Annual Symposium
2025

13 & 14 March
Louvain-La-Neuve

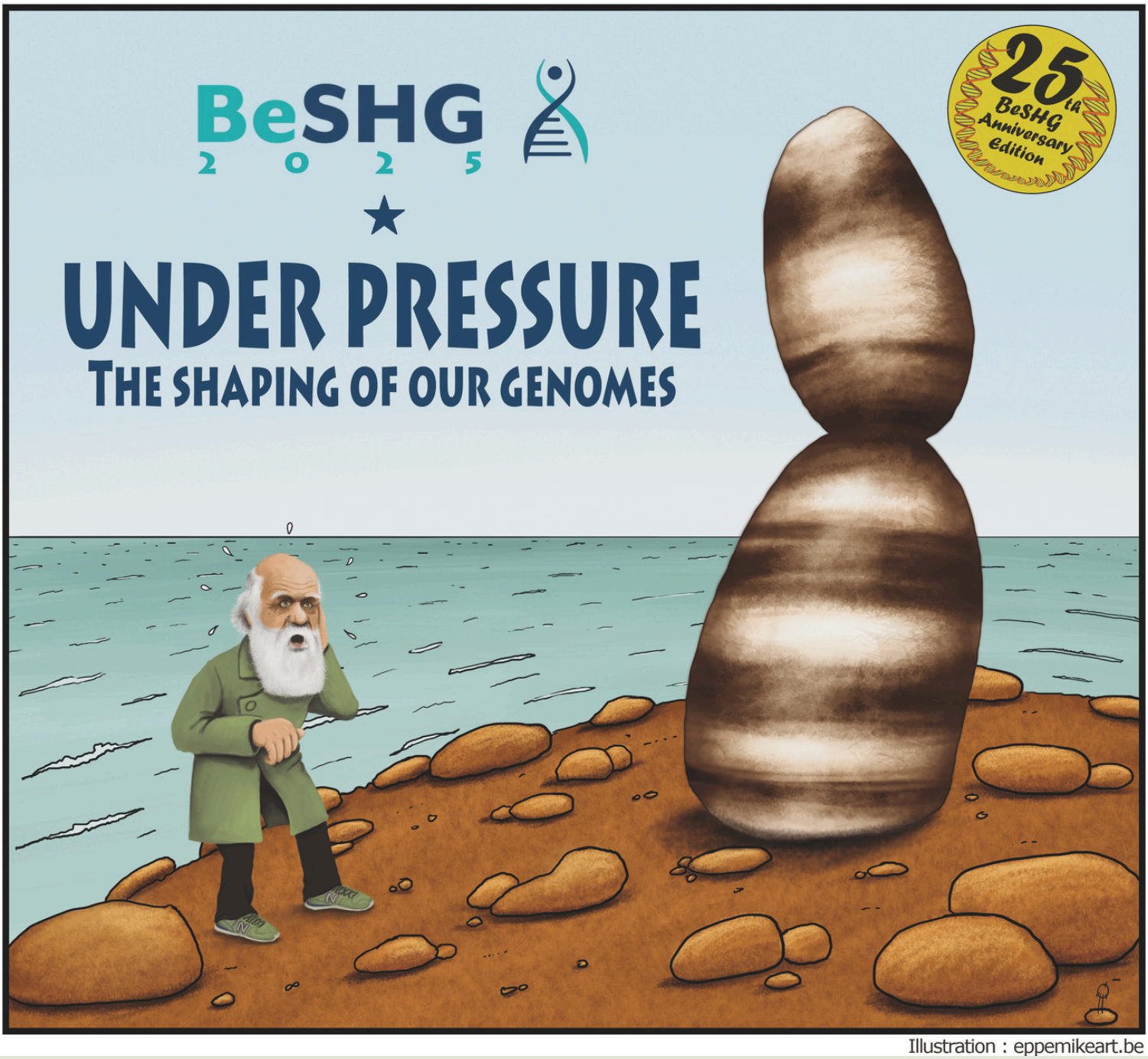


Illustration : eppemikeart.be

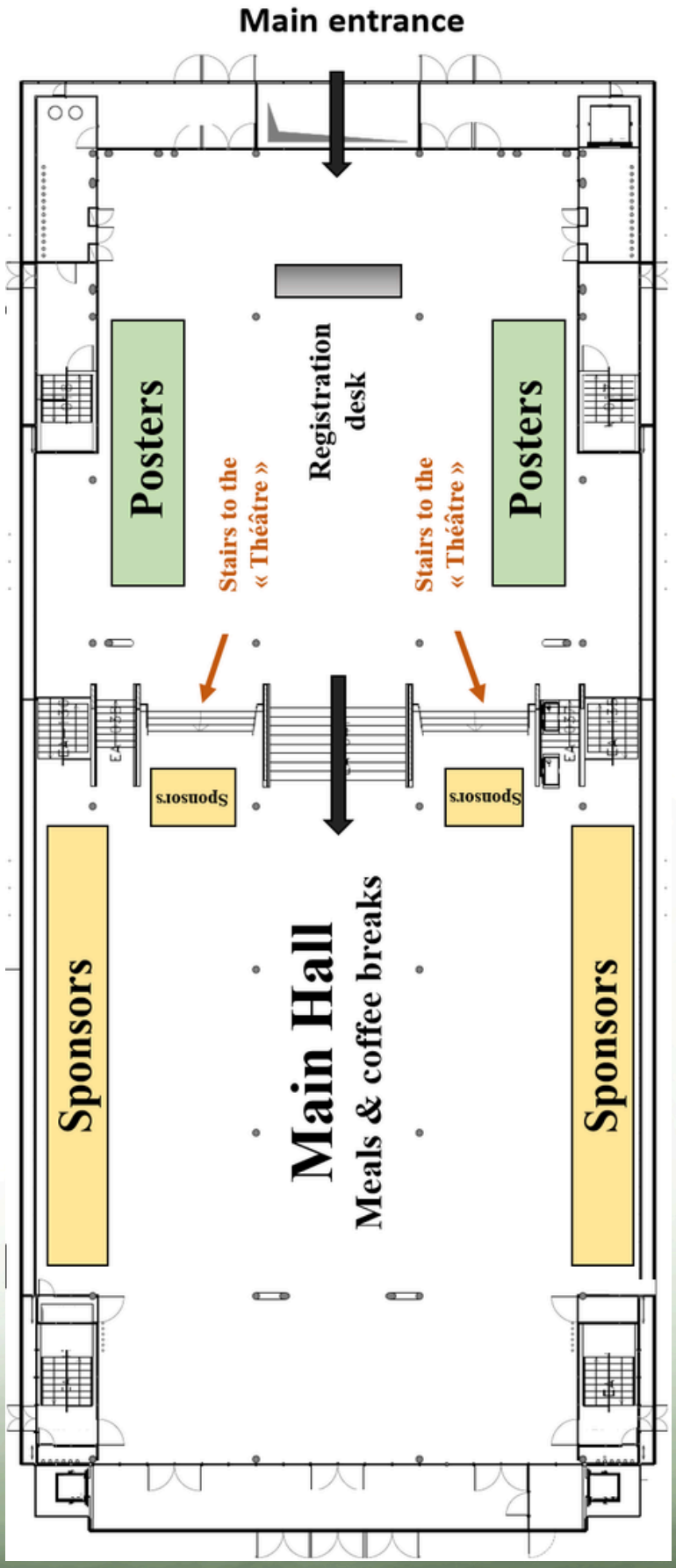
Roche

BeSHG is going greener !

1- Please recycle your badges by returning them at the end of the conference in the dedicated box at the reception

2- Certificates of attendance will be sent to you electronically by the BeSHG after the conference

Map of the venue



Dear BeSHG members,

On behalf of the local organizing committee members, and on behalf of the board of the Belgian Society of Human Genetics, we wish you a warm welcome and thank you for your attendance.

This event presents a unique opportunity to celebrate our vision and acknowledge the contributions of our pioneers. Science, infrastructure, resources, and the environment have changed dramatically in 25 years; we especially want to recognize those who have been instrumental in this progress.

For this anniversary edition, we had hoped for a truly exceptional guest.

While His Highness, King Philippe, and members of the Royal Family were unable to attend in person, this 25th annual edition is « Under The Patronage of Her Majesty The Queen ». Thus, we are extremely grateful that the Belgian Society of Human Genetics and the current meeting receive the support and interest of Her Majesty the Queen.

