

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 \(Hospital Sant Joan de Déu, Barcelona, Spain\)](#)

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 \(Univ. Children's Hospital, Salzburg; Helmholtz Institute Munich, Germany\)](#)

Successful 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](#)