

# PROGRAMME

# 21<sup>ST</sup>

INTERNATIONAL SYMPOSIUM  
OF THE PORTUGUESE SOCIETY  
FOR METABOLIC DISORDERS

RAISING AWARENESS  
ON INHERITED  
METABOLIC DISORDERS



26<sup>th</sup> - 29<sup>th</sup>  
March



 SANA METROPOLITAN HOTEL, LISBON



21<sup>ST</sup>

**INTERNATIONAL SYMPOSIUM  
OF THE PORTUGUESE SOCIETY  
FOR METABOLIC DISORDERS**

**RAISING AWARENESS ON INHERITED METABOLIC DISORDERS**

26<sup>th</sup> March | SPDM Working & Nutrition Groups Meeting

27<sup>th</sup> - 28<sup>th</sup> March | 21<sup>st</sup> SPDM International Symposium

29<sup>th</sup> March | Raising awareness in innovative therapies in IMD

## **SYMPOSIUM CHAIRPERSON**

| Daniel Costa Gomes, ULS Santa Maria

## **ORGANIZING COMMITTEE**

| Cristina Florindo, Faculdade de Farmácia, ULisboa

| Margarida F. B. Silva, Faculdade de Farmácia, ULisboa

| Patrícia Janeiro, ULS Santa Maria

| Patrícia Lipari Pinto, ULS Santa Maria

| Rita Loureiro, ULS Santa Maria

## **SCIENTIFIC COMMITTEE**

| Daniel Costa Gomes, SPDM Board

| Dulce Quelhas, SPDM President

| Esmeralda Martins, SPDM Board

| Hugo Rocha, SPDM Board

| Patrícia Janeiro, SPDM Vice-President

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## SCIENTIFIC PROGRAMME

### WEDNESDAY, 26<sup>TH</sup> MARCH

17:00 | SPDM Working Groups Meeting

18:00 | SPDM Nutrition Group Meeting

### THURSDAY, 27<sup>TH</sup> MARCH

09:00 | **Symposium Opening – Welcome Address on behalf of the SPDM**

*Symposium Chairperson - Daniel Costa Gomes*

09:15 | **Session I – Innovation in diagnostic methods for classic Inherited Metabolic Disorders (IMD)**

*Chairperson - Célia Nogueira, Porto, PT | Margarida F. B. Silva, Lisbon, PT*

09:15 | **Metabolomics to screen for classic IMD**

*George Ruijter, Rotterdam, NL*

09:40 | **Advances on integrative omics approaches to IMD diagnosis**

*Holger Prokisch, Munich, DE*

10:05 | **Quantitative proteomics in mitochondrial disease - where are we now?**

*Marco Moedas, Stockholm, SE*

10:30 | **Lipidomics in IMD for biomarker discovery**

*Frédéric Vaz, Amsterdam, NL*

10:55 | **Coffee Break**

11:30 | **Session II – Rethinking Newborn Screening (NBS)**

*Chairperson - Esmeralda Martins, Porto, PT | Laura Vilarinho, Porto, PT*

11:30 | **New challenges in NBS**

*Hugo Rocha, Porto, PT*

11:55 | **Challenges of using whole genome sequencing in population NBS**

*Robin Lachmann, London, UK*

12:20 | **Lessons from adult metabolic medicine**

*Fanny Mochel, Paris, FR*

12:45 | **Lunch Symposium**

**Cipaglucosidase alfa + miglustat: Now you can do more (+) for your Pompe patients (AMICUS)**

*Arlindo Guimas, Porto, PT*

14:15 | **Session III – Raising awareness on neurometabolic diseases**

*Chairperson - Patrícia Janeiro, Lisbon, PT | João Durães, Coimbra, PT*

14:15 | **Metabolic epilepsies - clinical and biochemical footprints of IMD**

*Ángeles García-Cazorla, Barcelona, ES*

14:40 | **Update on leukodystrophies and developing trials**

*Karen Gunanayagam, London, UK*

15:05 | **Movement disorder increases with age in argininosuccinic aciduria**

*Julien Baruteau, London, UK*

15:30 | **Coffee Break**

16:00 | **Session IV – Oral Communications**

*Chairperson - Teresa Campos, Porto, PT | Isaura Rodrigues, Porto, PT*

17:00 | **Session V – E-posters guided walk**

19:30 | **Departure to Dinner**



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## FRIDAY, 28<sup>TH</sup> MARCH