Thursday March 13th meeting, thanks to Han Brunner and Michel George, provides a wonderful opportunity for young scientists to present, discuss, and share their work.

The program on March 14 is designed to incorporate expertise, diversity, and needs, fostering future collaborations and interactions in our fields. We will also have the unique opportunity to debate about the future of genetic testing in Belgium in light of recent and future developments in clinical genetics, as analyses become more widely available for specialists and genetic screening in the population.

Working groups are the heart of BeSHG since the beginning. Their contribution to establish policies and guidelines allowed us to work and move forward together. After 25 years, it may be also a time to think about how working groups will operate in the future.

We express our sincere gratitude to our sponsors, whose significant contributions have been invaluable to this event. Your investment not only supports our organization but also facilitates dialogue and interactions that enrich our academic work. We especially thank Roche (Diamond) and, in alphabetical order, Agilent, AstraZeneca, Illumina, Limbus, Oxford Nanopore, and SeqOne (Gold), along with all our other sponsors. Please take the opportunity to visit their booths and speak with their representatives.

The future is bright for the next generation. We hope you have a productive meeting, taking full advantage of the scientific content and networking opportunities.

Please use the coffee and lunch breaks to connect with our national and international experts.

We look forward to seeing you again next year and in the years to come.

Yves Sznajer, local host
Center for Human Genetics,
CUSL UCLouvain

Damien Lederer
President, Belgian Society
of Human Genetic

Dear colleagues, dear friends, dear organisers

Thank you for asking me to say a few introductory words to this meeting celebrating the 25 th anniversary of the BeSHG. As some of you may know, I have been a dedicated clinical geneticist all along since, and even before, I became the director of the 8th center of medical genetics at the VUB in 1982. The first center at the KUL dates back to 1966 and the other 6 started their activities in between. I am still thankful to the other centers for helping me to start by sharing the funding by the government.

Now, I want to tell you a funny story concerning the BeSHG. When I was asked to say a few words today, I was honored and thought it was the right thing to do since I was convinced that I had been involved in the founding or rather the reactivation of the society, originally founded by Herman Vanden Berghe in the early days. I even thought that I had been the first president of the BeSHG for one year.

Preparing this short speech, I looked for archives by asking many of the colleagues who were or are active in the society, since my memory needed back-up, but no archives seem to be around. In my personal notes or… reports I could find limited information on the BeSHG.

In a report of one of our staff-meetings at the end of 2000, the organization of the first annual meeting in Liége on 9/2/2001 was mentioned with a note on wondering if my term was coming to an end ?

In a press-conference at the occasion of the the 30th anniversary of the High Council of Antropogenetics (now the college) in 2004, I read: In Belgium, medical geneticists convene regularly within the High Council mainly taking care of administrative matters while the BESHG, takes care of scientific matters in accordance with the High Council.

In my speech at the occasion of Willy Lissens retirement in 2014 I mentioned the BELBIS database and Belmolgen working group , initiated by Willy and still operating within the BeSHG.

In the annual reports of our center the meetings of the Society are mentioned with some information on its location and its content.

With the hope to find more information, I contacted ChatGPT as well as Perplexity and asked about the history of the society and its founders. Their answers were quick and similar but not very helpful.
I quote:

'The Belgian Society for Human Genetics (BeSHG) was established in March 2000 to serve as an official representative body for scientists involved in human genetics across Belgian universities and research institutes. The society aims to promote human genetics by supporting research, facilitating international collaboration, organizing scientific meetings, and addressing ethical issues in the field.
One of the key activities of the BeSHG is organizing annual meetings that serve as a platform for members to share research findings, discuss advancements, and explore collaborative opportunities'.

'In general, the establishment of such a society often involves a collaborative effort from various researchers and institutions across the country. Key figures in Belgian genetics, such as Herman Van den Berghe, known for his contributions to cancer cytogenetics, might have played roles in shaping the field. If you are looking for detailed information about the founders, it might be helpful to contact the BeSHG directly or consult their official publications or website for more specific details'

When I looked to the actual website, great was my surprise to see that it was Alain Verloes who was the first president in 2000. And when I talked to him on the phone, he not only confirmed that he had been the first president but he told me that the idea of founding a BeSHG did arise in Manchester when he, Koen Devriendt and Geert Mortier were having a drink in a pub after the dysmorphology meeting organized by Diane Donnai in 1999. Further documents or information are not available.

