EPNS Research Meeting: Alicante: 26-27 October 2018

Neurometabolic working group; Friday 26th October 2018; Block 2; 17.00-20.00

17.00-17.20
Natalia Juliá (Hospital Sant Joan de Déu, Barcelona, Spain)
New aspects of neurotransmitter defects

17.20-17.40
Heidy Suriel Baide (Vall d´Hebron Research Institute, Barcelona, Spain.)
Basal ganglia neurodegeneration in children, including mitochondrial encephalopathies.

17.40-18.00
Holger Prokisch (Helmholtz Institute, Munich, Germany)
Exome of 1500 Mitopathy patients

18.00-18.20
Francesca Nardecchia (Child Neurology and Psychiatry, Sapienza University, Rome, Italy)
Study of brain vulnerability to Phe in subjects with early treated PKU: assessment of higher cortical functions and cerebral connectivity with respect to blood and brain Phe

18.20-18.40
Sabine Jung-Klawitter (University Children´s Hospital, Heidelberg, Germany)
iPS cell-derived neuronal cells as a model to study neurometabolic diseases

18.40-19.00
Matthias Zielonka (University Children´s Hospital, Heidelberg)
Why zebrafish is a suitable model to study the mechanism of hyperammonemic encephalopathy?

19.00-19.20
Saskia Wortmann (Univ. Children´s Hospital, Salzburg; Helmholtz Institute Munich, Germany)
Succesful treatment with ketogenic diet in pediatric patients based on exome sequencing results

19.20-19.40
Johanna Uusimaa (University Hospital Oulu, Finland)
NHLRC2 variants associated with Fibrosis, Neurodegeneration, and Cerebral Angiomatosis (FINCA) – Characterisation of a novel multiorgan disease

19.40-20.00
wrap-up and final discussion