



21 e 22 Março 2013

Local:

Hotel Quinta das Lágrimas › Coimbra

IX International
Symposium
– Sociedade
Portuguesa
de Doenças Metabólicas ‹
SPDM

Coimbra, 21 and 22 March 2013



IX International Symposium of the Sociedade Portuguesa de Doenças Metabólicas - SPDM

Coimbra, 21 and 22 March 2013

Identifying disorders, modifying phenotypes

21 March

Identifying disorders

09:00-09:20	Open session	Direção da SPDM
09:20-11:15	From the bench to clinics and back	Manuela Grazina, Dulce Quelhas
09:20-09:40	Diagnosis of primary and secondary deficiencies of CoQ	Rafael Artuch Iriberry
09:40-10:00	Laboratorial study of polyols metabolism	Rúben Ramos
10:00-10:20	What is new about CDGs?	Gert Matthijs
10:20-10:40	Bile acid metabolism disorders	Peter Clayton
10:40 -11:00	Disorders of phospholipids and sphingolipids biosynthesis	Foudil Lamari
11:00 -11:15	Discussion	
11:15-11:45	Coffee-break	
11:45-13:00	Newborn screening	Ana Gaspar, Hugo Rocha
11:45- 12:05	Changing the incidence of IEM in Portugal after expanded NBS	LauraVilarinho
12:05-12:25	Screening for attenuated diseases - Pros and Cons	Esmeralda Martins
12:25-12:45	Screening LSDs -Pros and Cons	Roberto Giuliani
12:45-13:00	Discussion	
13:00-14:00	Lunch	
14:00-15:30	Inborn metabolic diseases in adults	Lélita Santos, Teresa Cardoso
14:00-14:20	The eye in adult IEM	Eduardo Silva
14:20-14:40	Treatable metabolic psychoses	Frederic Sedel
14:40-15:00	Acute porfrias	Charles Lourenço
15:00-15:15	Discussion	
15:15-15:45	Coffee-break	
15:45- 17:00	Free communications/ Poster presentations	M. Carmo Macário, Margarida Silva
17:00-18:30	SPDM Society Meeting	
20:00	Dinner	

22 March

Modifying phenotypes

8:30-10:00	Enzyme replacement therapy	Lúcia Lacerda, Sílvia Sequeira
8:30-08:50	ERT in Fabry disease	Charles Lourenço
08:50-09:10	Long term follow up in Gaucher type I disease	Carla Hollak
09:10-09:30	Enzyme replacement therapy in MPS IV- Preliminary data of MOOR-004	Paul Harmatz
09:30-09:45	Enzyme replacement therapy in Portugal - an overview	Ana Fortuna
09:45- 10:00	Discussion	
10:00- 11:00	Cell & organ transplantation	Emanuel Furtado, Isabel Gonçalves
10:00 -10:30	Hepatocytes and liver transplantation in inborn metabolic disorders	Etienne Sokal
10:30 -10:45	Liver transplantation in inborn metabolic disorders – the Portuguese experience	Sandra Ferreira
10:45- 11:00	Discussion	
11:00-11:30	Coffee break	
11:30- 12:45	Nutrition and supplementation in inborn metabolic disorders	Manuela Almeida, Rita Ferreira
11:30-11:50	Nutritional assessment	Júlio Rocha
11:50-12:10	The need of amino acid supplementation when protein substitutes are already given	Françjan van Spronsen
12:10-12:30	Essential fatty acids supplementation	Pablo Sanjurjo
12:30-12:45	Discussion	
12:45- 14:00	Lunch	
14:00-15:30	Small molecules therapy	Elisa Leão Teles, Paula Leandro
14:00-14:20	Genistein in Sanfilippo disease	Mercedes Pineda
14:20-14:40	Sapropterin in PKU	Amaya Bélanger-Quintana
14:40-15:00	Substrate inhibitor therapy in NP-C disease	Mercedes Pineda
15:00-15:15	Carglumic acid in the treatment of hyperammonemia	Pablo Sanjurjo
15:15-15:30	Discussion	
15:30- 16:00	Coffee-break	
16:00-17:15	Free communications/ Poster presentations	Patrícia Janeiro, xxx
17:15-17:30	Awards and scholarships delivering	Direção da SPDM
17:30	Closing remarks and farewell	Direção SPDM

Symposium registration:

	Until March 3 rd	After March 3 rd
Member SPDM	125.00 € □	150.00 € □
Non-member	150.00 € □	175.00 € □
Student*	100.00 € □	125.00 € □

The Symposium Registration includes:

Access to the Symposium sessions and documentation, coffee breaks, lunches and Symposium dinner

<http://www.asic.pt/index.php/formularios>