**ESN najaarsvergadering vrijdag 18 november 2016**

**Definitief programma:**

**9.30-10.15** Ontvangst en koffie

**10.15-10.30** Opening door voorzitter

**10:30-13:00 Omics in de erfelijke metabole ziekten**

*Voorzitter Dr. Fred Vaz*

**10.30-11.00** Ervaringen met genomics

*Dr. Peter van Hasselt*

**11.00-11.30** Going beyond diagnosis...delineating pathophysiology to identify treatment targets

*Dr. Clara van Karnebeek*

**11:30-12:00 Pauze**

**12:00-12.30** Functional exome sequencing – combinatie van glycomics en genomics

*Dr. Dirk Lefeber*

**12.30-13:00** Maatschappelijke aspecten van Omics-toepassingen in de gezondheidszorg

*Prof. Martina Cornell*

**13.00-14.00** Lunch

*Vergaderingen: 13:00-14:30 NVK Start in restaurant, vanaf 13:45 Amethist zaal*

*13:45-14:30 EMZ (biochemici) Ionazaal*

**14.30-15.45** vrije voordrachten

*Voorzitter Dr. Maaike de Vries*

**14.30-14.45** Diagnostic value of untargeted metabolomics in dried blood spots

*H.A. Haijes*

**14.45-15.00** No loss of GPI-anchored leucocyte proteins in flowcytometric assay in a patient with compound heterozygote PIGN mutations

*IMLW Körver-Keulart*

**15.00-15.15** A method for functional characterization of variants in genes encoding all aminoacyl-tRNA synthetases

*Dr. M. Mendes*

**15.15-15.30** Update of combined D, L-2-Hydroxyglutaric aciduria: new cases and restoration of the defect in vitro   
*A. Pop*

**15.30-15.45** LNAA supplementation as a possible alternative treatment for adult PKU patients: evidence in PKU mice

*D. van Vliet*

Afsluitende borrel