BeSHG/NVHG SYMPOSIUM 2019

Time	Thursday 19th September
9:30	Registration
10:30	Opening *Dr. M. van Haelst, president NVHG*
10:35	**CRISPR-Cas – from biology to applications** *Prof. Dr. John van der Oost*
	DNA Day Contest Award
11:15	Predicting Breast Cancer Risk using Rare and Common Variants *Prof. Dr. Douglas Easton*
12:00	**Lodewijk Sandkuijl Lecture**
12:40	Lunch
13:00	*Huishoudelijke vergadering VKGN*
13:00	*Huishoudelijke vergadering VKGL*
14:00	Hurdles for genetic research - what can we do about them?
14:00	*Background and objective of the session*
14:15	*Case 1 GDPR*
14:35	*Case 2 Multi-center METC approval*
14:55	*Open Mic (deelnemers dragen zelf voorbeelden/casusen aan)*
15:00	*Power Break*
15:15	*Case 3 Making genetic data FAIR*
15:35	*Discussion and action points*
16:00	Posters presentation (with coffee/tea)
17:30	**A result is only a result if it results in a result** *Prof. Dr. Peter de Knijff*

Social Evening Program

18:30	Pre-dinner drinks
19:00	Dinner
21:00	Networking event

Time	Friday 20th September
8:30	Registration (till 9:30)
8:45	**Identifying causative variants and genes in GWAS-identified IBD frisk loci** *Prof. Dr. Michel Georges*
9:25	**Integrated omics to accelerate diagnosis and therapy in inherited retinal diseases causing blindness** *Prof. Dr. Elfride De Baere*
10:05	Parallel sessions A/B
	A. Fertility and Pregnancy
10:15	*Preimplantation Genetic Testing with HLA matching: from counseling to birth and beyond / Martine De Rycke*
10:30	*TRIDENT-2: National Implementation of Genome-Wide Non-Invasive Prenatal Testing as a First-Tier Screening Test in the Netherlands / Karuna van der Meij*
10:45	*The landscape of pathogenic copy number variations in healthy, reproducing females / Kris Van Den Bogaert*
11:00	*In vitro fertilization does not increase the incidence of de novo copy number alterations in fetal and placental lineages / Masoud Zamani Esteki*
	B. Personalized Genomics
10:15	*Longitudinal assessment of the 313-SNP based Polygenic Risk Score for breast cancer risk prediction in a Dutch prospective cohort / Inge Lakeman*
10:30	*Towards the treatment of Cantú syndrome / Helen Roessler*
10:45	*NOTCH3 cysteine altering variants and their phenotypes in 92,456 whole exome sequenced participants of the Geisinger DiscovEHR initiative / Remco Hack*
11:00	*Towards personalized treatment of genetically classified refractory epilepsies using human induced pluripotent stem Cells (hIPSCs) as an ex-vivo tool / Eline van Hugte*
11:15	Break with Poster viewing *Coffee and tea will be served*
11:45	Parallel sessions C/D
	C. Diagnostic Opportunities
11:45	*Genotyping On ALL patients (GOALL); clinical implementation of high-throughput genotyping arrays. / Jeroen van Rooij*
12:00	*The added value of long-read amplicon sequencing for clinical applications / Kornelia Neveling*
12:15	*Reliable application of DNA-methylation signatures in genetic diagnostic testing / Peter Henneman*
12:30	*What if we would use a diagnostic multi-cancer gene panel for opportunistic screening? A study in 2,090 Dutch familial cancer patients / Helga Westers*

D. Biological Insight into Rare Disease

11:45 *Neurodevelopmental disorders: a next generation / Margot Reijnders*

12:00 *Transcriptome and protein analysis highlight the endosomal pathway in disease pathogenesis of metabolic CL syndrome / Lore Pottie*

12:15 *Noncoding structural variants disrupt the regulatory architecture of Rett genes / Eva D'haene*

12:30 *Loss of neutral sphingomyelinase-3 (SMPD4) links neurodevelopmental disorders to cell cycle and nuclear envelope anomalies / Daphne Smits*

Lunch and posterviewing

12:45 *Algemene Ledenvergadering NVHG*
Algemene Ledenvergadering BeSHG

14:00 **De novo mutations affecting male reproductive health**
Prof. Dr. Joris Veltman

14:40 **Human genetics at single cell resolution**
PD. Dr. Malte Spielmann

15:20 **Optical Mapping of 22q11.2 Low Copy Repeats reveals structural hypervariability**
Lisanne Vervoort

Awards 2019

16:00 *Young Investigator Award (for best thesis)*
Annual Award (for best oral presentation)
Poster Award (for best poster)
Puzzel prize

16:15 Closing Remarks

16:30 Meeting ends