BeSHG
Belgian Society for Human Genetics

ANNUAL SYMPOSIUM 2024

SINGLE CELLS

To care or not to care?

12 APRIL

LEUVEN

AstraZeneca

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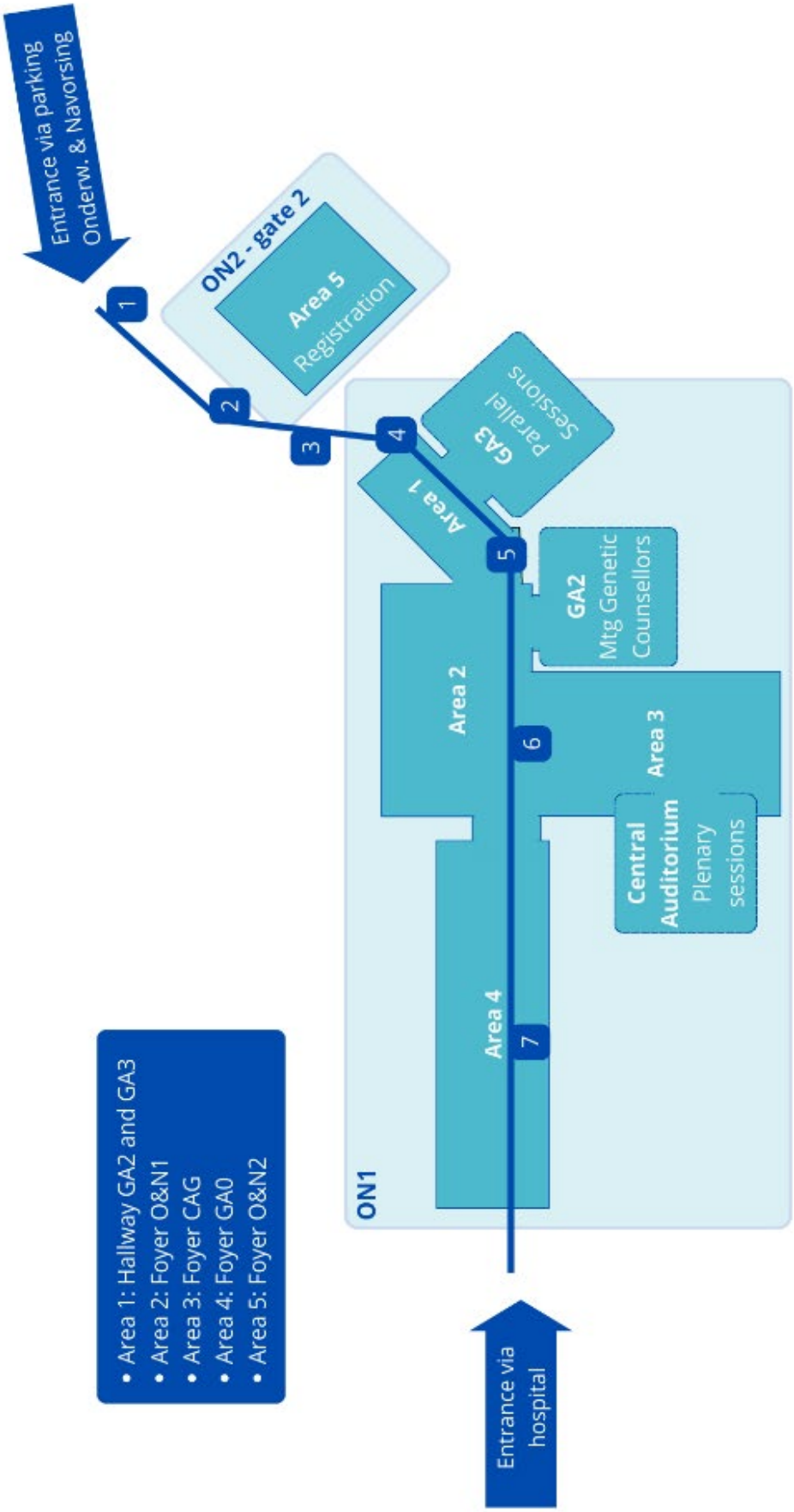
Roche

Locations


Entrance via parking Onderw. & Navorsing
1
ON2 - gate 2
Area 5
Registration
2
3
4
GA3
parallel
Sessions
Area 1
5
GA2
Mtg Genetic
Counsellors
Area 2
6
Area 3
Central
Auditorium
Plenary
sessions
Area 4
7
ON1
Entrance via hospital
• Area 1: Hallway GA2 and GA3
• Area 2: Foyer O&N1
• Area 3: Foyer CAG
• Area 4: Foyer GA0
• Area 5: Foyer O&N2

Dear Members,

It is my pleasure to welcome you at the 24th BeSHG meeting, here in Leuven.

