

## 19th International Symposium of the Portuguese Society for Metabolic Disorders

“The next steps in Inborn Errors of Metabolism: from newborn screening  
to palliative care”

29<sup>th</sup>-31<sup>th</sup> March 2023

Eurostars Oasis Plaza - Figueira da Foz, Portugal

| Wednesday 29 <sup>th</sup> March |                              |
|----------------------------------|------------------------------|
| 17:00                            | SPDM Nutrition Group Meeting |
| 17:45                            | SPDM Groups Meeting          |
| 19:00                            | Welcome drink                |

| Thursday 30 <sup>th</sup> March |  |                                       |
|---------------------------------|--|---------------------------------------|
| 08:30                           | Registration opening   |                                       |
| 09:00                           | Symposium Opening – Welcome address on behalf of the SPDM<br>Chairpersons: To be confirmed |                                       |
| 09:10                           | Session I – Inherited metabolic diseases in the lifetime                                   |                                       |
| 10:50                           | Chairpersons: TBC  |                                       |
| 09:10                           | Dietary management during the lifetime for IMD   | <i>Júlio Rocha, Portugal</i>          |
| 09:30                           | Reproductive health and other challenges in adults with an IMD                             | <i>Elaine Murphy, United Kingdom</i>  |
| 09:50                           | Neurometabolic disorders in adults: IMD and beyond   | <i>Fanny Mochel, France</i>           |
| 10:10                           | Palliative care for IMD patients   | <i>Cândida Cancelinha, Portugal</i>   |
| 10:30                           | Discussion   |                                       |
| 10:50                           | Coffee break   |                                       |
| 11:20                           | Session II - Familial dyslipidaemias   |                                       |
| 12:40                           | Chairpersons: To be confirmed  |                                       |
| 11:20                           | Defects of lipoprotein metabolism: from the genes to the clinic                            | <i>Adriaan Holleboom, Netherlands</i> |
| 11:40                           | Familial hypercholesterolaemia in children and adolescents - better safe than sorry        | <i>Albert Wiegman, Netherlands</i>    |
| 12:00                           | The Portuguese screening programs for familial dyslipidaemias                              | <i>Mafalda Bourbon, Portugal</i>      |
| 12:20                           | Discussion   |                                       |
| 12:40                           | Lunch and Satellite Symposium  |                                       |
| 14:30                           | Session III – Are we moving forward in lysosomal storage disorders?                        |                                       |
| 16:00                           | Chairpersons: To be confirmed  |                                       |
| 14:30                           | The future of enzyme replacement therapy   | <i>To be confirmed</i>                |
| 14:50                           | The path to gene therapy in lysosomal storage disorders                                    | <i>Francesca Tucci, Italy</i>         |

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| 15:10 | Registries in inherited metabolic diseases: how to move forward? | <i>Nadia Belmatoug, France</i> |
| 15:30 | Discussion   |                                |
| 16:00 | <b>Coffee break</b>  |                                |
| 16:30 | <b>Session IV – Oral communications I</b>                        |                                |
| 17:30 | <b>Chairpersons:</b> To be confirmed                             |                                |
| 17:30 | <b>SPDM General Assembly</b>                                     |                                |
| 19:00 | <b>Departure to dinner</b>                                       |                                |

| <b>Friday 31st March</b> |  |                                   |
|--------------------------|--|-----------------------------------|
| 09:00                    | <b>Session V – New approaches in porphyrias</b>  |                                   |
| 10:30                    | <b>Chairpersons:</b> To be confirmed   |                                   |
| 09:00                    | RNAi and the revolution in therapeutical approaches to inherited metabolic diseases: acute hepatic porphyria at the target | <i>Pedro Moreno, Portugal</i>     |
| 09:20                    | Mechanisms of neuronal damage in acute hepatic porphyrias  | <i>Miguel Oliveira, Portugal</i>  |
| 09:40                    | Sporadic and latent acute hepatic porphyria – an EPNET Centre Experience   | <i>To be Confirmed</i>            |
| 10:00                    | Discussion   |                                   |
| 10:30                    | <b>Coffee break and Poster view</b>  |                                   |
| 11:00                    | <b>Session VI – Thinking big and bold in inherited metabolic diseases</b>  |                                   |
| 12:20                    | <b>Chairpersons:</b> To be confirmed   |                                   |
| 11:00                    | Big data and artificial intelligence in metabolic medicine   | <i>Mário Silva, Portugal</i>      |
| 11:20                    | Newborn screening by WGS: opportunities and challenges   | <i>David Bick, United Kingdom</i> |
| 11:40                    | Aging in IMD: uncovering new phenotypes  | <i>Charles Lourenço, Brasil</i>   |
| 12:00                    | Discussion   |                                   |
| 12:20                    | <b>Session VII – Oral communications II</b>  |                                   |
| 13:00                    | <b>Chairpersons:</b> To be confirmed   |                                   |
| 13:00                    | <b>Lunch and Satellite Symposium</b>   |                                   |
| 14:30                    | <b>Session VIII – Unmet needs in inherited metabolic diseases</b>  |                                   |
| 15:30                    | <b>Chairpersons:</b> Paulo Gonçalves (RD-Portugal); Fátima Ventura (Infarmed); TBC   |                                   |
| 14:30                    | Unmet needs for the patient and the family   | <i>José Vilhena, Portugal</i>     |
| 14:50                    | The pharmacist and the specificities of IMD  | <i>Sara Dias, Portugal</i>        |
| 15:10                    | Reference Centres of IMD in Portugal - challenges and opportunities  | <i>Luisa Diogo, Portugal</i>      |
| 15:30                    | Discussion   |                                   |
| 15:50                    | <b>Session IX – Spotlight I</b>  |                                   |
| 16:30                    | <b>Coffee break</b>  |                                   |



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|--------------|---------------------------------|
| <b>17:00</b> | <b>Session X – Spotlight II</b> |
| <b>17:40</b> |                                 |
| <b>17:40</b> | <b>Awards and Final remarks</b> |
| <b>18:00</b> | <b>End of the Symposium</b>     |