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 ACBD5 deficiency: a single peroxisomal enzyme deficiency

 *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: Abstracts**

**Afsluitende borrel**