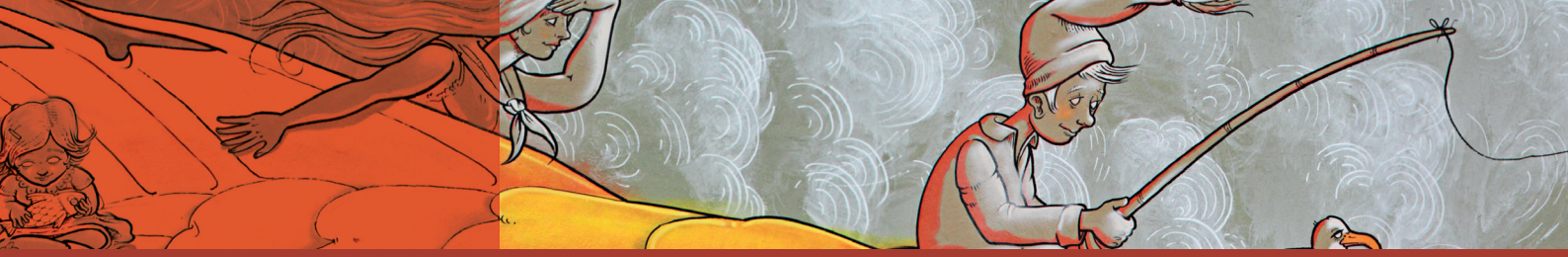

19TH INTERNATIONAL SYMPOSIUM OF THE PORTUGUESE SOCIETY FOR METABOLIC DISORDERS

THE NEXT STEPS IN INBORN ERRORS OF METABOLISM: FROM NEWBORN SCREENING TO PALLIATIVE CARE

28–30 MAR 2023

EUROSTARS OASIS PLAZA - FIGUEIRA DA FOZ - PT

PROGRAM

spdm
SOCIEDADE PORTUGUESA DE DOENÇAS METABÓLICAS

SCIENTIFIC PROGRAM

WEDNESDAY, 29TH MARCH

17:00	SPDM Nutrition Group Meeting
17:45	SPDM Group Meeting

THURSDAY, 30TH MARCH

08:30	Registration opening *Symposium Chairperson*
09:00	Symposium Opening – Welcome address on behalf of the SPDM
	SESSION I – INHERITED METABOLIC DISEASES IN THE LIFETIME *Chairpersons: Lelita Santos, Coimbra; João Durães, Coimbra; Maria João Guedes, Coimbra*
09:10	Dietary management during the lifetime for IMD - *Júlio Rocha, Lisbon*
09:30	Reproductive health and other challenges in adults with an IMD - *Elaine Murphy, London*
09:50	Neurometabolic disorders in adults: IMD and beyond - *Fanny Mochel, Paris*
10:10	Palliative care for IMD patients - *Cândida Cancelinha, Coimbra*
10:30	Discussion
10:50-11:20	Coffee break and Poster View
	SESSION II - FAMILIAL DYSLIPIDAEMIAS *Chairpersons: Ana Gaspar, Lisbon; Teresa Cardoso, Porto; Eulália Costa, Coimbra*
11:20	Defects of lipoprotein metabolism: from the genes to the clinic *Adriaan Holleboom, Amsterdam*
11:40	Familial hypercholesterolaemia in children and adolescents - better safe than sorry *Albert Wiegman, Amsterdam*
12:00	The Portuguese screening programs for familial dyslipidaemias - *Mafalda Bourbon, Lisbon*
12:20	Discussion
12:20 - 14:30	Lunch and Satellite Symposium
	SESSION III – ARE WE MOVING FORWARD IN LYSOSOMAL STORAGE DISORDERS? *Chairpersons: Esmeralda Rodrigues, Porto, Sandra Alves, Porto, Lúcia Lacerda, Porto*
14:30	The future of enzyme replacement therapy - *Eugen Mengel, Hochheim*
14:50	The path to gene therapy in lysosomal storage disorders - *Francesca Tucci, Milan*
15:10	Registries in inherited metabolic diseases: how to move forward? - *Nadia Belmatoug, Paris*
15:30	Discussion
16:00-16:30	Coffee break and Poster View
16:30	**SESSION IV – ORAL COMMUNICATIONS I** *Chairpersons: Sara Ferreira, Coimbra, Isabel Rivera, Lisbon*
17:30	**SPDM GENERAL ASSEMBLY**
19:00	**DEPARTURE TO THE SYMPOSIUM DINNER**

