



Leuven 12th April - Single Cells: to care or not to care?

Friday April 12, 2024

Moderators: *prof. Geert Mortier, PhD, MD (UZ Leuven) & dr. Damien Lederer, MD (IPG Gosselies; BeSHG)*

08:00 – 09:00 Registration

09:00 – 09:15 Welcome address by the president of the BeSHG, **dr. Damien Lederer, MD (IPG Gosselies, BE)**

09:15 – 11:00 Plenary session 1 – *CAG Auditorium – Area 3*

Chair persons: prof. Geert Mortier, PhD, MD (UZ Leuven) & dr. Damien Lederer, MD (IPG Gosselies; BeSHG)

9:15 An introduction to single cell and spatial multi-omics
prof. Bernard Thienpont, PhD & prof. Thierry Voet, PhD (KU Leuven, BE)

9:40 Single cell human genetics: from genotype to phenotype one cell at a time
prof. Malte Spielmann, PhD, MD (University Hospital Schleswig-Holstein in Kiel & Lübeck, DE)

10:20 Linking common and rare diseases using gene regulatory networks
prof. Lude Franke, PhD (University Medical Centre Groningen, NL)

11:00 – 11:30 BeSHG Annual General Assembly

11:30 – 12:00 Coffee Break + Meet your sponsor + Poster viewing

12:00 – 13:00 Selected Oral Presentations

12:00 Parallel session 1: Tools and tricks in genetics – *CAG Auditorium – Area 3*

Chair persons: prof. Bernard Thienpont, PhD (KU Leuven) & dr. Julie Soblet, PhD (UH Brussels; BeSHG)

- Evaluation of a novel antisense oligonucleotide therapy targeting a 5'UTR mutation in the RDH12 gene (**Manon Bouckaert, UGent**)
- Two models, one mutation: functional characterisation of a Brugada syndrome associated CACNA1C variant in stem-cell-derived cardiomyocytes and zebrafish (**Bert Vandendriessche, UA Antwerpen**)
- Dissection of the 3D genome and long non-coding RNAs in the regulation of FOXL2 in novel ovarian granulosa cell models (**Charlotte Matton, UGent**)
- Unsupervised ECG feature extraction using deep learning empowers discovery of genetic determinants of cardiac electrophysiology (**Ewa Sieliwarczyk, UA Antwerpen**)

12:00 Parallel session 2: Diagnosing and understanding malignant disease – *GA3 Auditorium – Area 1*

Chair persons: prof. Thierry Voet, PhD (KU Leuven), dr. Arvid Suls, PhD (UA Antwerpen; BeSHG)

- Clonal heterogeneity and evolution during treatment in high hyperdiploid b-cell acute lymphoblastic leukemia (**Margo Aertgeerts, KU Leuven**)
- The Role of Hexokinase 3 in Familial Hodgkin Lymphoma (**Hiba Maalouf, UCL Louvain**)
- IMPRESS: Improved methylation profiling using restriction enzymes and smMIP sequencing, combined with a new biomarker panel, creating a multi-cancer detection assay (**Thomas Vanpoucke, UA Antwerpen**)
- Importance of SPRED1 in cutaneous melanoma (**Charlotte Carton, KU Leuven**)

13:00 – 14:30 Lunch break + Meet your sponsor + Poster viewing

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14:30 – 16:00 Selected Oral Presentations

14:30 Parallel session 3: Functional insights and genomics – CAG Auditorium – Area 3

Chair persons: prof. Joris Vermeesch, PhD (KU Leuven) & prof. Leila Zahed, PhD (UCL Brussels; BeSHG)

- Comparative 3D genome analysis between neural retina and RPE reveals differential cis-regulatory interactions at retinal disease loci (**Eva D'haene, UGent**)
- WES analysis of rare families points to a pathogenic role for the cGAS/STING – IFN-I axis in SSC (**Pierre Maus, UCL Louvain**)
- Matrisome expression patterns in murine and human dorsal root ganglia (**Delfien Syx, UGent**)
- Comprehensive tissue specificity analysis identifies (novel) retina-specific, long non-coding RNAs (**Emma Delanote, UGent**)
- Identifying regulatory variation in the human brain through single-cell multiomics and whole genome sequencing (**Alexandra Pančíková, KU Leuven**)
- Unlocking the power of multiomics with Novaseq™ X (**Vivek Mishra, Illumina**)

14:30 Parallel session 4: Clinical genetics and gene discovery – GA3 Auditorium – Area 1

Chair persons: prof. Hilde Van Esch, PhD, MD (KU Leuven) & prof. Saskia Bulk, PhD, MD (CHU Liège; BeSHG)

- Mutations in sterile alpha motif domain containing 7 (SAMD7) cause autosomal recessive macular dystrophy with or without cone dysfunction (**Miriam Bauwens, UGent**)
- Loss-of-function mutations of the TIE1 receptor tyrosine kinase cause congenital and late-onset primary lymphedema (**Lucas Potier, UCL Louvain**)
- Loss-of-function of the Zinc Finger Homeobox 4 (ZFHX4) gene causes a novel neurodevelopmental disorder (**María del Rocío Pérez Baca, UGent**)
- Understanding the role of somatic genetic variation in causing sporadic Frontotemporal Lobar Degeneration (**Vanshika Bidhan, UA Antwerpen**)
- Non-invasive cell-free DNA methylation profiling of pregnant women for the diagnosis and early risk determination of preeclampsia (**Machteld Baetens, UGent**)
- Liquid biopsy, outlook for routine clinical implementation (**Dr Martens, AZ Delta, for Astra Zeneca**)

14:30 Parallel session 5: Psychosocial aspects in genetics – GA2 Auditorium – Area 1

Chair persons: mrs. Aude Lombard (IPG Gosselies), mrs. Ariane Van Tongerloo (UGent) & dr. Kathelijn Keymolen, MD (UZ Brussels; BeSHG)

- The role and tasks of the clinical (educational) psychologist at the genetic centres across Belgium (**Ann Swillen, UZ Leuven**)
- Family communication and its legislation (**Amicia Phillips, KU Leuven**)
- Psychosocial impact of communicating VUS (**Charlotte Spaas, UZ Antwerpen**)
- TOP and future reproductive decision-making (**Ileen Slegers, UZ Brussel**)
- Expanded carrier screening (**Eva Van Steijvoort, KU Leuven**)

16:00 – 16:30 Coffee break + Meet your sponsor + Poster viewing



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- 16:30 – 17:45** **Plenary session 2 – CAG Auditorium – Area 3**
Chair persons: prof. Patrick Callaerts, PhD(KU Leuven) & prof. Bert Callewaert, PhD, MD (UZ Gent; BeSHG)
- 16:30 Single-cell analyses of the tumor microenvironment during checkpoint immunotherapy
prof. Diether Lambrechts, PhD (VIB - KU Leuven, BE)
- 17:05 A Decade of Molecular Cell Atlases
prof. Stephen Quake, PhD (Stanford University, USA)
- 17:45 – 18:00** Closing Remarks + Best Poster + Presentation Award
prof. Gert Matthijs, PhD (KU Leuven; BeSHG) & dr. Damien Lederer, MD (IPG Gosselies; BeSHG)
- 18:00 – 20:00** Reception
- 20:00 – 22:00** Party

Scientific & local organization committee:

Prof. Patrick Callaerts, Prof. Gert Matthijs, Prof. Geert Mortier, Prof. Bernard Thienpont,
Prof. Hilde Van Esch, Prof. Joris Vermeesch, Prof. Thierry Voet

Local contact:

Liliane Geyskens, Annemie Puttemans, Lindsey Stevens