Single cell, to care or not to care ? Obviously, you care, in view of the success of this meeting with more than 450 participants. I would like to thank the KU Leuven and UZ Leuven teams for the wonderful program and organisation of the meeting all along this year. Thank you to the speakers for their time and to travel to Belgium to present us their last works. Thank you also to people who submitted abstracts, allowing high level parallel and poster sessions. Finally, thank you to the sponsors for their support and presence. Please meet them at their boots.

The development of innovative sequencing methods and data analysis allows genetic community to have a better understanding of cellular pathophysiological processes in developing bodies. It also gives more opportunities to look for the missing heritability of known and unknown diseases. Implementing these technologies in routine will be the next revolution in the labs and we will have to adapt in a coordinated effort.

If deep analyses are the future of genetics, the care of patients and therapies should remain the ultimate goals. Genetics is more and more a multidisciplinary discipline and medical specialists are involved in prescription of genetic analysis and result transmission. Still, it is important that clinical geneticists remain the corner stone between the lab and patients in view of the complexity of deep phenotyping and of (deep) variants interpretation. Training of clinical geneticists is now part of the medical formation, but the role of genetic counsellors is still not recognised in Belgium despite their daily important contribution to patients care. Therefore, we are happy to welcome the first meeting for Belgian psychologists and genetic counsellors in an afternoon parallel session.

Therapy is now becoming a reality in more and more conditions. Introduction of treatments will necessitate a coordinated effort between genetic centres in order to share available therapies, research protocols to give Belgian patients the best treatment opportunities. On the other hand, the cost is a real hurdle for some conditions and it is our responsibility to fight for a fair price. This will request actions from genetic centres and BeSHG together with patients associations.

Lastly, Belgian genetics has always been strong and united thanks to the working groups. New working groups have appeared and others are becoming obsolete due to changes in genetic diagnostic techniques and new challenges, such as therapy, IVDR, recognition of lab specialists and genetic counsellors. Therefore, we intend to contemplate the future of working groups in the next year during a special day.

Thank you every one, technicians, biologists, researchers, bioinformaticians, clinicians, genetic counsellors and psychologists to allow day to day care to patients and to maintain Belgian genetics to a high level since many years.

Enjoy the meeting

Damien Lederer

President of BeSHG

Program

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 Sieliwonczyk, 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

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 NovaseqTM 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
  (G. 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

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

General information

Abstracts

All abstracts can be found on the website and via the following QR code: https://beshg.be/meeting/abstracts

Catering

Coffee breaks will be in area 1-4 (O&N1).
Lunch will be in area 1-4 (O&N1) and in area 5 (O&N2).
The reception will be in area 1-4 (O&N1).

Conference badges

Upon registration you will receive your name badge. Please wear your badge at all times to promote networking and to assist staff in identifying you.

Contact

General E-mail: BeSHG-admin@uantwerpen.be

If you have any queries or comments, please do not hesitate to contact a member of the crew, who will be pleased to help you. You can find the crew members at the meeting premises.

Internet access

The WiFi code will be displayed in the different areas of the event.

Privacy

Audio or video recording is not permitted. In case the presenter is asking the audience not to make screenshots of the slides, we kindly ask you to respect this. Professional photos will be taken during the event. If you don't wish to be present in these official event photos, we advise you to avoid the photographer

Party

The party will take place at the 'Alma', in the ALO building (Blauwe straat - poort 5).

Poster sessions

Posters will be displayed throughout the day, in areas 1 - 5. All posters will be numbered and assigned to one of the areas. Please display your poster at the right location upon arrival. Crew members will be present to assist.

Presentations

Please provide an electronic copy of your presentation to a member of the organizing team who will be based near the entrance of the meeting room in the morning.

