Programma ESN najaarssymposium 22 november 2018

Locatie: Het Huis Utrecht (Boorstraat 107, 3513 SE Utrecht)

9.30-10.00 Ontvangst en koffie

10.00-10.15 Opening door de voorzitter (Prof. Nanda Verhoeven-Duif)

10:15-11:45 Diagnostic guidelines
Voorzitter: Dr. Kees Schoonderwoerd (ErasmusMC)

10.15-11:00 Nieuwe richtlijn: Diagnostiek bij kinderen met een ontwikkelingsachterstand (NVK)
Dr. Clara van Karnebeek

11:00-11.45 Best-practise voor de diagnostiek van mitochondriële aandoeningen
Dr. Richard Rodenburg

11.45-12.45 Vergaderen en discussie 'Open space' met koffie/thee
NVK, EMZ, INVEST
'Open space' voor overige aanwezigen

12.45-13.30 Lunch

13.30-13.45 Uitreiking stimuleringsbeurzen

13:45-14.30 Vrije voordrachten en korte pitches
Voorzitter: Prof. Carla Hollak (Amsterdam UMC)

13.45-14.00 Thoracolumbar kyphosis in MPS I: a natural history study and an international consensus procedure for the development of a clinical practice guideline
G Kuiper

14:00–14:30 Korte pitches (max 5 min)

Sarcopenia; physical functioning, body composition and protein intake in adult patients with mitochondrial disease compared with matched healthy controls – DYNAMO study
H Zweers

Longitudinal analysis of the effect of hematopoietic cell transplantation on ocular disease in children with Mucopolysaccharidosis I shows ongoing disease progression
BTA van den Broek

Direct-infusion based untargeted metabolomics unveils biochemical profiles of inborn errors of metabolism in cerebrospinal fluid
HA Haijes

Differential methylation in D-loop region of mitochondrial DNA isolated from muscle biopsies and skin fibroblast of myopathy patients associates with the disease
A Mposhi

Possible pathogenetic mechanism of liver failure in X-linked protoporphyria caused by gain of function mutation in *ALAS2*
A Vanlander

Development and clinical consequences of white matter lesions in Fabry disease: a systematic review
S Körver

14.30-14.55 Pauze

14.55-15.40 Vrije voordrachten
Voorzitter: Dr. Charlotte Lubout (UMCG)

14.55-15.10 What you might be missing. The unknown sister of lactase-deficiency: sucrase-isomaltase deficiency
B Jacobs

15.10-15.25 Fibroblast growth factor 21 as a biomarker for long-term complications in organic acidemias
F Molema

15.25-15.40 Clinical and biochemical phenotype of 11 patients with bi-allelic mutations in TANGO2
P Verloo

Afsluitende borrel