22 November 2007

09h15 Welcome

Screening in inherited metabolic diseases (IMD): new perspectives
Chairs: Filomena Eusébio, Paula Garcia
09h30 Neonatal screening: recent advances (L. Vilarinho)
09h50 *Screening of lysosomal storage diseases (LSDs) (C. Sá Miranda)*
10h10 Discussion

Inherited metabolic diseases in adults
Chairs: P. Sanjurjo, Caldeira Ferreira
10h20 Aging with IMD (A. Cabral)
10h45 IDM: adult onset (J. Walter)
11h10 Discussion
11h20 ***Coffee break***

New approaches for "old diseases"
Chairs: I. T. Almeida, E. Rodrigues
11h50 *Tyrosinemia type I (P. Mckiernan)*
12h15 *Aminoacidophaties and organic aciduries (J. Walter)*
12h40 *Discussion*
12h50 **Lunch and poster viewing**
14h30 Overview: apoptosis and neurodegenerative diseases (R. Boustany)

Peroxisome and peroxisomal diseases (PD)
Chair: M. Lemos, A. Gaspar
15h05 Peroxisome biogenesis (J. Azevedo)
15h25 PD: Clinical and diagnostic aspects (J. Berger)
15h50 Discussion.
16h00 **Coffee break**
16h30 **Free communications**
Chairs: M. Silva, E. Martins
16h30 **Sattelite meeting: nutrition and dietary treatment in IMD** (A. McDonald, M. Pons)
Chairs: A. Guerra, A. Baldellou

23 November 2007

Endosome lysosome system
Chairs: M. João Saraiva, C. Sá Miranda
09h00 Intracellular traffic (M. Seabra)
09h25 LSDs and "new" biomarkers (J. Aerts)

Neuronal ceroid lipofuscinosis
Chairs: S. Carpenter, G. Ribeiro
09h50 Pathophysiology and molecular biology (T. Braulke)
10h15 Clinical and diagnostic aspects (A.Jalanko)
10h40 Discussion
10h50 **Coffee break and poster viewing**

LSDs: new therapeutical perspectives
Chairs: G. Pintos, S. Sequeira
11h20 Enhancement therapy: an overview (E. Schuchman)
11h50 Stem cells transplantation and gene therapy: an overview (T. Cox)
12h20 Discussion
12h30 **Lunch and poster viewing**

LSDs- enzyme replacement therapy and substrate deprivation: problematic organs and therapeutic pitfalls
Chairs: T. Cox, E. Leão Teles
Bone
14h00 Outcome in Gaucher disease (A.M. Martins, M. Rocca)
14h30 Outcome in MPS II and MPS VI Therapies (P. Harmatz)
Central nervous system
14h50 Outcome in Sphigolipidoses and in MPS (M. Pineda)
Kidney
15h10 Outcome in Fabry disease (J. P. Oliveira)
Heart
15h30 Outcome in Fabry disease (J. L. Zamorano)
Skeletal muscle
15h50 Outcome in Pompe disease (S.Carpenter, M. Deschauer)
16h20 New Biomarkers of Mucopolysaccharidoses (C. Simonaro)
16h40 *Discussion*
17h15 **Coffee break and poster viewing**
17h30 **Free communications**
Chairs: L. Diogo, M. Luís Cardoso

24 November 2007

Ethical problems and inherited metabolic diseases
Chairs: L. Aldamiz-Echevarria, A. Cabral
09h00 Genetic diseases and ethical problems (A.S. Carvalho)
IMD: reference centres organization
Chairs: A. Ferreira, J. Lavinha
09h30 European Community perspectives (J. Lavinha)
09h50 Perspectives of Portuguese Health Care Authorities (L.Nunes)
10h10 **Role of the scientific and patients association in IMD reference centers organization**

Scientific associations:
Society of IMD in Portugal (A. Cabral)
Societies of IMD in Spain (D. Lamuño, M. I. Couce)
Society of IMD in Brazil (A. M. Martins)

Portuguese patients associations:
APL (F. Beirão)
APOFEN (R. Barros)
Raríssimas (P. Costa)
10h50 Discussion
11h15 **Coffee break**
11h30 **Prizes: communications and posters**
International panel of judges
12h00 Closing remarks
12h30 SPDM General Assembly

V Symposium- Portuguese Society of Metabolic Disorders

New and Controversial Perspectives on Metabolic Diseases