An introduction to single cell and spatial multi-omics

Thierry Voet

Laboratory of Reproductive Genomics, Department of Human Genetics, KU Leuven, Belgium / Director Leuven Institute for Single Cell Omics (LISCO), KU Leuven, Belgium

Thierry Voet graduated with a Master in Bioscience Engineering: Cell and Gene Biotechnology from the University of Leuven (KU Leuven, Belgium), and holds an inter-university post-graduate in Human Genetics. Following his PhD in the Human Genome Laboratory at KU Leuven, he pioneered single-cell microarray analyses, and in 2010, he joined in the Cancer Genome Project at the Wellcome Sanger Institute (Cambridge, UK) to explore next-generation sequencing technologies for single-cell genomics, which led to an Associate Faculty membership. In 2014, he became Associate Professor at KU Leuven following a tenure track, and was appointed Professor in 2017 and Full Professor in 2022. In 2021, he founded the interdisciplinary KU Leuven Institute for Single Cell Omics (LISCO) together with 33 colleagues, of which he is currently director. His research focuses on (1) the development of methods for single-cell and spatial multi-omics, and (2) their application to study the biology of cells in human development, aging and disease.

Bernard Thienpont

Laboratory for Functional Epigenetics, Department of Human Genetics, KU Leuven, Belgium / Leuven Institute for Single Cell Omics (LISCO), KU Leuven, Belgium

Professor Bernard Thienpont trained as a bioengineer and did his doctoral research in Leuven, where he made important contributions to genetics: he identified a new genetic syndrome and a new gene that causes heart defects. He then moved to Cambridge, UK, studying the epigenetic causes of heart disease, and afterwards to the Vesalius Research Center, where he did cancer epigenetics research. Since 2018, he heads the Laboratory for Functional Epigenetics in Leuven, where he and his team try to discover epigenetic causes and biomarkers of human diseases using advanced genomic technologies. However, his main goal is always translational: improving patient care.

Abstract

The analysis of the genome, epigenome, transcriptome, proteome and/or metabolome from single cells is transforming our understanding of cell biology in health and disease. In a decade, the field has seen tremendous technological advances that enable crucial new insights into molecular mechanisms that govern development, physiology and pathogenesis. In this tutorial we highlight the main advances in the fast-developing field of single-cell and spatial (multi)omics technologies and demonstrate their value for fundamental cell biology and clinical translational research.

Single-cell analyses of the tumor micro-environment during checkpoint immunotherapy

Diether Lambrechts

VIB – KU Leuven Center for Cancer Biology, Belgium and Department of Human Genetics, KU Leuven

Professor Diether Lambrechts is Science Director of the VIB-KU Leuven Center for Cancer Biology, Group Leader at the VIB and Full Professor in the Department of Human Genetics at the University of Leuven, Belgium.

Prof. Lambrechts was trained as an engineer at the University of Leuven where he also pursued his PhD. He then worked at the Wellcome Trust Centre for Human Genetics at the University of Oxford, UK, before joining the VIB as an independent Group Leader in 2008. He currently holds a Kuang-Piu professorship chair at Zhejiang University. He has won several awards, including the Karel-Lodewijk Award for Human Medicine; the Galenus Prize for Pharmacology; the AstraZeneca Award for Translational Research and the Agilent Thought Leadership Award.

The expertise of the Laboratory for Translational Genetics headed by Prof. Lambrechts is focused on tackling important questions in oncology by translating genome-scale data sets into clinically applicable knowledge. Investigations are based on the application of cutting-edge sequencing technologies and bioinformatics, and on the seamless integration of genomic data sets with clinical and fundamental biological information to generate novel insights and potent biomarkers for the field of oncology. Prof. Lambrechts lab has developed a special interest in studying the mechanisms underlying genomic scars and developing biomarkers for them. He already succeeded in developing a novel microsatellite instability marker. This test was commercialised by Biocartis NV as the IdyllaTM MSI Test and recently also received FDA approval. His group also developed with the gynaeco-oncology team at the University Hospital Leuven an academic HRD test to guide treatment of patients with high-grade serous ovarian cancer towards PARP inhibition.

