



SCIENTIFIC PROGRAMME | 5TH APRIL

09:45	Welcome
10:00 - 11:15	SESSION I - LESSONS FROM NBS Chairs: Hugo Rocha, Anabela Oliveira
10:00	45 years of Newborn Screening: lessons learned // Laura Vilarinho, Porto, PT
10:20	Clinical follow-up of mild diseases – paediatric experience // Teresa Campos, Porto, PT
10:40	Clinical follow-up of mild diseases – adult overview // Arlindo Guimas, Porto, PT
11:00	Discussion
11:15 - 11:45	COFFEE BREAK AND POSTER VIEW
11:45 - 13:00	SESSION II – CURRENT AND FUTURE OF IEM DIAGNOSIS Chairs: Anabela Bandeira, Sara Ferreira
11:45	Classification of variants in the clinical laboratory: Challenges and solutions // Belén Pérez, Madrid, SP
12:05	NGS - implications for clinical diagnosis // Fabiana Ramos, Coimbra, PT
12:25	New biochemical markers // Rafael Artuch, Barcelona, SP
12:45	Discussion
13:00 - 15:00	LUNCH AND POSTER VIEW
15:00 - 16:15	SESSION III – NEW APPROACHES TO IEM TREATMENT Chairs: Ana Paula Leandro, Esmeralda Rodrigues
15:00	Breastfeeding in IEM // Júlio Cesar Rocha, Lisboa, PT
15:25	New options for treatment in CDG // Mercedes Serrano, Barcelona, SP
15:50	Discussion
16:10 - 16:45	COFFEE BREAK AND POSTER VIEW
16:45 - 18:15	SESSION IV - OVERVIEW OF SPDM PROJECTS Chairs: Esmeralda Martins, Manuela Grazina
16:45	Improve management of MADD patients – insights and achievements // Bárbara Henriques, Lisboa, PT
17:00	Improvement of new amino acid content tables to be used as a tool for the treatment of protein inherited metabolic disorders // Carla Vasconcelos, Porto, PT
17:15	A patient reported experience measure (PREM) for quality improvement of pediatric inherited metabolic disorders care // Teresa Campos, Porto, PT
17:30	Rare diseases reference centers network knowledge management // Sandra Mexia, Lisboa, PT
17:45	Acquired Vitamin B12 Deficiency in Newborns: Positive Impact on Newborn Health through Early Detection // Patrícia Lipari Pinto, Lisboa, PT
18:00	Discussion
18:15	AWARDS AND FINAL REMARKS
18:45	End of the Symposium