Looking at the addendum of the Moniteur Belge/ Belgisch Staatsblad of 6/2/2001, I found that the members of the first board were Alain Verloes, Geert Mortier, Marc Abramowicz, Inge Liebaers, Pascale Cochaux, Koenraad Devriendt, Frank Kooy and Lionel Van Malderghem. So I was not the president but I was involved.

The aim of the society was specified, more or less like ChatGPT told me before. The Society had to promote human genetics by stimulating genetic research, promote collaboration of scientists in the field in Belgium and abroad, organize scientific meetings and take care of permanent education, inform the general public, reflect on the impact of genetic technology on the society including ethics, develop applications of the technology to the human being in a correct way.

I am happy to be able to conclude that the aim of the Society as described in the Moniteur/Staatsblad in 2001 has been achieved. Not only do we, as geneticists, meet at the interesting annual meetings, but the different working groups are also active and stimulate, the collaboration between the 8 centers for medical genetics. Of course, It would have been nice to be able to tell the story of the BeSHG in a more accurate way. Maybe we can still try to complete the gaps to realize how the society became as it is today thanks to many of you.

Inge Liebaers, Annual meeting of the BeSHG, Louvain La Neuve, March 14,2025

Scientific Program

Day 1 : March 13, Genetics Retreat

Location: Foyer du lac

Registration : 8:00 – 9:00

9:00-9:05: Welcome address by the president of the BeSHG, ***Dr Damien Lederer*** (IPG)

9:05-9:15: Presentation of the moderators: ***Professor Han Brunner*** (NL) and ***Professor Michel Georges*** (BE)

9:15-10:30: Oral presentations I

- 9 :15-9 :30 : ***Jay Devine*** : Revealing the developmental-genetic basis for the human cranial vault.
- 9 :30 -9 :45 : ***Kristine Hovhannesyan*** : BabyDetect project: interim results and some challenges in newborn screening.
- 9 :45-10 :00 : ***Lisa Hamerlinck*** : Structural variants disrupt a critical regulatory region downstream of FOXG1
- 10 :00 -10 :15 : ***Eline Van Vooren*** : Discovery, replication and characterization of protein, in vitro and iPSC-RPE stem cell models of a novel dominant RPE65- retinopathy, an actionable RPE disease
- 10 :15-10 :30 : ***Mathijs van der Lei*** : Biallelic GAD2 variants cause a early-onset developmental encephalopathy

10:30-11:00: Coffee break

11:00-12:30: Oral presentations II

- 11 :00-11 :15 : ***Janah Vandenhoeck*** : Methylation Biomarkers can Distinguish Pleural Mesothelioma from Healthy Pleura and other Pleural Pathologies
- 11 :15 -11 :30 : ***Dogan Akdeniz*** : Using the Genetically Encoded Calcium Indicator NCaMP7 in hiPSC-derived cardiomyocytes to model Brugada Syndrome
- 11 :30-11 :45 : ***Pierre Maus*** : PRKD2 as a candidate gene modulating T cell activation in familial Systemic sclerosis
- 11 :45 -12 :00 : ***Edith De Bruycker*** : Unravelling the proxisome of photoreceptor-specific nuclear receptor NR2E3 reveals a potential molecular link between transcriptional regulation and splicing in human retina
- 12 :00-12 :15 : ***Lucas Potier*** : Functional validation of novel TIE1 variants as causes for primary lymphedema
- 12 :15-12 :30 : ***Robin De Putter :*** Diagnosing HEreditary predisposition syndromes for Childhood cancer: Implementation in clinical PRactice (DHECIPR)– preliminary results

