Sociedade Portuguesa de Doenças Metabólicas

VIII Internacional Symposium SPDM
3-4 November 2011 - Porto
Fundação Cupertino de Miranda

& 1º Encontro Luso-Brasileiro e de Outros Países de Lingua Oficial Portuguesa
1st Luso-Brazilian and Other Countries of Portuguese Official Language Meeting - 5th November

Inborn Errors of Metabolism and Neurodegeneration

NOVEMBER 3rd

08:00	Registration
09:00	Symposium Opening - Wellcome Adress
09:15	Opening lecture: IMD and neuropathology - *J. Campistol, Barcelona*

Session I - Amino acids and cerebral organic acidurias neuropathology

09:55	D-2 and L-2-hydroxyglutaric aciduria: What is new? - *C. Jakobs, Amsterdam*
10:20	Glutaric aciduria type I - Pathomechanisms of neurodegeneration in glutaryl-CoA dehydrogenase deficiency - *S. Kolker, Heidelberg*
10:45	Role of neuroprotectors on neurochemical changes observed in hyperprolinemic rats - *A. T. Wyse, Porto Alegre*
11:10	Neurological signs in hyperammonemias - *C. Dionisi-Vici, Rome*
11:40	Coffee Break and Poster view

Session II - Mitochondrial disorders and neurodegeneration

12:10	Mitochondrial medicine - S. *DiMauro, New York*
12:35	Mitochondrial disorders and Alzheimer disease - *C. Oliveira, Coimbra*
13:00	Lunch

Session III - Neuropathomecanisms in complex molecules diseases

14:30	Neuropathology in congenital disorders of glycosylation - *J. Jaeken, Leuven*
14:55	ERAD of mutant glucocerebrosidase and its implications in Gaucher disease - *M. Horowitz, Telavive*
15:20	Antioxidans halt axonal degeneration and disability in X- adrenoleucodystrophy mouse model: towards a clinical trial - *S. Fourcade, Barcelona*
15:45	Ether-phospholipid deficiencies: from human disorders to mouse models - *P. Brites, Porto*
16:10	Coffee Break and Poster view

Session IV - Oral communications

16:40	Oral Communications
19:40	Dinner

Organizing Committee
Elisa Leão Teles | Clara Sá Miranda | Laura Vilarinho | Jorge Azevedo | Esmeralda Rodrigues | Esmeralda Martins

Scientific Committee
Elisa Leão Teles | Clara Sá Miranda | Isabel Tavares de Almeida | Ana Gaspar | Paula Garcia

more information: www.spdm.pt

Secretariat: IBMC.INEB Event Management Office | Antonieta Correia | acorreia@ibmc.up.pt |

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NOVEMBER 4th

Session V - Oral communications

09:00 Oral communications and Late Break News

Session VI - Neuropathic lysosomal storage disorders – from storage to cellular damage

10:15 Dissecting neuronal degeneration: NCL from genes to function - *T. Braulke, Hamburg*

10:40 Metachromatic leukodystrophy: consequence of sulphatide accumulation and therapeutic perspectives - *V. Gieselmann, Bonn*

11:05 Coffee Break and Poster view

Session VII – Neuropathic LSD – new insights in diagnosis, treatment and research

11:35 Mucopolysaccharidosis type I: Cell, Protein and Gene Therapy - *J. Tolar, Minnesota*

12:00 Mucolipidosis type II: molecular changes in the brain of a novel mouse model - *T. Braulke, Hamburg*

12:25 Neuroimaging in IMD: magnetic resonance pattern and spectroscopy - recognition in white matter disease - *M. van der Knaap, Amsterdam*

13:00 Lunch

Session VIII - Inherited Metabolic Diseases: important diagnosis clues

14:30 Neurological presentation of IMD in adult - *J.M. Saudubray, Paris*

14:55 Niemann Pick type C: from diagnosis suspicion to treatment monitoring - *C. Lourenço, Ribeirão Preto*

15:20 Closing lecture: Exome sequencing will change our life - *R. Wevers, Nijmegen*

15:45 Coffee Break and Poster view

Session IX - Professional and patient associations meeting

16:15 Patient in first place/O doente em primeiro lugar - *A. Nunes/P. Silva, Porto*

17:00 Discussion

17:15 Awards. Closing remarks

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NOVEMBER 5th - 1st Luso-Brazilian and Other Countries of Portuguese Official Language Meeting

Doenças Hereditárias do Metabolismo - Do diagnóstico, do tratamento e da investigação - que desafios

- Programas de Rastreio Neonatal

- Doenças de Sobrecarga Lisossomal: do diagnóstico ao acompanhamento do doente

- Os caminhos da Investigação na área das DHM

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