: where are we going? Johannes Zschocke, Innsbruck, AT
9:25 - 9:45	Untargeted metabolomics in a diagnostic setting Judith Jans, Utrecht, NL
9:50 – 10:05	Invited short oral communication: Aminoacyl-tRNA synthetase defects: development of novel diagnostic strategies Marisa Mendes, Amsterdam, NL
10:05 - 10:20	Invited short oral communication: Biomarkers characterization in organic acidurias: development of a metabolomics platform Sara Violante, New York, USA
10:20 - 10:35	How to face big data - Open discussion <i>Hugo Rocha, Porto, PT</i>
10:40 - 11:10	Coffee break
11:10 - 13:00	Session II – Lysosomal disorders Chairpersons: Anabela Oliveira, Lisboa, PT and Paula Garcia, Coimbra, PT
11:10 -11:30	Cystinosis in adolescent and adult patient: recommendations for transition and the comprehensive care <i>Gema Ariceta, Barcelona, SP</i>
11:35 - 11:55	Lysosomal lipase acid deficiency: from suspicion to treatment François Feillet, Vandoeuvre les Nancy, FR
12:00 - 12:20	Fabry Disease 10 years on: progress or progression? Robin Lachmann, London, UK
12:25 – 12:40	Invited short oral communication: Immune alterations in Progranulin mutation carriers have functional consequences in NCL: data from a human NCL model Ana Luisa Cardoso, Coimbra, PT
12:40 – 12:55	Invited short oral communication: Cholesterol 24S-hydroxylase as a new therapeutic target in Niemann-Pick type C disease Elsa Rodrigues, Lisboa, PT
12:55 - 13:55	Lunch



14:00 - 15:45	Session III – Neurometabolic disorders Chairpersons: Mireia del Toro, Barcelona, SP and José Pedro Vieira, Lisboa, PT
14:00 - 14:20	Treatable newborn and infant seizures due to IEM Barbara Plecko, Zurich, CH
14:25 – 14:45	The biochemistry of vitamin B6 dependent epilepsy Nanda Verhoeven–Duif, Utrecht, NL
14:50 - 15.10	Metabolic epilepsies of late childhood and adolescence Georg Hoffmann, Heidelberg, DE
15:15 -15:30	Neuronal–ceroid lipofuscinosis (NCL) and epilepsy: clinical cases and therapeutic perspectives <i>Sofia Quintas, Lisboa, PT</i>
15:35 – 15:50	Invited short oral communication: Vitamin B6 is essential in serine <i>de novo</i> biosynthesis Rúben Ramos, Utrecht, NL

15:50 - 16:10 **Coffee Break**

16:10 - 17:40 Short oral communications (from selected abstracts) Chairpersons: Teresa Cardoso, Porto, PT and Esmeralda Rodrigues, Porto, PT

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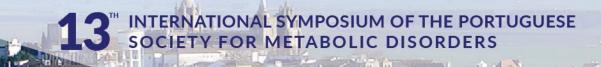


Friday, March 17th

09:00-10:30	Short oral communications (from selected abstracts) Chairpersons: Paula Leandro, Lisboa,PT and Júlio Rocha, Porto, PT
10:30-11:00	Coffee-break
11:00 -13:00	Session IV – Looking beyond the nutritional treatment: a new reality Chairpersons: Margarida B. Silva, Lisboa, PT and Esmeralda Martins, Porto, PT
11:00 -11:20	Nutritional management in MSUD: the experience from the USA guidelines Rani Singh, Decartur - Georgia, USA
11:25 – 11:45	The therapeutic potential of Metformin in MSUD Arvind Ramanathan, Novato –California, USA
11:50 - 12:10	LCFA oxidation defects: treatment strategies Ute Spiekerkoetter, Freiburg, DE
12:15 - 12:35	Triheptanoin acid in LCFA oxidation defects Jerry Vockley, Pittsburgh - Pennsylvania, USA
12:40 - 13:00	MCAD deficiency – whom to treat and how Soeren Gersting, Munch, DE
13:05 – 14:05	Lunch
14:10 – 15:40	Session IV (cont) – Looking beyond the nutritional treatment: a new reality Chairpersons: Patricia Janeiro, Lisboa, PT and Rita Castro, Lisboa,, PT
14:10 -14:30	
11.10 11.50	PKU and sapropterin dihydrochloride: management of very young children Ania Muntau, Hamburg, DE
14:35 – 14:55	
	Ania Muntau, Hamburg, DE Cobalamin related remethylation disorders and MTHFR deficiency: the role of the guidelines in the harmonisation of diagnosis, treatment and follow-up

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- 16:05 17:05 Short oral poster communications (from selected posters) Chairpersons: Ana Cristina Ferreira, Lisboa, PT and Dulce Quelhas, Porto, PT 17:05 - 17:15 Awards / Closing Remarks 17:15 End of the Meeting
- 17:30 Assembleia Geral (membros da SPDM) / Annual General Meeting (SPDM members)

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