Location: Foyer du lac

12:30-14:00: Lunch break

14:00-15:30: Oral presentations III

- 14 :00-14 :15 : ***Dries Vanisterbecq*** : Unraveling VUS in BRAF: A path to improve diagnosis, prognosis, and therapy in cancer patients
- 14 :15 -14 :30 : ***Aneta Malesa*** : A novel form of autosomal dominant spondylocostal dysostosis in three unrelated families caused by the same heterozygous pathogenic variant in MESP2
- 14 :30-14 :45 : ***Wannes Renders*** : Genetic background of patients with childhood onset cardiomyopathy: results from a retrospective cohort study
- 14 :45 -15 :00 : ***Mathilde Geysens*** : Clinical evaluation of long-read sequencing based episignature detection in developmental disorders
- 15 :00-15 :15 : ***Marina Horvat*** : Cardiovascular effects of fibrillin impairment in zebrafish
- 15 :15-15 :30 : ***Irene Valdivia*** : Unravelling shared pathomechanisms in syndromic thoracic aortic aneurysm disorders using single-cell RNA sequencing

15:30-16:00: Coffee break

16:00-17:00: Wrap up and lecture by ***Professor Han Brunner*** (BE): Why we have the diseases we have

- Announcement of the 3 best student oral presentations
- Concluding remarks

Scientific Program

Day 2 : March 14, 25th Anniversary Conference

Location: Le Théâtre

Registration and coffee : 8:00 – 9:00

Session 1 : 09 :00 – 10 :45

- 9 :00-9 :15 : Welcome address by the president of the BeSHG, ***Dr Damien Lederer*** (IPG), the head of the organizing center (UCL), ***Professor Yves Sznajer,*** and one of the BeSHG founding members, ***Em. Professor Inge Liebaers*** (VUB)

9 :15-10:45 : **Invited speakers**

- 9 :15-10 :00 : ***Professor Michel Georges*** (BE) Coevolution of balanced polymorphisms in the host and its intestinal microbiome *(Chairpersons: Professor Alain Verloes , Doctor Anne De Leener)*
- 10 :00 -10 :45 : ***Dr Jennifer Meadows*** (SE) Comparative Genomics through the lens of Zoonomia: mammalian evolution and complex traits *(Chairpersons : Professor Frank Kooy, Professor Nisha Limaye)*

10 :45-11 :15 : Coffee break and poster session

Session 2 : 11 :15-13 :00

- 11:15-12:00 : **Invited speaker : *Professor Molly Przeworski*** (USA) Why do human germline mutation rates depend on age and sex? *(Chairpersons : Professor Geert Mortier, Professor Jean-Baptiste Demoulin)*
- 12 :00-12 :30 : General Assembly
- 12 :30-13 :10 : Best student oral presentations *(Chairpersons : Professor Geert Mortier, Professor Jean-Baptiste Demoulin)*

13 :10 -14 :30 : Lunch and poster session

Session 3 : 14 :30-16 :00 : **Invited speakers**

- 14:30 - 15:15 : ***Professor Sam Behjati*** (UK) Childhood cancer predisposition, penetrance and mosaicism *(Chairpersons : Professor Marc Abramowicz, Doctor Pascal Brouillard)*
- 15:15 - 16:00 : ***Dr Hilary Martin*** (UK) The differential effects of common and rare genetic variants on cognitive performance across development *(Chairpersons : Professor Koenraad Devriendt, Doctor Raphael Healers)*

Location: Le Théâtre

16 :00-16 :30 : Coffee break and surprise

Session 4 : 16 :30-18 :00 **Round Table Discussion : The Future of Genetic Testing**

Moderator : ***Professor Han Brunner***

Participants : ***Professor Sam Behjati, Doctor Candy Kumps, Professor Inge Liebars, Ms. Ileen Slegers, Mr. Thomas Minten***

- Best poster and best student presentation awards

Concluding remarks : ***Dr Damien Lederer***

18 :00 – 22 :00 Drinks, Dinner and 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, lunch, reception and party will be in the main Hall

Badges
Please wear your conference badge at all times to promote networking and to assist staff in identifying you.

Assistance
If you have any questions or comments, please do not hesitate to contact a member of the ***Genetics Squad***, identifiable by their white T-shirt with a fun drawing on the back.

Internet access
Wi-Fi code: **BESHG2025** Password: **underpressure**

Privacy
Audio or video recordings are not permitted. Please respect the wish of speakers if they ask you not to take any photos of their presentation.

Poster sessions
Posters will be displayed from 9am till 6 pm in the registration hall at the main entrance of Aula Magna.

Presentations
Please provide an electronic copy of your presentation to a member of the organizing team at the entrance of the Theater.

