IX International Symposium – Sociedade Portuguesa de Doenças Metabólicas – SPDM

Identifying disorders, modifying phenotypes

21st March — Identifying disorders

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

22nd March — Modifying phenotypes

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

Curso Satélite 23 de março 2013

MPS and the Heart Meeting

Doença Cardíaca nas Mucopolissacaridoses

Consulte http://www.scomunicacao.com/heartmeeting/