Sociedade Portuguesa de Doenças Metabólicas

VII INTERNATIONAL SYMPOSIUM

4 - 5 November 2010 | Algarve, Portugal

4 November 2010

09:00 **Symposium Opening - Welcome Address on behalf of the SPDM**
(I. Tavares de Almeida, Lisboa, PT)

Session I - Homocystinuria / Creatine Syndromes / Methylmalonic Aciduria
Chairpersons: *I.Tavares de Almeida; M. Duran*

09:15 Disorders of intake, absorption and transport of vitamin B12 (M. Schiff, Paris, FR)
09:40 Remethylation disorders – homocystinuria: how to treat (C. Dionisi-Vici, Rome, IT)
10:05 Creatine deficiency syndromes (G. Salomons,Amsterdam, NL)
10:30 **Coffee Break**
11:00 Methylmalonic aciduria: outcome of Mutase, CblA and CblB deficiency (S. Grünewald, London, UK)
11:25 Methylmalonic aciduria – Sucla 2 and Epimerase (C. Dionisi-Vici, Rome, IT)
11:50 New challenges for the treatment of Methylmalonic Aciduria (B. Pérez, Madrid, ES)
12:15 **Poster View**
13:00 **Lunch**

Session II - Metabolic Disorders: Clues in Unsolved Cases
Chairpersons: E. Leão Teles (Porto, PT); C. Mendonça (Faro, PT)

14:00 Dysmorphism and metabolic disease (S. Grünewald (London, UK)
14:25 MRI patterns and metabolic diseases (J. Gärtner, Gottingen, DE)
14:50 Hypotonic babies and IEM (B. Plecko, Graz, AT)
15:15 Neonatal seizures in IEM (J. Campistol, Barcelona, ES)
15:40 **Coffee Break**

Session III – Oral Communications
Chairpersons: A. Gaspar (Lisboa, PT); P. Leandro (Lisboa, PT)

16:15 Mevalonic aciduria and congenital CMV infection: clinical challenge in diagnosis and management (S. Soares, Porto, PT)
16:30 High frequency of Methionine Adenosyltransferase (MAT) I/III deficiency in newborn screening due to a single dominant mutation (A. Marcão, Porto, PT)
16:45 mtDNA copy number mutations screening in Portuguese patients: results of the first year (M. J. Santos, Coimbra, PT)
17:00 Infantile-onset disorders of cross-talk between nuclear and mitochondrial genomes (C. Nogueira, Porto, PT)
17:15 Liver failure due to deoxyguanosine linase deficiency in four Portuguese patients (F. Silva, Coimbra, PT)
17:30 Chemical chaperoning of a mutant form of PDH E1α protein: the effect of L-arginine (P. Leandro, Lisboa, PT)

17:45 **LATE-BREAKING NEWS**
Variant MADD due to riboflavin transporter defect presenting as Brown-Vialetto-van Laere Syndrome in three patients (M. Duran, Amsterdam, NL)

18:00 **Poster View**

5 November 2010

Session IV – Oral Communications
Chairpersons: S. Sequeira (Lisboa, PT); E. Rodrigues (Porto, PT)

09:00 Clinical outcomes of long-term enzyme replacement therapy for 6 children with Gaucher disease (P. J. Nóbrega, Porto, PT)
09:15 A novel pathogenic variant of the human medium-chain acyl-CoA dehydrogenase (MCAD): Gly377Val (F. Ventura, Lisboa, PT)
09:30 Short-chain-hydroxyacyl-CoA dehydrogenase deficiency: the clinical relevance of an early diagnosis and report of four new cases (E. Martins,Porto, PT)
09:45 Analysis of evolution of Tyrosinemia type 1 patients treated with Nitisinone - a single centre experience: a deeper face of iceberg (T. Campos, Porto, PT)
10:00 The formation of human phenylalanine hydroxylase heteromeric proteins (J. Leandro, Lisboa, PT)
10:15 Functional recovery of 6-pyruvoyl-tetrahydropterin synthase deficiency by antisense therapy (S. Brasil, Madrid, ES)
10:30 **Coffee Break**

Session V – New Challenges in Old Disorders
Chairpersons: E. Martins (Porto, PT); I. Rivera (Lisboa, PT)

11:00 BH4 treatment in PKU: Selection of the patients and importance of the genotypes (N. Blau, Zurich, CH)
11:25 BH4 treatment in PKU: what we know now? (A. Bélanger-Quintana, Madrid, ES)
11:50 Classical Galactosemia: new approaches to old problems (J. Fridovich-Keil, Atlanta, USA)
12: 15 Tyrosinemia type I: the past, the present, the future (F. van Spronsen, Groningen, NL)
12:40 **Lunch**

Session VI – Lysosomal Diseases
Chairpersons: C. Sá Miranda (Porto, PT); P. Garcia (Coimbra, PT)

13:45 Niemann-Pick Disease type C: from diagnosis to treatment (M. Pineda, Barcelona, ES)
14:10 Treatment of children and adults with Pompe Disease (J. de Vries, Rotterdam, NL)
14:35 Gaucher Disease: new advances in therapy (T. Cox, Cambridge, UK)
15:00 Fabry Disease: early diagnosis to therapy (U. Ramaswami, Cambridge, UK)
15:25 **Coffee Break**

Session VII - Poster Presentations
Chairpersons: L. Diogo (Coimbra, PT); L. Vilarinho (Porto, PT)

16:00 Short Oral e-poster presentations

17:00 **Awards / Closing Remarks**

17:15 **End**