29 October 2009

Mitochondrial Respiratory Chain Diseases
Chairs: Luisa Diogo, Manuela Grazina

09:00 Diagnostic Work-up of Mitochondrial Cytopathies - Guidelines (F. Scaglia)
09:20 Coenzyme Q10 Deficiency and Mitochondrial Disorders (R. A. Iriberri)
09:40 Hepatic mtDNA Depletion Syndromes in Childhood (F. Scaglia)
10:10 Progress and Prospects in Therapy for Mitochondrial Diseases (R. W. Taylor)
10:30 **Coffee-break**

Inborn Errors of Metabolism and Epilepsy
Chairs: Ana Gaspar, Conceição Robalo

11:00 Sodium Valproate – Therapeutic Aspects (C. F. Ribeiro)
11:20 Sodium Valproate and Inborn Errors of Metabolism (M. Silva)
11:40 Cerebral Folate Deficiency: Cause or Consequence of Disease? (R. A. Iriberri)

12:00 **Welcome Ceremony**
Rui Batista, Catarina Oliveira, Elisa Leão Teles, Luisa Diogo
12:30 **Lunch**

Outcome and Quality of Life in…
Chairs: Margarida Leite, Sandra Paiva

13:30 Galactosemia (A. Bosch)
14:00 Glycogen Storage Disease Type 1 (A. Cabral)

Aminoacid Catabolism Disorders: Therapeutic Approaches
Chairs: Isabel Gonçalves, Jorge Salles Marques

14:20 Orthotopic Liver and Hepatocyte transplantation (E. Sokal)
14:50 Current role of Carglumic Acid in Hyperammonaemia (V. Vassili)
15:20 Sapropterin Dihydrochloride in Phenylketonuria (A. Burlina)
15:50 **Coffee-break**

16:15 **Free Communications**
Chairs: Esmeralda Rodrigues, Hugo Rocha

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The State of the Art…
Chairs: Isabel Tavares de Almeida, Maria Carmo Macário

09:00 Defects in Polyol Metabolism (C. Jakobs)
09:30 Neurotransmitter Disorders (A. G. Carzola)
10:00 Inborn Errors of Metabolism in Adult Neurology (F. Sedel)
10:30 **Coffee-break**

11:00 **The State of the Art…**
Chairs: Esmeralda Martins, Lígia Almeida

11:00 D and L 2-Hydroxyglutarica Acidurias (C. Jakobs)
11:20 Citrin Deficiency (F. Scaglia)

11:40 **Oral Posters Presentation**
Chairs: Paula Garcia, Dulce Quelhas
13:00 **Lunch**

Advances in Lysosomal Storage Diseases
Chairs: Clara Sá Miranda, Sílvia Sequeira

14:00 Clinical Trials in Mucopolyssacharidosis II (P. Harmatz)
14:20 Guidelines in the Follow-up of Mucopolyssacharidosis (M. Beck)
14:40 Stem Cell Transplantation in Lysosomal Disorders (J. J. Boelens)
15:10 Miglustat in Niemann-Pick C Disease (M. Pineda)
15:40 **Coffee-Break**

Inborn Errors of Metabolism as Rare Disorders
Chairs: Rui Vaz Osório, Laura Vilarinho

16:00 Portuguese National Plan for Rare Diseases (L. Nunes)
16:30 Clinical and Laboratorial Training (A. Cabral, I. Tavares de Almeida)
17:00 Reference Centres (E. Leão Teles)
17:20 Patient Associations Perspectives
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17:50 **Closing Remarks**
Elisa Leão Teles, Paula Garcia

VI SIMPÓSIO INTERNACIONAL
International Symposium
Sociedade Portuguesa de Doenças Metabólicas