Han Brunner

Why we have the diseases we have

Professor of Human Genetics, Dept Human Genetics Radboudumc Nijmegen and Maastricht UMC, the Netherlands

Han Brunner is the former Head of the Institute of Human Genetics at Nijmegen Radboudumc, and at Maastricht Medical Center, in the Netherlands. Han promotes rapid implementation of genomic technologies in Medicine, which is advantageous for patients, and families. Genomic approaches and can make the care for people with rare diseases more effective.
His scientific work has shown that new mutations are the main cause of intellectual disability, which led to the acceptance of exome sequencing as a first-tier test in neurodevelopmental disorders. Recent work from his group established that heterozygote selection is relevant to the overall landscape of autosomal recessive diseases in European populations.

Abstract

Most intellectual disability is due to new mutations. New mutations occur at any position in the genome and in any gene. However, large genes present a larger mutational target and this explains why ARID1B (Coffin-Siris) and ANKRD11 (KBG syndrome) are very common. The rate of mutation increases with the age of the father. Some mutations are positively selected in the testis and for this reason occur more commonly than would be expected. This includes PTPN11 (Noonan syndrome) and FGFR3 (achondroplasia).
The landscape of recessive diseases is quite similar in the Dutch, British, and Estonians. However, the genetic variants in these disease genes are largely different.
This suggests that we constantly lose pathogenic variants because carriers are at a (very small) reproductive disadvantage.
We conformed in a recent study that heterozygous carriers of an autosomal recessive variant are:
- more likely to be childless,
- have completed less education
From this we conclude that carriers of recessive ID variant have slightly more difficulty finding a partner.

Michel Georges:

Coevolution of balanced polymorphisms in the host and its intestinal microbiome

Professor Emeritus, University of Liège, Belgium

Michel Georges is Professor in Genetics and Genomics at the University of Liège, and the head of the Unit of Animal Genomics. He played an instrumental role in establishing the GIGA Institute and its director from 2016 to 2023. He devoted his carrier to the development of genomic tools for the identification of genes and mutations underlying complex traits of agronomic and medical importance. With his collaborators, he cloned the double-muscling gene, discovered polar overdominance, identified the first mammalian phenotype due to perturbed microRNA regulation, discovered a novel CNV generating mechanism underlying colour-sidedness, identified several QTN at single-base pair resolution, and demonstrated that ABO genotype alters the microbiome by affecting GalNAC concentrations. His laboratory has contributed to multiple GWAS and post-GWAS studies for Inflammatory Bowel Disease. Michel Georges was awarded the Wolf Prize in Agriculture in 2007, the Francqui Prize in Biomedical Sciences in 2008, and an ERC advanced grant in 2013. He has been a member of NAS since 2013. He obtained his DVM from the University of Liège (1983), a MSc in Molecular Biology from the Free University of Brussels (1985), and his habilitation from the University of Liège (1991). From 1989 to 1993 he was senior scientist at Genmark and adjunct professor in Human Genetics in Salt Lake City, Utah.

Abstract

We have recently shown that A(B)O genotype affects the abundance of the p.75.a5 Erysipelotrichaceae genus in the cecum and feces of pigs. We have shown that this effect was mediated by modulating the concentrations of free GalNAc in the intestine, and that the affected bacteria are able to use GalNAc as carbon source using a non-inducible GalNAc operon. We have also shown that the A(B)O alleles of Suidae may have persisted for 10 million years and are therefore most likely undergoing balancing selection as in primates. We further show that the GalNAc operon is polymorphic in p.75.a5, and that the corresponding alleles respond in an antagonistic manner to A(B)O genotype. Thus, it appears that a balanced polymorphism in the host may maintain a coevolved balanced polymorphism in the intestinal microbiota. Recent data indicate that a similar phenomenon is operating in humans.

Jennifer Meadows

Comparative genomics through the lens of Zoonomia: mammalian evolution and complex traits.

Department of Medical Biochemistry and Microbiology, Uppsala University, Sweden

Dr Jennifer Meadows is based at Uppsala University, Sweden. Jennifer uses comparative genomics, both within and across the species border, to dissect and characterise the natural variation in genome structure and regulation that underpins heritable complex traits. While her team has a special focus on using domestic dogs as model of human health, the current scale of tools and resouces available mean that they can also explore the basics of genome prcoessing and the fundamental questions of what it means to be human.