Recently, he has developed a special interest in dissecting the tumor microenvironment using single-cell technologies. He was among the first to characterize the lung tumor stroma (Lambrechts et al., Nat Med 2018) and in comparing stromal cells residing in tumor versus normal tissues across various cancer types (Qian et al. Cell Res 2020). Currently, he is focusing his team's efforts on studying changes in the tumor microenvironment during checkpoint immunotherapy at single-cell level (Bassez et al., Nat Med 2021). In this context, his lab is also applying the most recent spatial single-cell technologies to better understand how response and/or resistance to treatment is spatially embedded within the tumor microenvironment (Franken et al., Immunity 2024).

Abstract

Cancer immunotherapy using immune-checkpoint blockade (ICB) has created a paradigm shift in the treatment of advanced-stage cancers. In terms of lives saved and person-years restored, these therapies promise to be more significant than any other form of cancer treatment. However, one of the major limitations of ICB is that it provides durable clinical responses only in a fraction of patients. Single-cell technologies have been exceptionally instrumental in highlighting how checkpoint immunotherapy works in some patients, and why not in other patients. During my talk, I will highlight which T-cells respond to these therapies, which surrounding microenvironments are needed to provide durable responses and where in the tumor tissue these T-cells need to be located. I will also highlight how combinations of checkpoint inhibitors provide tremendous potential to enhance the clinical benefits of ICB, and how single-cell technologies can again be instrumental to develop novel synergistic therapies and combine them with approved checkpoint inhibitors.

Single cell human genetics: from genotype to phenotype one cell at a time

Malte Spielmann

Professor and Chair of Human Genetics Universität zu Lübeck, Director of Human Genetics at the University Hospital Schleswig-Holstein in Kiel & Lübeck

Dr. Spielmann is the director and chair of Human Genetics at the University of Lübeck and a research group leader at the Max Planck Institute for Molecular Genetics in Berlin. The main focus of his work is to understand the role of noncoding mutations and SVs as the cause of human disease and their influence on the 3D architecture of the genome. He has extensive experience with the clinical application of NGS technologies and the analysis of WGS data. Recently, the lab has also pioneered the development and application of single cell technologies to study human disease. Dr. Spielmann was a Heisenberg fellow of the DFG and is a member of the DFG priority programme "Spatial Genome Architecture in Development and Disease" (SPP 2202). He also serves on the program committees of the German and European Society of Humane Genetics. He has published more than 60 articles in peer-review journals with an h-index of 29 and over 5700 citations.

Abstract
Single-cell sequencing is a powerful approach that can detect genetic alterations and their phenotypic consequences in the context of human development, with cellular resolution. Whether germline or somatic in nature, some of these mutations may have significant genotypic impact and lead to diseased cellular phenotypes, either systemically or confined to a tissue. Single-cell sequencing enables the detection and monitoring of the genotype and the consequent molecular phenotypes at a cellular resolution. It offers powerful tools to compare the cellular lineage between 'normal' and 'diseased' conditions and to establish genotype-phenotype relationships. By preserving cellular heterogeneity, single-cell sequencing, unlike bulk-sequencing, allows the detection of even small, diseased subpopulations of cells within an otherwise normal tissue. Indeed, the characterisation of biopsies with cellular resolution can provide a mechanistic view of the disease. While single-cell approaches are currently used mainly in basic research, it can be expected that applications of these technologies in the clinic may aid the detection, diagnosis and eventually the treatment of rare genetic diseases as well as cancer. I will present data on several prospective applications of the technology in human genetics, namely the single-cell, whole-embryo phenotyping of mammalian developmental disorders, annotation of the non-coding genome using single-cell functional genomics, and the use of single-cell sequencing data for in silico variant prioritisation.

A Decade of Molecular Cell Atlases

Stephen Quake

Head of Science, Chan Zuckerberg Initiative and President, Chan Zuckerberg Biohub Network
Lee Otterson Professor of bioengineering and Applied Physics, Stanford University

Stephen Quake is Head of Science at the Chan Zuckerberg Initiative, overseeing a shared, comprehensive strategy across CZI's science program and technology teams, the CZ Biohub Network, and the Chan Zuckerberg Institute for Advanced Biological Imaging.

He has invented many measurement tools for biology, including new DNA sequencing technologies that have enabled rapid analysis of the human genome, and microfluidic automation that allows scientists to efficiently isolate individual cells and decipher their genetic code.

