

4th November 2004

09:30 **Welcome and opening session**

First session

Chairperson: Clara Sá Miranda

10:00 Sphingolipids and Sphingolipidoses: Metabolism at Membrane Water Interface (K. Sandhoff, Bonn)

10:40 Lysosomal Storage Disorders: New Therapeutics Strategies

(T. Cox, Cambridge)

11:00 **Pausa para café / Coffee Break**

Second session

Chairpersons: M de Jesus Feijó / Olga Amaral

11:30 Gaucher Disease: Ten Years of Enzyme Replacement Therapy Experience (N. Belmatoug, Paris)

11:50 Gaucher Disease: First Experience with Substrate Reduction Therapy in Spain (P. Giraldo, Zaragoza)

12:10 Biomarkers Role in the Diagnosis and Management of Gaucher Disease (H. Aerts, Amsterdam)

12:30 Discussion

12:45 **Lunch**

14:00 **Oral communications: Gaucher Disease**

Chairpersons: Teresa Tasso / Fátima Ferreira

Third session

Chairpersons: João P Oliveira / M João Pais

15:00 Fabry Disease as a Model for a Systemic Vasculopathy (R. Schiffmann, Bethesda)

15:30 Enzyme Replacement Therapy of Fabry Disease (W. Wilcox, LA)

15:50 Discussion

16:05 **Oral communications: Fabry Disease**

Chairpersons: Idalina Beirão / Jorge Dickson

16:30 **Coffee Break**

Fourth session

Chairpersons: Maria J Saraiva / Sílvia Sequeira

17:00 Niemann-Pick Type C Pathogenic Mechanisms and Implications in Therapeutic Approaches (M. Vanier, Lyon)

17:20 The Niemann-Pick type C Disease in Spain: Development of a Clinical Database, a Disability Scale and a New Protocol to Evaluate Substrate Reduction Therapy (M. Pineda, Barcelona)

17:35 Substrate Reduction Therapy for Glycosphingolipid Storage Disorders of the Brain (R. Lachmann, Cambridge)

17:55 GM2 Gangliosidosis B1 Variant in Portuguese Population: Phenotype / Genotype Correlation, (M. Maia, Porto)

18:10 Discussion

5th November 2004

First session

Chairpersons: Elisa Leão Teles / Paula Garcia

09:00 Mucopolysaccharidosis Epidemiology (J. Hopwood, Adelaide)

09:30 MPS I – Hurler Disease: Enzyme Replacement Therapy Experience (J. Muenzer, Chapel Hill)

09:50 MPS VI – Maroteaux-Lamy: Phase III Clinical Trial Results (P. Harmatz, Oakland)

10:10 Enzyme Replacement Therapy of MPS II – Hunter Disease – Phase III Clinical Trial Results (J. Muenzer, Chapel Hill)

10:30 Pompe Disease: Enzyme Replacement Therapy (H. Mandel, Haifa)

10:50 Discussion

11:05 **Coffee Break**

11:30 **Oral communications: MPSs and Pompe Disease**

Chairpersons: Henriqueta Marques, Jorge Marques

12:30 **Almoço / Lunch**

Second session

Chairpersons: Mónica Sousa / Ana Gaspar

14:00 Niemann - Pick A and B: an Overview, Animal Models and New Therapies (E. Schuchman, New York)

14:40 Morquio A animal Model and Enzyme Replacement Therapy (S. Tommatsu, Missouri)

15:00 Discussion

15:15 **Coffee Break**

Third session

Chairpersons: P Moradas Ferreira, Laura Vilarinho

15:45 Endosomes Related Organelles Biogenesis and Genetics Disorders (J. Bonifacino, Bethesda)

16:15 Signal Transduction Defects, Emerging Causes of Genetic Encephalopathy. (J. Jaeken, Leuven)

16:45 Discussion

Fourth session

Chairpersons: Isabel Tavares de Almeida, Clara Sá Miranda

17:00 Early Diagnosis of Lysosomal Storage Disorders (J. Hopwood, Adelaide)

17:30 Tandem Mass Spectrometry Applications in Lysosomal Storage Disorders Screening (P. Meikle, Adelaide)

17:50 Newborn Screening Using Enzymatic Assays in Blood Spots (N. Chamoles, Buenos Aires)

18:10 Discussion

6th November 2004

First session

Chairpersons: Isabel Azevedo / Elisa L. Teles

09:00 Medical Education in Inherited Metabolic Diseases – a Challenge (P. Sanjurjo, Bilbao; A. Cabral, Lisboa)

09:45 **Oral communications: Metabolic Diseases in General**
Chairpersons: Aguinaldo Cabral / Isabel Rivera

10:45 **Coffee Break**

11:15 **Round table**

Chairpersons: L. Diogo (Coimbra), L. Aldamiz-Echevarria (Bilbao)

Ethical Legal and Social Aspects Related with Early Diagnosis and Therapy of Inherited Metabolic Diseases

- Ethical Aspects. A. Quintanilha

- Social and Legal Aspects. Maria de Belém Roseira

- Position of the Ministry of Health. (Portuguese Health Ministry delegate)

- Role of Social Assistant. L. Almeida (H. Stª Maria)

- Role of the Patients Association. (Member of the Portuguese Association of LSD Patients)

12:45 **Closing session and Awards presentation for the best communications (oral and poster)**