Abstract

Comparative genomics allows us to look both within populations and across the species boundary. This talk will touch on both. Through the reference free alignment of 240 diverse placental mammals, the Zoonomia Consortium resource encompasses 100 million years of evolution, viewed at single base pair resolution. The constrained evolutionary footprints in this data can be used to ask questions such as, what are the essential elements of being a mammal, be these at the protein coding or genome regulatory level. Leveraging the considerable omics data available from studies of humans, mice, and domestic animals, we can infer the constrained and likely functional regions of non-model organisms, opening new avenues of research for at-risk or hard to access populations. By delving into the opposite side of the evolutionary coin, acceleration, species specific innovations can be identified, including those that make us human. This talk will illustrate some of these questions, and highlight how with more than 6,000 extant mammals spanning every continental niche, our evolutionary questions are only tempered by our imaginations.

Molly Przeworski

Why do human germline mutation rates depend on sex and age?

Alan H. Kempner Professor of Biological Sciences and Systems Biology, Columbia University, USA

Molly Przeworski is a population geneticist at Columbia University. She is interested in how and why fundamental processes such as mutation and recombination differ among vertebrate species, as well as in the dynamics of natural selection in humans. Molly received a B.A. in Mathematics from Princeton University and a Ph.D. in Evolutionary Biology at the University of Chicago, then conducted postdoctoral research in Statistics at the University of Oxford. Before moving to Columbia University, she was a researcher at the Max Planck Institute for Evolutionary Anthropology and a faculty member at Brown University and the University of Chicago. She is a member of the American Academy of Arts and Sciences and the National Academy of Sciences in the United States, and received the 2023 Scientific Achievement Award from the American Society for Human Genetics.

Abstract

Germline mutation is the source of all heritable differences and therefore of fundamental importance. In mammals, it has long been appreciated that mutation rates are higher in fathers, particularly older fathers. The textbook view has long been that these patterns reflect replication errors that accrue during spermatogenesis. I will present multiple lines of evidence that call this view into question. I will argue instead that current data are best explained by a much larger role of DNA damage in the genesis of germline mutations than previously appreciated, and draw implications for why mutation rates depend on sex and age.

Sam Behjati

Childhood cancer predisposition, penetrance and mosaicism

Group Leader, Wellcome Sanger Institute; Honorary Consultant Paediatric Oncologist, Cambridge University Hospitals; Professor of Paediatric Oncology, University of Cambridge

I am Group Leader at the Wellcome Sanger Institute, Cambridge (UK). My group focuses on investigating the fetal origin of childhood cancer. We use traditional phylogenetic approaches to reconstruct the early fetal history of tumours whilst applying cutting-edge single cell technologies to define the developmental origin of tumours in quantitative molecular terms. I remain clinically active as an Honorary Consultant Paediatric Oncologist at Addenbrooke's Hospital in Cambridge.

Abstract

In this talk I will show phylogenetic studies in childhood cancers that investigate childhood cancer predisposition, penetrance and mosaicism.

Hilary Martin

The differential effects of common and rare genetic variants on cognitive performance across development

Group leader in Human Genetics, Wellcome Sanger Institute

Hilary started her university studies in Australia and then completed her PhD with Peter Donnelly at the Wellcome Trust Centre for Human Genetics in Oxford in 2015. There, she worked on an eclectic set of projects ranging from rare disease genomics to population genetics of the platypus. She moved to the Sanger Institute initially as a postdoc with Jeff Barrett, and then started her own group there in 2018. Her group analyses large-scale genetic and electronic health record data to explore the genetic architecture of neurodevelopmental disorders and traits, as well as complex diseases in populations with high levels of consanguinity.

