

PROGRAMME

21ST

INTERNATIONAL SYMPOSIUM
OF THE PORTUGUESE SOCIETY
FOR METABOLIC DISORDERS

RAISING AWARENESS
ON INHERITED
METABOLIC DISORDERS



26th - 29th
March



SANA METROPOLITAN HOTEL, LISBON



21ST

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

RAISING AWARENESS ON INHERITED METABOLIC DISORDERS

26th March | SPDM Working & Nutrition Groups Meeting

27th - 28th March | 21st SPDM International Symposium

29th 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



SCIENTIFIC PROGRAMME

(preliminary)

WEDNESDAY, 26TH MARCH

17:00 | SPDM Working Groups Meeting

18:00 | SPDM Nutrition Group Meeting

THURSDAY, 27TH 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)**

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)**

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 | Networking lunch

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

14:15 | **Metabolic epilepsies - clinical and biochemical footprints of IMD**
Angeles Garcia Cazorla, Barcelona, ES

14:40 | **Update on leukodystrophies and developing trials**
Giorgia Ceravolo, 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**

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

19:30 | Departure to Dinner



SCIENTIFIC PROGRAMME

(preliminary)

FRIDAY, 28TH MARCH

09:00	Session VI – Development and challenges for new therapies in IMD
09:00	The basis for mRNA-based therapies in IMD <i>Fátima Ventura, Lisbon, PT</i>
09:25	mRNA therapy for lysosomal disorders <i>Sandra Alves, Porto, PT</i>
09:50	Mitochondrial disorders - hope for the future? <i>Shamima Rahman, London, UK</i>
10:15	New treatment approaches for CDG <i>Dulce Quelhas, Porto, PT</i>
10:40	Coffee Break
11:15	Session VII – Revisiting nutritional approaches in IMD
11:15	Exploring asymptomatic childhood methionine cycle disorders <i>Patrícia Lipari Pinto, Lisbon, PT</i>
11:40	Supplementation for performance and health in patients with phenylketonuria <i>Domingo Gonzalez-Lamuño, Santander, ES</i>
12:05	Personalised parenteral nutrition for acute IMD decompensations <i>Marjorie Dixon, London, UK</i>
12:30	Networking lunch
14:00	Session VIII – Selected E-posters Communications
15:00	Session IX – Moving from research and clinical practice to digital health and decision making in IMD
15:00	The European Health Data Space: impact for disease management at the individual and population level <i>Cátia Sousa Pinto, Lisbon, PT</i>
15:25	Sustainability in the development of the treatments for IMD <i>Rui dos Santos Ivo, Lisbon, PT</i>
15:50	Using data science for better healthcare and opportunities in IMD <i>João Guimarães, Lisbon, PT</i>
16:15	Coffee Break
16:45	Session X – SPDM grants communications
16:45	Can enzyme replacement therapy revert <i>iNKT</i> cell dysfunction in acid sphingomyelinase deficiency patients? <i>Fátima Macedo, Porto, PT</i>
17:05	Functional and structural impact of 10 <i>ACADM</i> missense mutations on human medium chain acyl-CoA dehydrogenase <i>Ana Paula Leandro, Lisbon, PT</i>
17:30	Awards and Final Remarks
17:30	End of the Symposium