SCIENTIFIC PROGRAM

FRIDAY, 31ST MARCH

	SESSION V – NEW APPROACHES IN PORPHYRIAS *Chairpersons: Arlindo Guimas, Porto; Sónia Moreira, Coimbra; Eulália Costa, Coimbra*
09:00	Acute hepatic porphyria – The experience of a reference center in Brazil - *Charles Marques Lourenco, Ribeirão Preto*
09:20	Biochemical and Molecular Update on Porphyria Diagnosis - *Filipa Ferreira, INSA Porto*
09:40	The RNAi therapeutics revolution: from bench to bedside and back again - *Pedro Moreno, Porto*
10:00	Discussion
10:30-11:00	Coffe Break and Poster View
	SESSION VI – THINKING BIG AND BOLD IN INHERITED METABOLIC DISEASES *Chairpersons: Laura Vilarinho, Porto; Ana Cristina Ferreira, Lisbon; Anabela Oliveira, Lisbon*
11:00	Challenges and Opportunities for using of real world data in metabolic medicine - *Mário Silva, Lisbon*
11:20	Newborn screening by WGS: opportunities and challenges - *David Bick, London*
11:40	Aging in IMD: uncovering new phenotypes - *Charles Lourenço, Ribeirão Preto*
12:00-12:20	Discussion
	SESSION VII – ORAL COMMUNICATIONS II *Chairpersons: Joana Salgado, Coimbra; João Gomes, Coimbra*
13:00-14:30	Lunch
	SESSION VIII – UNMET NEEDS IN INHERITED METABOLIC DISEASES *Chairpersons: Paulo Gonçalves, Lisbon; Fátima Ventura, Lisbon; Rui Tato Marinho, Lisbon*
14:30	Unmet needs for the patient and the family - *José Vilhena, Coimbra*
14:50	The pharmacist and the specificities of IMD - *Sara Dias, Lisbon*
15:10	Reference Centres of IMD in Portugal - challenges and opportunities - *Luisa Diogo, Coimbra*
15:30-15:50	Discussion
	SESSION IX – SPOTLIGHT I *Chairpersons: M. Helena Santos, Vila Nova de Gaia; Nanci Batista, Coimbra*
15:50	Diet quality and saproterin dihydrochloride (BH4) use in children with phenylketonuria (PKU) - *Maria Inês Gama, Lisbon*
16:10	Novel insights into treatment strategies for hyperammonemia-associated urea cycle disorders and organic acidurias. - *Margarida Silva, Lisbon*
16:30-17:00	Coffee Break and Poster View
	SESSION X – SPOTLIGHT II *Chairpersons: Paulo Castro Chaves, Portugal; Paula Leandro, Lisbon*
17:00	Something stinks': impaired hydrogen sulfide and cysteine persulfide production by cystathionine β-synthase variants identified in classical homocystinuria patients - *João Vicente, Lisbon*
17:20	Improve management of MADD patients: a curated database with clinical, molecular and cellular information - *Bárbara Henriques, Lisbon*
17:40	**AWARDS AND FINAL REMARKS**
18:00	**END OF THE SYMPOSIUM**

ORAL COMMUNICATIONS

THURSDAY, 30TH MARCH

16h30 – 17h30 | Session IV

OC 01	Impact of structural GLA protein changes on peripheral GLA activity and substrate accumulation in Fabry disease patients
OC 02	MCADD patients: how to face cardiac function biomarkers during acute episodes?
OC 03	Help comes from unexpected places: how a tiny fairy and a tropical fish may help us model Mucopolysaccharidoses
OC 04	Clinical and laboratory findings in Glycogen Storage Disease Type V: results from a retrospective observational study in a tertiary hospital
OC 05	Glutaric acidemia type 1: diagnosis, clinical features, and outcome in a Portuguese cohort
OC 06	Forty-three Years After The Start Of Neonatal Screening In Portugal: The Results of a Retrospective Cohort Study with 113 Adult PKU Patients

FRIDAY, 31TH MARCH

12h20 – 13h00 | Session VII

OC 07	The hole in the whole: mitochondrial DNA deletions screening
OC 08	Restoring cholesterol homeostasis in neurons by AAV-mediated CYP46A1 delivery is not sufficient to stall the progression of Niemann-Pick type C disease
OC 09	Palliative care in children with inherited metabolic diseases: why does it matter?
OC 10	Boosting insights on the immunopathology of PMM2-CDG

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