

20th International SPDM Symposium "The changing paradigm of Inherited Metabolic Disorders in Portugal"

Scientific Programme

5th April 2024

09:45	Welcome
10:00 11:15	Session I - Lessons from NBS
10:00	<ul style="list-style-type: none"> 45 years of Newborn Screening: lessons learned <i>Laura Vilarinho, Porto, PT</i>
10:20	<ul style="list-style-type: none"> Clinical follow-up of mild diseases – paediatric experience <i>Teresa Campos, Porto, PT</i>
10:40	<ul style="list-style-type: none"> Clinical follow-up of mild diseases – adult overview <i>Arlindo Guimas, Porto, PT</i>
11:00	<ul style="list-style-type: none"> Discussion
11:15 11:45	Coffee Break and Poster View
11:45 13:00	Session II – Current and future of IEM diagnosis
11:45	<ul style="list-style-type: none"> NGS - implications for clinical diagnosis <i>Fabiana Ramos, Coimbra, PT</i>
12:05	<ul style="list-style-type: none"> New biochemical markers <i>Rafael Artuch, Barcelona, SP</i>
12:25	<ul style="list-style-type: none"> Classification of variants in the clinical laboratory: Challenges and solutions <i>Belén Pérez, Madrid, SP</i>
12:45	<ul style="list-style-type: none"> Discussion

13:00 15:00	Lunch and Poster View
15:00 16:15	Session III – New approaches to IEM treatment
15:00	<ul style="list-style-type: none"> Breastfeeding in IEM <i>Júlio Cesar Rocha, Lisboa, PT</i>
15:20	<ul style="list-style-type: none"> Modulating microbiome to treat IEM – <i>TBC</i>
15:40	<ul style="list-style-type: none"> New options for treatment in CDG <i>Mercedes Serrano, Barcelona, SP</i>
16:00	<ul style="list-style-type: none"> Discussion
16:15 16:45	Coffee Break and Poster View
16:45 18:15	Session IV - Overview of SPDM Projects
16:45	<ul style="list-style-type: none"> Improve management of MADD patients: a curated database with clinical, molecular and cellular information - <i>Bárbara Henriques, Lisboa, PT</i>
17:00	<ul style="list-style-type: none"> Melhoramento das novas tabelas de composição do teor e dos aminoácidos a utilizar como ferramenta para o tratamento das doenças hereditárias do metabolismo proteico – <i>Carla Vasconcelos, Porto, PT</i>
17:15	<ul style="list-style-type: none"> PhD project – <i>Teresa Campos, Porto, PT</i>
17:30	<ul style="list-style-type: none"> Rare diseases reference centers network knowledge management – <i>Sandra Mexia, Lisboa, PT</i>
17:45	<ul style="list-style-type: none"> Acquired Vitamin B12 Deficiency in Newborns: Positive Impact on Newborn Health through Early Detection – <i>Patrícia Lipari Pinto, Lisboa, PT</i>
18:00	<ul style="list-style-type: none"> Discussion

18:15	Awards and Final Remarks
18:45	End of the Symposium