

Scientific Programme

Thursday, 15 March

08:00

Registration Opening

08:45

Symposium Opening - Welcome Address on behalf of the SPDM
Local organization representatives

Session I – New players in Inborn Errors of Metabolism

Moderators: Isabel Rivera, *PT* and Luisa Azevedo, *PT*

09:00

Metabolic adaptation as a protective strategy against infectious diseases

Ana Rita Carlos | *Lisboa, PT*

09:30

Gut microbioma and hyperammonemia – problem or solution?

David Shen | *Philadelphia, USA*

10:00

Epigenetic changes and phenotypic variation in IEM

Joseph H. Nadeau | *Seattle, USA*

10:30 Coffee Break

Session II – Inborn Errors of Metabolism cross talk with Immune system

Moderators: Patrícia Janeiro, *PT* and Fátima Macedo, *PT*

11:00

Inborn errors of metabolism underlying primary immunodeficiency's – an emerging field

Joris van Montfrans | *Utrecht, ND*

11:30

Impacts of free amino acids on the immune system

Thomas Vorup-Jensen | *Aarhus, DK*

Session III – New Inborn Errors of Metabolism

Moderators: Laura Vilarinho, PT, Paulo Castro Chaves, PT

12:00

Blood platelets and genome sequencing to unravel unknown metabolic diseases

Kathleen Freson | *Leuven, BE*

12:30

Odyssey to a new disorder

Ron Wevers | *Nijmegen, ND*

13:00 Poster View / Lunch

Session IV – Congenital Disorders of Glycosylation

14:00

Moderators: Esmeralda Martins, PT and Luísa Diogo, PT

Lessons from NGS approach to CDG diagnostic

Gert Mathijs | *Leuven, BE*

14:30

Treating CDGs – always a challenge

Jaak Jaeken | *Leuven, BE*

15:00 Coffee Break

Session V – Selected Free Communications

Moderators: Maria Carmo Macário, PT and Paula Leandro, PT

15:30 Beginning of the Session

16:55 End of the Session

17:10 SPDM Assembleia Geral

Session VI – Selected Free Communications

Moderators: Daniel Gomes *PT* and Anabela Bandeira, *PT*

08:00-09:00

Session VII – LSD - still much to learn

Moderators: Lúcia Lacerda, *PT* and Sílvia Sequeira, *PT*

09:10

Immunity and Inflammation in LSD

Brian Bigger | *Manchester, UK*

09:35

Bone growth in lysosomal storage disorders – role of autophagy and new paths for treatment

Carmine Settembre | *Naples, IT*

10:00

Bridging the age spectrum of neurodegenerative storage diseases

Barry Boland | *Cork, IR*

10:25 Coffee break

Session VIII – LSD treatment – reality and expectations

Moderators: Paul Harmatz, *USA* and Elisa Leão Teles, *PT*

11: 00

Advances in gene therapy for LSD

Brian Bigger | *Manchester, UK*

11:25

Treatment with pentosane polysulphate in patients with MPS I

Calogera Simonaro | *New York, USA*

Session IX – Selected free Communications

Moderators: Arlindo Guimas, *PT* and Ana Cristina Ferreira, *PT*

11:50 Beginning of the Session

12:40 End of the Session

12:45 Poster View / Lunch

IV SPDM Nutrition meeting (SPDM-GN)

Session XI – Inborn Errors of Metabolism nutritional and pharmacological treatment – Present and future

Moderators: Paula Garcia, PT and Anabela Oliveira, PT

14:35

Vitamin and Micronutrients Metabolism – role in IEM treatment

Ana Moráis López | *Madrid, SP*

15:00

Sapropterin treatment of PKU- the Portuguese picture

Pku think tank | PT

15:20

Carbaglumic acid – lessons from treating organic acidurias

Sufin Yap | *Sheffield, UK*

15:45 Coffee Break

Session XII – Selected poster Communications

16:00 – 17:00

Moderators: Júlio Rocha PT, Teresa Cardoso PT

17:00 Awards / Closing Remarks

Isabel Tavares de Almeida, SPDM President

17:30 SPDM Assembleia eleitoral