VERENIGING KLINISCHE GENETICA NEDERLAND

Assistentenlog/Wetenschapsdag VKGN/VKGL

21 maart 2019, In de Driehoek, Willemsplantsoen 1 C, 3511 LA Utrecht

9.30 uur	**Ontvangst met koffie/thee**
9.55 uur	**Welkom**
10.00-12.00 uur	**Ochtendsessie** *(voorzitter: Mala Misra-Isrie)*
10.00-10.15 uur	Orbital telorism and its relation to genetics, cognition, and brain morphology in the general population - *Hieab H.H. Adams, ErasmusMC*
10.15-10.30 uur	Loss of function of SETD1A causes a distinct neurodevelopmental disorder in humans, whereas knock down of the orthologue Set1 shows impaired memory in Drosophila. – *Joost Kummeling, RadboudUMC*
10.30-10.45 uur	GLS gain- and loss of function mutations - *Lynne Rumping, Amsterdam UMC locatie AMC*
10.45-11.00 uur	Reclassification of diagnostic whole exome sequencing results of 2014 - *Daniëlle Bosch, UMCU*
11.00-11.15 uur	Incidental findings in clinical exome sequencing - *Vyne van der Schoot, MUMC+*
11.15-11.30 uur	Rapid whole exome sequencing to identify the underlying genetic cause in fetuses with sonographic anomalies – *Chantal Deden, RadboudUMC*
11.30-11.45 uur	Next-generation phenotyping using computer vision algorithms in rare genomic neurodevelopmental disorders – *Sandra Jansen, RadboudUMC*
11.45-12.00 uur	Whole-exome sequencing identifies heterozygous de novo stop-loss mutations in HBB resulting in an elongated β-globin chain in two children with severe anemia- *Tamara Koopman, LUMC*
12.00-13.00 uur	**Lunch**
13.00-16.00 uur	**Middagsessie** *(voorzitter: Marjolijn Jongmans)*
13.00-13.15 uur	Polygenic Risk Scores: from Epidemiological Studies to Clinical Applications – *Philip Jansen, Amsterdam UMC locatie VUmc*
13.15-13.30 uur	Clinical applicability of the 313-SNP based Polygenic Risk Score for breast cancer risk prediction – *Inge Lakeman, LUMC*
13.30-13.45 uur	Rapid DNA-testing for hereditary breast cancer - *Beppy Caanen, MUMC+*
13.45-14.00 uur	Colorectal polyps and carcinoma in Birt-Hogg-Dubé syndrome – *Irma van de Beek, Amsterdam UMC*
14.00-14.15 uur	Declining APC and biallelic MUTYH mutation detection rates in polyposis patients – *Diantha Terlouw, LUMC*
14.15-14.45 uur	**Thee- en koffiepauze**
14.45-15.00 uur	Mutational landscape and patterns of clonal evolution in relapsed pediatric acute lymphoblastic leukemia - *Esmé Waanders, UMCU*
15.00-15.15 uur	TRIM28 haploinsufficiency predisposes to Wilms tumor – *Illja Diets, RadboudUMC*
15.15-15.30 uur	Consequences of Diagnosing a Tumour Predisposition Syndrome in Children with Cancer: a literature review – *Floor Postema, AMC*
15.30-16.00 uur	**Juryberaad en borrel**
16.00-17.00 uur	**Prijsuitreiking en vervolg borrel**