Quake is also the Lee Otterson Professor of Bioengineering and professor of applied physics at Stanford University. He was an Investigator of the Howard Hughes Medical Institute from 2006 to 2016.

Abstract

In recent years there has been tremendous progress towards deep molecular characterization of cell types using single cell transcriptome sequencing, creating so-called "Cell Atlases". These atlases provide a basic understanding of how different cell types of the same organism – which all share the genome – make distinct use of subsets of genes from the genome to create a variety of distinct cell types across tissues with specialized functions. I will discuss the cell atlases we have developed at the CZ Biohub which span fly, mouse, lemur and human, and discuss some of the history and technological innovation which led to these whole organism atlases.

Linking common and rare diseases using gene regulatory networks

Lude Franke

Professor of functional genomics, University Medical Centre Groningen, The Netherlands

Lude Franke develops and applies computational methods to understand the downstream molecular consequences of genetic risk factors. We studied the effect of somatic copy number alterations on gene-expression-levels (Fehrmann et al, Nature Genetics 2015) and studied the effect of genetic single-nucleotide polymorphism on methylation (Bonder et al, Nature Genetics 2017) and gene expression (i.e. expression quantitative trait loci), both in blood using bulk data (Zhernakova et al, Nature Genetics 2017 and Vosa et al, Nature Genetics 2021) and single-cell data (Van der Wijst, Nature Genetics 2018). Recently, we did this in brain (De Klein, Nature Genetics 2023). We lead the eQTLGen and single-cell eQTL consortia (see eQTLGen.org), where we use federated meta-analyses approaches to generate comprehensive maps how genetic variation is exerting cell-type and context-specific effects on gene expression, with the aim to help identify and validate drug targets.

Abstract

In the last few years many genetic risk factors have been found for a variety of common diseases through genome-wide association studies (GWAS). Through functional genomics approaches, e.g. by using expression quantitative trait loci (eQTLs) it is possible to gain insight into the molecular downstream consequences of these variants. Such knowledge can reveal drug targets, and can also be used to (in)validate such targets. Very recently, we have observed that the genetic risk factors for common diseases are actually very informative for rare diseases as well. It turns out that these common variants usually map near genes that can be connected to each other through gene regulatory network, and that the key genes that link these genes together, happen to be genes that cause severe, but rare forms of these diseases when mutated. In my presentation I will detail ongoing work on understanding the downstream molecular consequences of GWAS hits, how these downstream genes converge on a limited number of key genes through so-called gene regulatory networks that can cause rare forms of these diseases when mutate

Poster presentations

Poster	*Area*	
P1	A3	Germline mutations in DDX41 in a Quebec childhood cancer cohort of 484 patients: prevalence and case report ***Edith Sepulchre***
P2	A3	Copy Number Variation Analysis for HBOC testing reveals a recurrent CHEK2 duplication of exons 3-7 ***Katleen Janssens***
P3	A3	Non-invasive Markers of an Early Response to Immune Checkpoint Blockade in Breast Cancer ***Aurelie Mechels***
P4	A3	Pathogenic variants in HGF give rise to primary lymphoedema by loss of function. ***Pascal Brouillard***
P5	A3	Pathogenic EPHB4 variants of lymphatic-related non-immune hydrops fetalis and CM-AVM2 have discernible functional effects ***Héloïse Poullet***
P6	A3	Transcriptomic analysis of ROS1+ non-small cell lung cancer reveals an enrichment of nucleotide synthesis and cell adhesion pathways ***Marc Terrones***
P7	A3	The novel translocation t(114)(p33q32) juxtaposes IGH to TAL1, causing TAL1 overexpression in a T-ALL patient ***Amber Verhasselt***
P8	A3	DNA methylation biomarker panel can distinguish Pleural Mesothelioma from healthy pleura and blood ***Nele De Meulenaere***
P9	A3	Optical genome mapping – Evaluation as a routine diagnostic test in chronic lymphocytic leukemia ***Barbara Dewaele***
P10	A3	Copy-number analysis of plasma cell-free DNA identifies clonal mosaicisms years before being detectable in peripheral blood cells ***Stefania Tuveri***
P11	A3	Epigenetics and chemical risk assessment: an interdisciplinary framework using human data sets ***Eugenia Casella***
P12	A3	Optimization of the routine diagnostic workflow for Myeloid/Lymphoid Neoplasms with eosinophilia and Tyrosine Kinase gene fusions (MLN-TK) by optical genome mapping ***Justine Vanhevel***
P13	A3	Decoding the evolution and heterogeneity of Peripheral T-cell Lymphoma through deep multiomic sequencing ***Ruben Cools***
P14	A3	A functional single cell screen reveals a role for the epigenome in cancer cell heterogeneity ***Paulien Van Minsel***
P15	A3	NBAtlas: A harmonized single-cell transcriptomic reference atlas of human neuroblastoma tumors ***Noah Bonine***
P16	A3	Single-cell CRISPR screening characterizes transcriptional deregulation in T-cell Acute Lymphoblastic Leukemia ***Sofie Demeyer***
P17	A3	CD8+ T cells in end-organ damage in Lupus Nephritis ***Laura Watteyne***

