Portuguese Neurological Society / Portuguese Society for Metabolic Disorders

Neurologic presentations of Metabolic Disorders

Formação pós graduada

Coimbra, 20 april 2018

Main auditorium, Coimbra Hospital and Universitary Centre

Registrations: SPDM (www.spdm.org.pt/) and SPN (www.spneurologia.com) sites

9:45 ***Registration opening***

10:00 ***Opening session***
Maria Carmo Macário (Course Coordinator), Neurology Department/Reference Centre for Inborn Errors of Metabolism, Coimbra Hospital and Universitary Centre, Coimbra, Portugal.

10:10 – 11:30 ***Leukodystrophies and Vascular disorders***

10:10 ***Leukodystrophies associated with inborn error of metabolism and other genetic leukoencephalopathies***
Vanishing white matter disease | Metachromatic leukodystrophy
Adult-Onset Autosomal Dominant Leukodystrophy with Autonomic Symptoms | Hereditary diffuse leukoencephalopathy with spheroids
Ettore Salsano, Unit of Neurodegerative and Neurometabolic Rare Diseases, IRCCS Foundation "Carlo Besta" Neurological Institute, Milan, Italy.

10:45 ***Metabolic causes of Vascular disorders***
Hyperhomocystinemia | Fabry disease
Miguel Viana Baptista, Neurology Department, Egas Moniz Hospital, West Lisbon Hospital Centre, Lisbon, Portugal.

11:20 ***Discussion***

11:30 ***Coffee Break***

11:45 – 13:00 ***Encephalopathy and Epilepsy***

11:45 ***Metabolic causes of Encephalopathy***
Intoxication syndromes (e.g. urea cycle disorders)
Mitochondrial encephalomyopathy, latic acidosis and stroke-like episodes (MELAS)
Francisco Sales, Neurology Department, Coimbra Hospital and Universitary Centre, Coimbra, Portugal.

12:20 ***Metabolic causes of Epilepsy***
Ceroid lipofuscinoses | Cerebral creatine deficiency
Myoclonic epilepsy with ragged red fibers (MERRF)
Francisco Sales, Neurology Department, Coimbra Hospital and Universitary Centre, Coimbra, Portugal.

12:50 ***Discussion***

13:00 – 14:30 ***Lunch***

14:30 - 16:15 ***Movement disorders and Spastic paraparesis***

14:30 ***Metabolic causes of Movement disorders***
Gaucher disease | Niemann-Pick type C | Biotin-responsive basal ganglia disease
Maria Carmo Macário, Serviço de Neurologia/Centro de Referência de Doenças Hereditárias do Metabolismo, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal.

15:15 ***Adrenomyeloneuropathy, Krabbe disease, Cerebrotendinous xanthomatosis, and other rare metabolic disorders***
Krabbe disease | Cerebrotendinous xanthomatosis
X-linked Adrenoleukodystrophy
Ettore Salsano, Unit of Neurodegerative and Neurometabolic Rare Diseases, IRCCS Foundation "Carlo Besta" Neurological Institute, Milan, Italy.

16:00 ***Discussion***

16:15 ***Coffee Break***

16:30 - 17:20 ***Neuromuscular disorders***
Pompe disease |Mitochondrial Respiratory chain disorders
Anabela Matos, Neurology Department, Coimbra Hospital and Universitary Centre, Coimbra, Portugal.

17:20 - 17:40 ***Sample collection in Metabolic disorders***
Sample collection, storage and transport | Emergency sample collection
João Durães, Neurology Department/Reference Centre for Inborn Errors of Metabolism, Coimbra Hospital and Universitary Centre, Coimbra, Portugal.

17:40 - 17:50 ***Evaluation***

17:50 – 18:00 ***Take-home message and Closing Remarks***
Maria Carmo Macário, Neurology Department/Reference Centre for Inborn Errors of Metabolism, Coimbra Hospital and Universitary Centre, Coimbra, Portugal.