Final program Joint BeSHG/NVHG Meeting 2022

Topic: New advances and tools towards personalized medicine of genetic disorders.

Thursday 21 April 2022

8:45-9:30 Registration

9:30-9:45 Welcome and introduction presidents BeSHG and NVHG

9:45-11:15 PLENARY SESSION 1 – Chairs: Ligia Mateiu & Roland Kuiper

- **Musa Mhlanga** (Radboud University) - A chromatin-regulated biphasic circuit coordinates inflammation and trained immunity.
- **Mihaela Zavolan** (University of Basel) - Cell-type-specific expression of 3' UTR isoforms: quantification, modelling, prediction of functional relevance.

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

11:45-13:00 Selected oral presentations

- Parallel session 1 – Chairs: Julie Soblet & Lude Franke
    - Eva D'haene - Mapping the 3D genome of the human retina and its role in retinal disease
    - Ilana Struys - Single-cell evaluation of DNA damage in offspring after prenatal exposure to chemotherapy
    - Meng Yuan - Hybrid Autoencoder with Orthogonal Latent Space for Robust Population Structure Inference
    - Dilys Weijers - Mutational processes in a Dutch cohort of children with Constitutional Mismatch Repair Deficiency
    - Bert van der Zwaag - Scaling up Whole Exome Sequencing library prep in a ISO15189 accredited genome-diagnostics laboratory.
- Parallel session 2 – Chairs: Damien Lederer & Mieke van Haelst
    - Simon Tavernier - An unexpected moonlighting function of GTF3A in anti-herpesviral immunity: a new monogenic cause of herpes simplex encephalitis?
    - Nika Schuermans - Loss of adipocyte phospholipase gene PLAAT3 causes lipodystrophy with neurological features due to inactivated arachidonic acid-mediated PPARγ signaling
    - Martina De Bortoli - Somatic activating PIK3R1 and non-hotspot PIK3CA mutations associated with a newly identified clinical phenotype: Capillary Malformation with Dilated Veins (CMDV)
    - Hamide Yildirim - MAN2C1, a new gene associated with the development of cortical malformations
    - Matthew Wilson - CAMLG-CDG: a novel Congenital Disorder of Glycosylation linked to defective membrane trafficking

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

14:00-15:15 Selected oral presentation

- Parallel session 3 – Chairs: Kathelijn Keymolen & Erik Sistermans
    - Karuna van der Meij - A cross-country comparison of women's perspectives on non-invasive prenatal testing in Belgium and the Netherlands
    - Jasper Linthorst - 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
    - Armelle Duquenne - Multicentric longitudinal performance monitoring of different non-invasive prenatal screening technologies used in Belgium
    - Catharina J. Heesterbeek - Noninvasive Prenatal Test results indicative of maternal malignancies: A nationwide genetic and clinical follow-up study
    - Virginie Bros-Facer - NICUSeq: A Trial to Evaluate the Clinicak Utility of Human Whole-Genome Sequencing (WGS) Compared to Standard of Care in Acute Care Neonates and Infants (NICU-Seq).
- Parallel session 4 – Chairs: Nisha Limaye & Gijs Santen
    - Gaby Schobers - Generic genome sequencing: one lab flow for all
    - Machteld Baetens - GENType: all-in-one preimplantation genetic testing by pedigree haplotyping and copy number profiling suitable for third-party reproduction
    - Marlen Lauffer - The Dutch Center for RNA Therapeutics: a center to develop antisense oligonucleotide therapies for patients with nano-rare mutations
    - Pieter Verdyck - Analysis of the genomewide BAF profiles of selected SNPs allows reliable aneuploidy detection in preimplantation embryos, independent of haplotyping.
    - Rajendra Kumar Chauhan - Automated qPCR and NGS assays for Biomarker Validation and Discovery

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

15:40-17:15 PLENARY SESSION 2: Chairs: Gert Matthijs & Johan den Dunnen

- **Ruben Kok** - Introduction to 1+M Genomes project
- **Frederik Coppens** - Towards an interoperable infrastructure for human genomic data in Belgium.
- **André Uitterlinden** - Genome of Europe, and The Netherlands.
- **Andres Metspalu** (Uni of Tartu) - From Biobanking to Personal Medicine.
- **Wendy van Zelst-Stams** & **Elfride De Baere** - Debate with moderators

17:15-18:00 Galjaard lecture by **Cisca Wijmenga** – (Chair: Mieke van Haelst)

19:00-0:00 Gala Dinner and Party

Friday 22 April 2022

8:45-9:15 Registration

9:15-10:45 PLENARY SESSION 3 – Chairs: Bert Callewaert & André Uitterlinden

- **Peter Devilee** (Leiden UMC) - Working on topics related to the regulation of gene expression, particularly at the post-transcriptional and translational level, in the Risk-adjusted breast cancer screening: are we ready?
- **Rens Reeskamp** (Amsterdam UMC) - Polygenic scores for cardiovascular diseases: ready for clinical use?

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 – Chairs: Frank & Terry Vrijenhoek
    - Tamara Jarayseh - A tapt1 knockout zebrafish line with aberrant lens development and impaired vision models human pediatric cataract
    - Munevver Burcu Cicekdal - 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
    - Elke Bogaert - SRSF1 haploinsufficiency is responsible for a new syndromic form of developmental delay including marfanoid habitus with intellectual disability
    - Mathijs van der Lei - Live mouse tracker reveals autistic symptoms in the Fmr1 KO mouse model
    - María del Rocío Pérez Baca - A novel neurodevelopmental syndrome caused by loss-of-function of the Zinc Finger Homeobox 3 (ZFHX3) gene
- Parallel session 6 – Chairs: Saskia Bulk & Aimee Paulussen
    - Jordy Dekker - Routine transcriptome sequencing improves diagnosis for neurodevelopmental disorders by identifying pathogenic effects of non-coding, putatively benign and missed variants
    - Jette Bakhuizen - Overview of cancer predisposition syndromes in a national, unselected cohort of 836 children with a neoplasm
    - Bahar Sedaghati-khayat - Polygenic risk scores predict overweight and obesity in the Dutch population
    - Yoeri Sleyp - The potential of 3D facial analysis to recognize monogenic autism in the spectrum
    - Robin de Putter - BRCA testing to identify patients eligible for PARPi treatments across different indications

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

14:00-15:30 PLENARY SESSION 4 – Chairs: Lut Van Laer & Lisenka Vissers

- **Roddy Walsh** (Amsterdam UMC) - Enhancing rare variant classification with gene and disease-specific quantitative approaches.
- **Rosa Rademakers** (Uantwerpen) - New insights and unsolved questions in the genetics of frontotemporal lobar degeneration.

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