

# 13<sup>TH</sup> INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS



Thursday, March 16<sup>th</sup>

- 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  
*Hugo Rocha, Porto, PT*
- 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  
*Gema Ariceta, Barcelona, SP*
- 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**

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## 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*

The biochemistry of vitamin B6 dependent epilepsy

14:25 – 14:45 *Nanda Verhoeven–Duif, Utrecht, NL*

14:50 – 15:10 Metabolic epilepsies of late childhood and adolescence

*Georg Hoffmann, Heidelberg, DE*

Neuronal–ceroid lipofuscinosis (NCL) and epilepsy: clinical cases and therapeutic perspectives

15:15 -15:30 *Sofia Quintas, Lisboa, PT*

15:35 – 15:50 *Invited short oral communication:*

Vitamin B6 is essential in serine *de novo* 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 17<sup>th</sup>

- 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, Decatur - 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 PKU and sapropterin dihydrochloride: management of very young children  
*Ania Muntau, Hamburg, DE*
- 14:35 – 14:55 Cobalamin related remethylation disorders and MTHFR deficiency: the role of the guidelines in the harmonisation of diagnosis, treatment and follow-up  
*Manuel Schiff, Paris, FR*
- 15:00 – 15:20 Riboflavin, *MTHFR* genotype and blood pressure: a personalized approach to reducing disease risk  
*Helene McNulty, Northern Ireland, UK*
- 15:25 – 15:40 *Invited short oral communication:*  
Homocysteine metabolism disruption may affect the crosstalk of histone methylation dynamics with gene regulation  
*Rúben Esse, Boston, USA*

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- 15:40 – 16:00      **Coffee break**
- 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)**

## Secretariat:



it's (ideas, tools & solutions)  
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