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| <b>09:00</b> | <b>Session VI – Development and challenges for new therapies in IMD</b><br>Chairperson - Arlindo Guimas, Porto, PT   Margarida Coelho, Porto, PT   |
| 09:00        | <b>The basis for mRNA-based therapies in IMD</b><br>Fátima Ventura, Lisbon, PT   |
| 09:25        | <b>mRNA therapy for lysosomal disorders</b><br>Sandra Alves, Porto, PT   |
| 09:50        | <b>Mitochondrial disorders - hope for the future?</b><br>Shamima Rahman, London, UK  |
| 10:15        | <b>New treatment approaches for CDG</b><br>Dulce Quelhas, Porto, PT  |
| <b>10:40</b> | Coffee Break   |
| <b>11:15</b> | <b>Session VII – Revisiting nutritional approaches in IMD</b><br>Chairperson - Júlio César Rocha, Lisbon, PT   Vânia Magalhães, Porto, PT  |
| 11:15        | <b>Exploring asymptomatic childhood methionine cycle disorders</b><br>Patrícia Lipari Pinto, Lisbon, PT  |
| 11:40        | <b>Supplementation for performance and health in patients with phenylketonuria</b><br>Domingo González-Lamuño, Santander, ES   |
| 12:05        | <b>Personalised parenteral nutrition for acute IMD decompensations</b><br>Marjorie Dixon, London, UK   |
| <b>12:30</b> | Lunch Symposium<br><b>Unmet Needs of ARG1-D Patients: New Perspectives (IMMEDICA)</b><br>Elisa Leão Teles, Porto, PT   |
| <b>14:00</b> | <b>Session VIII – Selected E-posters Communications</b><br>Chairperson - Ana Cristina Ferreira, Lisbon, PT   Hugo Rocha, Porto, PT   |
| <b>15:00</b> | <b>Session IX – Moving from research and clinical practice to digital health and decision making in IMD</b><br>Chairperson - Bárbara J. Henriques, Lisbon, PT   Daniel Costa Gomes, Lisbon, PT |
| 15:00        | <b>The European Health Data Space: impact for disease management at the individual and population level</b><br>Cátia Sousa Pinto, Lisbon, PT   |
| 15:25        | <b>Using data science for better healthcare and opportunities in IMD</b><br>João Guimarães, Lisbon, PT   |
| 15:50        | <b>Sustainability in the development of the treatments for IMD</b><br>Rui Santos Ivo, Lisbon, PT   |
| <b>16:15</b> | Coffee Break   |
| <b>16:45</b> | <b>Session X – SPDM grants communications</b><br>Chairperson - Dulce Quelhas, Porto, PT   Isabel Tavares de Almeida, Lisbon, PT  |
| 16:45        | <b>Can enzyme replacement therapy revert iNKT cell dysfunction in acid sphingomyelinase deficiency patients?</b><br>Fátima Macedo, Porto, PT   |
| 17:05        | <b>Functional and structural impact of 10 ACADM missense mutations on human medium chain acyl-CoA dehydrogenase</b><br>Ana Paula Leandro, Lisbon, PT   |
| <b>17:30</b> | <b>Awards and Closing Remarks</b>  |
| <b>17:30</b> | <b>End of the Symposium</b>  |
| <b>18:00</b> | <b>SPDM General Assembly</b>   |

## SATURDAY, 29<sup>TH</sup> MARCH

Raising awareness in innovative therapies in IMD