VERENIGING KLINISCHE GENETICA NEDERLAND

Agenda 15 december

Adres: Academiegebouw, Domplein 29, 3512 JE Utrecht

9.30 uur		**Ontvangst met koffie/thee**
9.55 uur		**Welkom door voorzitter LOGcie**
10.00-11.45 uur		**Ochtendsessie** Voorzitter: Mieke van Haelst
10.00-10.15 uur		A Dutch multicenter study of the cardiac and genetic features of noncompaction cardiomyopathy in adults and children and it's relation to outcome. *J. van Waning - Erasmusmc*
10.15-10.30 uur		Discovery of novel genes for intellectual disability and multiple congenital anomalies in the next generation sequencing era: lessons learnt *M. Misra-Isrie –VUmc*
10.30-10.45 uur		Combination-PGD: clinical aspects and ethical considerations *V. van der Schoot - MUMC*
10.45-11.00 uur		Update on Leukodystrophies: A Historical Perspective and Adapted Definition *S. Kevelam – Radboudumc*
11.00-11.15 uur		Taking the next steps in validating NOTCH3 cysteine correction as a therapeutic approach for CADASIL. *G. Gravesteijn – LUMC*
11.15-11.30 uur		Mutations in ATP6V1E1 or ATP6V1A Cause Autosomal Recessive Cutis Laxa *T. Gardetchik – Radboudumc*
11.30-11.45 uur		Effect of intranasal insulin on development and behaviour in Phelan-McDermid syndrome: a randomized, double-blind, placebo-controlled trial *R. Zwanenburg – UMCG*
11.45-12.45 uur		**Algemene Ledenvergadering VKGN**
12.45-13.30 uur		**Lunch**
13.30-16.00 uur		**Middagsessie** Voorzitter: Maartje Nielsen
13.30-13.45 uur		Mutations in *KDM3B* cause intellectual disability and might be associated

		with cancer predisposition. *I. Diets - Radboudumc*
13.45-14.00 uur		Evaluation of current prediction models for Lynch Syndrome Updating the PREMM1,2,6 model to identify PMS2 mutation carriers. *A. Goverde - Erasmusmc*
14.00-14.15 uur		Performance of BRCA1/2 mutation prediction models in male breast cancer patients. *S. Moghadasi - LUMC*
14.15-14.30 uur		Long-term safety, quality and ethics of Preimplantation genetic diagnosis *M. Heijligers - MUMC*
14.30-14.45 uur		Exome sequencing detects a truncating NKX2-3 mutation in a family with intestinal varices. *C. Kerkhofs - MUMC*
14.45-15.15 uur		**Thee** Voorzitter vervolg programma: Marjolijn Jongmans
15.15-15.30 uur		Reverse phenotyping in patients with de novo mutations in PHIP leads to the recognition of a novel intellectual disability syndrome *S. Jansen – Radboudumc*
15.30-15.45 uur		Diagnostics of Syndromal and Monogenic Obesity: a Series of Interesting Cases *L. Kleinendorst - AMC*
15.45-16.00 uur		Bed to bench: cerebellar knock-out model of Chd7 *C. de Geus - UMCG*
16.00-16.30 uur		**Juryberaad + borrel**
16.30-17.15 uur		**Prijsuitreiking + vervolg borrel**