Erfelijke Stofwisselingsziekten Nederland

Programma ESN najaarssymposium 16 november 2017

9.30-10.00 Ontvangst en koffie

10.00 Opening door de voorzitter

10:05-11:15 New mechanisms

Voorzitter Dr. Monique de Sain-van der Velden, UMCU

10.05-10.30 Upstream SLC2A1 translation initiation causes GLUT1 deficiency syndrome
Dr. Erik-Jan Kamsteeg, Radboudumc

10.30-10.55 Deficiency of peroxisome-ER tether protein ACBD5 causes a defect in peroxisomal very long chain fatty acid metabolism.

Prof. dr. Hans Waterham, AMC

10.55-11.20 Living on the edge: from computational model to MCADD patient

Prof. dr. Barbara Bakker, UMCG

11.20-11.30 Uitreiking stimuleringsbeurzen

11.30-11.45 Pauze

11.45-12.45 Vergaderen

NVK, EMZ, INVEST

12.45-13.30 Lunch

13.30-14.30 New phenotypes

Voorzitter Dr. Hidde Huidekoper, ErasmusMC

13.30-13.50 A young lady with a late fetal loss and increased bile acids
Prof. dr. David Cassiman, UZ Leuven

13.50-14.10 Adenosine kinase: a rare disorder of methylation

Prof. Dr. Henk Blom, UMC Freiburg

14.10-14.30 Newborn screening reveals novel phenotypes of fatty acid oxidation disorders

Dr. Gepke Visser, UMCU

14.30-14.45 Pauze

14.45-15.30: Vrije voordrachten

Individual dietary intervention in adult patients with mitochondrial disease: the DINAMITE study, *Heidi Zweers, Radboudumc*

Hypermorphic variant in *GLS1* unveils a highly conserved genetic restrictor of glutaminase activity, *Lynne Rumping, UMCU*

Ketone ester drink boosts muscle energy balance in patients with VLCAD deficiency, *Jeannette Bleeker, UMCU*

Subjective cognitive complaints and symptoms of depression are highly prevalent in Fabry disease and are not related to objective cognitive impairment, *Simon Körver, AMC*

Afsluitende borrel