NVHG AM2022

PROGRAM

JOINT MEETING BeSHG/NVHG

New advances and tools: towards personalized medicine of genetic disorders

Thursday 21 April 2022

08:45-09:30 Registration

09:30-09:45 Welcome and introduction presidents BeSHG and NVHG

09:45-11:15 **PLENARY SESSION 1 (Spatial genomics/transcriptomics)**

Chairs: Ligia Mateiu & Roland Kuiper

- Musa Mhlanga (Radboud University)
- Mihaela Zavolan (University of Basel)

11:15-11:45 Coffee break / Meet the sponsors / poster viewing

11:45-13:00 Selected oral presentations

Parallel session 1 (BS+BI)

Chairs: Julie Soblet & Lude Franke

- Mapping the 3D genome of the human retina and its role in retinal disease - Eva D'haene
- Single-cell evaluation of DNA damage in offspring after prenatal exposure to chemotherapy - Ilana Struys
- Hybrid Autoencoder with Orthogonal Latent Space for Robust Population Structure Inference - Meng Yuan
- Mutational processes in a Dutch cohort of children with Constitutional Mismatch Repair Deficiency - Dilys Weijers

Parallel session 2 (CL – novel gene/syndrome)

Chairs: Damien Lederer & Mieke van Haelst

- An unexpected moonlighting function of GTF3A in anti-herpesviral immunity: a new monogenic cause of herpes simplex encephalitis? - Simon Tavernier
- Loss of adipocyte phospholipase gene PLAAT3 causes lipodystrophy with neurological features due to inactivated arachidonic acid-mediated PPARγ signaling - Nika Schuermans
- Somatic activating PIK3R1 and non-hotspot PIK3CA mutations associated with a newly identified clinical phenotype: Capillary Malformation with Dilated Veins (CMDV) - Martina De Bortoli
- MAN2C1, a new gene associated with the development of cortical malformations - Hamide Yildirim
- CAMLG-CDG: a novel Congenital Disorder of Glycosylation linked to defective membrane trafficking - Matthew Wilson

13:00-14:00 Lunch break / Meet the sponsors / poster viewing

14:00-15:15 Selected oral presentation

Parallel session 3 (CL – NIPT)

Chairs: Kathelijn Keymolen & Erik Sistermans

- A cross-country comparison of women's perspectives on non-invasive prenatal testing in Belgium and the Netherlands - Karuna van der Meij
- The presence of viral DNA in a cohort of 108,349 Dutch NIPT samples and its relation to characteristics in pregnancy and cell-free DNA biology - Jasper Linthorst
- Multicentric longitudinal performance monitoring of different non-invasive prenatal screening technologies used in Belgium - Armelle Duquenne
- Noninvasive Prenatal Test results indicative of maternal malignancies: A nationwide genetic and clinical follow-up study - Catharina Heesterbeek

Parallel session 4 (Methodology)

Chairs: Nisha Limaye & Gijs van Santen

- Generic genome sequencing: one lab flow for all - Gaby Schobers
- GENType: all-in-one preimplantation genetic testing by pedigree haplotyping and copy number profiling suitable for third-party reproduction - Machteld Baetens
- The Dutch Center for RNA Therapeutics: a center to develop antisense oligonucleotide therapies for patients with nano-rare mutations - Marlen Lauffer
- Analysis of the genomewide BAF profiles of selected SNPs allows reliable aneuploidy detection in preimplantation embryos, independent of haplotyping - Pieter Verdyck

15:15-15:40 Coffee break / Meet the sponsors / poster viewing

15:40-17:15 **PLENARY SESSION 2 (1+M Genomes)**

Chairs: Gert Matthijs & Johan den Dunnen

- Introduction to project - Ruben Kok
- Plan of action 'Belgium' - Frederik Coppens
- Plan of action 'The Netherlands' - André Uitterlinden
- Genome Estonia - Andres Metspalu, University of Tartu

Debate

Moderators: Wendy van Zelst-Stams & Elfride De Baere

17:15-18:00 NVHG Galjaard lecture

Chair: Mieke van Haelst

19:00-00:00 Gala Dinner and Party

Friday 22 April 2022

08:45-09:15 Registration

09:15-10:45 **PLENARY SESSION 3 (Polygenic risk scores)**

Chairs: Bert Callewaert & André Uitterlinden

- Prof. Peter Devilee (Leiden UMC)
- Dr. Rens Reeskamp (Amsterdam UMC)

10:45-11:15 General assembly (BeSHG / NVHG)

11:15-11:45 Coffee break / Meet the sponsors / poster viewing

11:45-13:00 Selected oral presentations

Parallel session 5 (BS – animal work)

Chairs: Frank Kooy & Terry Vrijenhoek

- A tapt1 knockout zebrafish line with aberrant lens development and impaired vision models human pediatric cataract - Tamara Jarayseh
- Single-cell transcriptional dynamics and in vivo enhancer assays provide insight into gene regulatory networks of PRDM13 and IRX1 implicated in North Carolina macular dystrophy - Munevver Burcu Cicekdal
- SRSF1 haploinsufficiency is responsible for a new syndromic form of developmental delay including marfanoid habitus with intellectual disability - Elke Bogaert
- Live mouse tracker reveals autistic symptoms in the Fmr1 KO mouse model - Mathijs van der Lei
- A novel neurodevelopmental syndrome caused by loss-of-function of the Zinc Finger Homeobox 3 (ZFHX3) gene - María del Rocío Pérez Baca

Parallel session 6 (CL – diagnostics)

Chairs: Saskia Bulk & Aimee Paulussen

- Routine transcriptome sequencing improves diagnosis for neurodevelopmental disorders by identifying pathogenic effects of non-coding, putatively benign and missed variants - Jordy Dekker
- Overview of cancer predisposition syndromes in a national, unselected cohort of 836 children with a neoplasm - Jette Bakhuizen
- Polygenic risk scores predict overweight and obesity in the Dutch population - Bahar Sedaghati-khayat
- The potential of 3D facial analysis to recognize monogenic autism in the spectrum - Yoeri Sleyp

13:00-14:00 Lunch break / Meet the sponsors / poster viewing

14:00-15:30 **PLENARY SESSION 4 (VUS)**

Chairs: Lut Van Laer & Lisenka Vissers

- Prof. Roddy Walsh (Amsterdam UMC)
- Prof. Rosa Rademakers (University of Antwerpen)

15:30-16:00 Coffee break / Meet the sponsors / poster viewing

16:00-17:00 Surprise act

Chair: Gert Matthijs

17:00-17:30 Closing remarks and best poster / oral presentation awards

17:30 Reception

GENETICS RETREAT

BeSHG

NVHG