Poster	*Area*	
P18	A3	A novel multiplex droplet digital PCR assay for simultaneous detection of eight frequent cancer types ***Isabelle Neefs***
P19	A3	Towards Improved Parsortix® efficiency for circulating tumor cell enrichment in pancreatic cancer ***Nele Vandenbussche***
P20	A3	Identification of a germline single-exon deletion of BARD1 gene in a breast cancer patient detected by sequencing-depth method ***Jérôme Coupier***
P21	A3	Single-cell alternative splicing analysis reveals novel tumor antigens predicting response to anti-PD1 checkpoint immunotherapy ***Jieyi Xiong***
P22	A5	Single nuclei transcriptomics of a Human In Vivo Neuronal Xenotranplantation Model for neurodevelopmental disorders linked to MECP2 ***Nona Merckx***
P23	A5	Decoding the mechanisms of neuronal wiring: a lesson from Trisomy 21 ***Francisco Pestana***
P24	A5	Shprintzen-Goldberg syndrome: a case report and review of literature ***Chatelain Camille***
P25	A5	Benchmarking of methods for DNA methylome deconvolution ***Kobe De Ridder***
P26	A5	A lethal multisystemic syndrome due to a novel mitochondrial disease with “gruyere-like” anomalies? ***Isabelle Maystadt***
P27	A5	Commingling analysis combined phenotyping for identification of 3D facial traits with potential major gene effects ***Meng Yuan***
P28	A5	Clinical features and developmental trajectories in school-aged children with 16p11.2 deletion ***Jente Verbesselt***
P29	A5	Structural variants as a cause of autosomal-dominantly inherited microtia ***Marie De Borre***
P30	A5	BeSolveRD: The Belgian Genome Resource to Resolve Rare Diseases ***Kris Van Den Bogaert***
P31	A5	Long-read sequencing and optical genome mapping fully characterize unresolved structural variation ***Griet De Clercq***
P32	A5	Myhre syndrome in adulthood: clinical variability and emerging genotype-phenotype correlations ***Eva Vanbelleghem***
P33	A5	Monoallelic RBBP4 variants describe a novel NuRDopathy associating neurodevelopmental delay and recurrent dysmorphic features ***Tanguy Demaret***
P34	A5	Three-dimensional facial phenotype analysis in Koolen-de Vries and Jansen-de Vries syndrome ***Jolijn Verseput***
P35	A5	Validation of the TrueDepth camera for accessible 3D facial scanning in clinical genetics **Kobe Vanheusden**

