VERENIGING KLINISCHE GENETICA NEDERLAND

Agenda Wetenschapsdag VKGN 25 maart 2021

Adres: Digitaal

8.30-9.10		**Aanmelden via link in e-mail**
9.10 uur		**Welkom door dagvoorzitter**
9.15-12.00 uur		**Ochtendsessie**
9.15-9.30 uur		Gido Gravesteijn, postdoc, LUMC Vascular NOTCH3 aggregation load in CADASIL patients is associated with *NOTCH3* variant position
9.30-9.45 uur		Laura Claus, arts-onderzoeker, UMCU *KidneyNetwork: Using kidney-derived gene expression data to predict and prioritize novel genes involved in kidney disease*
9.45-10.00 uur		Lisette Leeuwen, AIOS Klinische genetica, UMCG *Genetic diagnoses using chromosomal analyses and exome sequencing in a prospective cohort of fetuses with structural anomalies*
10.00-10.15 uur		Marjoleine Broekema, AIOS Klinische genetica, Amsterdam UMC *FPLD3-associated PPARγ mutants define subclasses of target genes*
10.15-10.30 uur		Jeroen Smits, ANIOS /arts-onderzoeker, Klinische genetica, Radboudumc *A RIPOR2 in-frame deletion is a frequent and highly penetrant cause of adult-onset hearing loss*
10.30-11.00 uur		**Pauze**
11.00-11.15 uur		Nienke van Engelen, arts-onderzoeker, Prinses Máxima Centrum voor kinderoncologie *Cancer spectrum and penetrance in a national cohort of patients with a loss-of-function germline SMARCA4 alteration*
11.15-11.30 uur		Diantha Terlouw, arts-onderzoeker, LUMC *APC mosaicism testing in milder polyposis phenotypes reveals pks+ E.coli bacteria as possible additional explanation for the development of colorectal adenomas*
11.30-11.45 uur		Noah Helderman, student gnk / biomed wetensch, LUMC *The Molecular Profile of MSH6-Associated Colorectal Carcinomas From Patients With Lynch Syndrome*
11.45-12.00 uur		Janna Hol, arts-onderzoeker, Prinses Máxima Centrum voor kinderoncologie *High rate of (epi)genetic predisposing factors and an important role for DIS3L2 in a nationwide Wilms tumor cohort*
12.00-13.00 uur		**Lunch**
13.00-16.00 uur		**Middagsessie**
13.00-13.15 uur		Lot Snijders Blok, AIOS / arts-onderzoeker, Radboudumc *A clustering of missense variants in the crucial chromatin modifier WDR5 defines a new neurodevelopmental disorder*

13.15-13.30 uur		Danielle Bosch, AIOS Klinische genetica, UMCU *De novo recurrent variants in U2AF2 RNA-binding domains in an intellectual disability syndrome*
13.30-13.45 uur		Eva van Walree, arts-onderzoeker, Amsterdam UMC *Impairment of the MSH4-MSH5 heterodimer results in infertility due to male meiotic arrest and premature ovarian failure*
13.45-14.00 uur		Vivian Vernimmen, AIOS klinische genetica, MUMC *Neurofibromatosis type 1 and the next generation: is preimplantation genetic testing the solution?*
14.00-14.30 uur		**Pauze**
14.30-14.45 uur		Jessica Bos, ANIOS Klinische genetica, Amsterdam UMC *Vascular Ehlers-Danlos syndrome – A comprehensive natural history study in the Dutch patient cohort, preliminary results*
14.45-15.00 uur		Rosan Lechner, arts-onderzoeker, ErasmusMC *Whole-exome sequencing 677 aneurysm patients identifies multiple rare variants in the proprotein convertase FURIN causing impaired TGFβ family signaling.*
15.00-15.15 uur		Job Verdonschot, AIOS Klinische genetica, MUMC *Clustering of the cardiac transcriptome of dilated cardiomyopathy patients reveals opposite molecular signatures among patients with truncating and missense TTN variants*
15.15-15.30 uur		**Juryberaad**
15.30-16.00 uur		**Prijsuitreiking**