Poster	*Area*	
P36	A5	Early-onset cognitive decline as a feature of adult-onset D-2-Hydroxyglutaric Aciduria Type 1: A case report. ***Miel Theunis***
P37	A5	Evaluating the added value of aneuploidy screening on clinical outcomes in PGT-M embryos ***Machteld Baetens***
P38	A5	The potential of long-read whole genome sequencing based preimplantation genetic testing ***Yan Zhao***
P39	A5	New paradigms in clinical geneticist practice in the era of emerging therapies: examples of two pediatric syndromes. ***Julie Harvengt***
P40	A4	Single-cell profiling of Copy Number Variants in brain organoids ***Paula Longás***
P41	A4	A rational and flexible genetic approach to produce customized extracellular vesicles ***Lukas Hyka***
P42	A4	Characterization of anti-heparan sulfate proteoglycan Nanobodies ***Pascale Zimmermann***
P43	A4	Identification of the exosomal protein syntenin as a coordinator of degradative and secretory autophagy ***Cristóbal Cerda Troncoso***
P44	A4	Zebrafish as a model to study pain in Ehlers-Danlos syndromes ***Zoë Malfait***
P45	A4	Exploring the spectrum: Phenotypic characterization of COL4A2 dysfunction in zebrafish ***Merlijn Nemegeer***
P46	A4	Towards creation and characterization of COL2A1-SEDC and BGN-SEMD iPSC-derived chondrocyte models ***Pauline De Kinderen***
P47	A4	Genetic uptake in Loeys-Dietz syndrome genes is highest in spontaneous coronary artery dissection patients with extra-coronary arterial involvement ***Ivanna Fedoryshchenko***
P48	A4	Loeys-Dietz syndrome associated with truncating variants in PMEPA1 gene: First description of the phenotypic spectrum in three Belgian families. ***Claire Fouquet***
P49	A4	Reduced penetrance and variable expressivity are pitfalls in counseling for ectrodactyly ***Sien Van Daele***
P50	A4	RUNX2-related metaphyseal dysplasia with maxillary hypoplasia: a rare skeletal disorder resembling SFRP4-related Pyle disease ***Ewa Hordyjewska-Kowalczyk***
P51	A4	Assessing the contribution of non-coding variation in craniofacial development and disease ***Katerina Rapti***
P52	A4	Management of achondroplasia in Belgium: Overview of the current practice based on a multicentric survey. ***Julie Harvengt***
P53	A4	The neurodevelopmental and facial phenotype in individuals with a TRIP12 variant ***Mio Aerden***
P54	A4	Structural variants disrupt a critical regulatory region downstream of FOXG1 ***Lisa Hamerlinck***

Poster	*Area*	
P55	A4	Cardiovascular effects of fibrillin impairment in zebrafish ***Marina Horvat***
P56	A4	Oocyte/zygote/embryo maturation arrest: a functional and clinical study expanding the phenotype of NOBOX variants ***Annelore Van Der Kelen***
P57	A4	Expanding genotype-phenotype associations in biglycan-related Meester-Loeys syndrome ***Anne Hebert***
P58	A4	Genetic testing in patients with Congenital Heart Disease ***Maxim Verlee***
P59	A4	Enhanced insights into the genetic architecture of 3D cranial vault morphology using pleiotropy-informed conditional GWAS ***Seppe Goovaerts***
P60	A4	An integrated approach to uncover the genetic architecture of inherited blindness in a consanguineous Iranian cohort ***Lieselot Vincke***
P61	A4	Homozygous pathogenic MYH3 variant associated with arthrogryposis and lingual dystonia. ***Charlotte Mouraux***
P62	A4	Screening for RASopathies in a prenatal setting in Belgium ***Lise Boon***
P63	A4	Objective 3D phenotyping uncovers subclinical facial features in a 3q26 deletion/KBG syndrome dual diagnosis ***Michiel Vanneste***
P64	A4	Exome sequencing reveals homozygous frameshift variation in SPIDR causing ovarian dysgenesis ***Ozlem Okutman***
P65	A4	Understanding human linkeropathies: study of the phenotypic and molecular consequences of defective biosynthesis of the glycosaminoglycan tetrasaccharide linker region ***Jana De Troyer***
P66	A1	Physical activity in men and DNA methylation signatures in sperm: A genome-wide association study ***Eugenia Casella***
P67	A1	Progressive familial intrahepatic cholestasis type 4 (PFIC4): high clinical relevance of a rapid genetic diagnosis. ***Charlotte Masson***
P68	A1	Genetic counseling of the idiopathic pulmonary fibrosis in the university hospital of Liège ***Lena Kukor***
P69	A1	Novel UNC45B compound heterozygous variants in a child with congenital heart defects and progressive myopathy. ***Thomas Delguste***
P70	A1	Two unrelated cases of prenatal Shwachman-Diamond syndrome: a diagnosis complicated by a rare clinical presentation and a pseudogene ***Nathalie Vanden Eynde***
P71	A1	A novel RPE65-related dominant retinopathy caused by founder variant c.1555G>A p.(Glu519Lys) enriched in Belgian patients with a recognizable pattern dystrophy spectrum ***Eline Van Vooren***
P72	A1	Prevalence and characteristics of genetic disease in adult kidney stone formers ***Eric Olinger***

Poster	*Area*	
P73	A1	A genetic analysis of familial aggregation in inflammatory bowel disease multiplex families ***Deborah Jans***
P74	A1	Molecular insights into Pompe Disease: PPARgamma, COL13A1 and cellular glycogen metabolism ***Sophie Uyttebroeck***
P75	A1	Genetic background modulates aortic aneurysm formation and mortality in a SMAD3 mouse model ***Jotte Rodrigues Bento***
P76	A1	Unraveling calcium handling in Brugada Syndrome using the NCaMP7 GECI in hiPSC-derived cardiomyocytes ***Dogan Akdeniz***
P77	A1	Retrospective analysis of virtual gene panel analysis for genodermatoses reveals a high diagnostic yield in daily practice ***Aude Beyens***
P78	A1	Investigation of strategies to block downstream effectors of AT1R mediated signaling to prevent aneurysm formation in Marfan Syndrome ***Irene Valdivia Callejon***
P79	A1	Genetic risk stratification in the Leuven familial pulmonary fibrosis cohort ***Nathalie Veyt***
P80	A1	Impact of donor-derived cell-free DNA fraction assessment in monitoring kidney transplant recipients: Insights from a European prospective cohort study ***Angelica Pagliazzi***
P81	A1	Alterations in neurite outgrowth of dorsal root ganglia neurons from a mouse model of classical Ehlers-Danlos syndrome ***Ruben Vanlerberghe***
P82	A1	Overview of neuromuscular disorders epidemiology and diagnostic approach at the Neuromuscular Reference Center of Liège Belgium. ***Charlotte Mouraux***
P83	A1	Deciphering the human colon cell heterogeneity in Parkinson's disease ***Markus Boesch***
P84	A1	Polygenic risk scores in Alzheimer's disease risk prediction and the role of APOE ***Yasmina Abakkouy***
P85	A1	A midbrain nuclei atlas to characterize the genetic and epigenetic alterations in Parkinson's disease ***Anna Diacofotaki***
P86	A5	Investigation of epigenomic alterations in the presence of chromosomal instability during human preimplantation development using single cell multi-omics ***Thomas Lefevre***
P87	A5	WiNGS SV: A platform for analysis and federated sharing of structural variants ***Benjamin Huremagic***
P88	A5	Genomics Core Dashboard: a tool for the automatic annotation of variants in a clinical setting. ***Wouter Bossuyt***
P89	A5	Single-cell and spatial (multi-)omics for biomedical research @ KU Leuven ***Katy Vandereyken***
P90	A5	Beyond the Bench: Collaborative Genomics Analysis with WiNGS API ***Nishkala Sattanathan***
P91	A5	Advances in CRISPR-mediated knock-in of disease-related variants in zebrafish ***Elyne De Neef***

Poster	*Area*	
P92	A5	Development of a minimally Invasive RNA-Seq protocol for the diagnosis of Rare Disorders ***Erika D'haenens***
P93	A5	Computational methods to decode epigenetic regulation in tumor heterogeneity in a single cell CRISPR screen dataset ***Qian Yu***
P94	A5	Integrating multigenerational sequencing data to enhance de novo variant detection ***Laurens Hannes***
P95	A5	Baby Detect, molecular newborn screening: workflow and analytical validation ***Kristine Hovhannesyan***
P96	A5	Using low-coverage non-invasive prenatal screening data for polygenic risk score calculation ***Sara Becelaere***
P97	A5	Genetic history and health in Medieval Europe with the Belgian town of Sint-Truiden as a model ***Owyn Beneker***
P98	A5	FACS separation of sperm cells for enhanced forensic DNA profiling in sexual assault casework ***Kristina Fokias***
P99	A5	Nanopore long read whole genome sequencing in developmental disorders ***Mathilde Geysens***
P100	A5	Detailed analysis of pathogenic tandem repeats in a large control cohort ***Wouter De Coster***
P101	A5	Reassessment of long-read technology performance ***Erika Souche***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Notes

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Thanks for your attendance

Scientific committee in Leuven

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

Local organisation committee

Liliane Geyskens, Annemie Puttemans, Lindsey Stevens

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25

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BeSHG-admin@uantwerpen.be