Abstract

Common and rare genetic variants that impact adult cognitive performance also predispose to rare neurodevelopmental conditions involving cognitive deficits in children. However, their influence on cognition across early life remains poorly understood. Here, we investigate the contribution of common genome-wide and rare exonic variation to cognitive performance across childhood and adolescence primarily using the Avon Longitudinal Study of Parents and Children (n=6,495 unrelated children). We show that the effect of common variants associated with educational attainment and adult cognitive performance increases as children age. Conversely, the negative effect of deleterious rare variants attenuates with age. Using trio analyses, we show that these age-related trends are driven by direct genetic effects on the individual who carries these variants rather than indirect genetic effects mediated via the family environment. We further find that the increasing effects of common variants are stronger in individuals at the upper end of the phenotype distribution, whereas the attenuating effects of rare variants are stronger in those at the lower end. Concordant results were observed in the Millenium Cohort Study (5,920 children) and UK Biobank (101,232 adults). The effects of common and rare genetic variation on childhood cognitive performance are broadly comparable in magnitude to those of other factors such as parental educational attainment, maternal illness and preterm birth. The effects of maternal illness and preterm birth on childhood cognitive performance also attenuate with age, whereas the effect of parental educational attainment does not. We show that the relative contribution of these various factors differs depending on whether one considers their contribution to phenotypic variance across the entire population or to the risk of poor outcomes. Our findings may help explain the apparent incomplete penetrance of rare damaging variants associated with neurodevelopmental conditions. More generally, they also show the importance of studying dynamic genetic influences across the life course and their differential effects across the phenotype distribution.

Posters accepted as oral presentations on March 13

Poster	*Area*	
O1	A	PRKD2 as a candidate gene modulating T cell activation in familial Systemic sclerosis - ***Pierre Maus***
O2	A	Revealing the developmental-genetic basis for the human cranial vault - ***Jay Devine***
O3	A	Biallelic GAD2 variants cause an early-onset developmental encephalopathy - ***Mathijs van der Lei***
O4	A	Unravelling shared pathomechanisms in syndromic thoracic aortic aneurysm disorders using single-cell RNA sequencing - ***Irene Valdivia***
O5	A	Clinical evaluation of long-read sequencing-based episignature detection in developmental disorders - ***Benjamin Huremagic***
O6	A	Functional validation of novel TIE1 variants as causes for primary lymphedema - ***Lucas Potier***
O7	A	Unraveling VUS in BRAF: A path to improve diagnosis, prognosis, and therapy in cancer patients - ***Dries Vanisterbecq***
O8	A	A novel form of autosomal dominant spondylocostal dysostosis in three unrelated families caused by the same heterozygous pathogenic variant in MESP2 - ***Aneta Malesa***
O9	A	Using the Genetically Encoded Calcium Indicator NCaMP7 in hiPSC-derived cardiomyocytes to model Brugada Syndrome - ***Dogan Akdeniz***
O10	A	Cardiovascular effects of fibrillin impairment in zebrafish - ***Marina Horvat***
O11	A	Methylation Biomarkers can Distinguish Pleural Mesothelioma from Healthy Pleura and other Pleural Pathologies - ***Janah Vandenhoeck***
O12	A	BabyDetect project: interim results and some challenges in newborn screening - ***Kristine Hovhannesyan***
O13	A	Diagnosing HEreditary predisposition syndromes for Childhood cancer: Implementation in clinical PRactice (DHECIPR)– preliminary results - ***Robin De Putter***
O14	A	Genetic background of patients with childhood-onset cardiomyopathy: results from a retrospective cohort study - **Wannes Renders**
O15	A	Discovery, replication and characterization of protein, in vitro and iPSC-RPE stem cell models of a novel dominant RPE65- retinopathy, an actionable RPE disease - ***Eline Van Vooren***
O16	A	Unravelling the proxisome of photoreceptor-specific nuclear receptor NR2E3 reveals a potential molecular link between transcriptional regulation and splicing in human retina - ***Edith De Bruycker***
O17	A	Structural variants disrupt a critical regulatory region downstream of FOXG1 - ***Lisa Hamerlinck***

Poster presentations

Poster	*Area*	
P1	A	Development of a CRISPR/Cas9-engineered zebrafish model to investigate the role of tgfb2 in Loeys-Dietz syndrome - ***Justine Verbiest***
P2	A	DiGerential impact of EPHB4 likely pathogenic variants between lymphatic and CM-AVM2-related phenotypes - ***Héloïse Poullet***
P3	A	Impact of selection and medieval migrations on human genetic diversity in Northwest Europe - ***Toomas Kivisild*** (absent)
P4	A	Proof-of-concept of DNA methylation-based multi-cancer detection in liquid biopsies using IMPRESS - ***Nele De Meulenaere***
P5	A	Family-level impact of extensive germline predisposition screening in childhood cancer: A multi family member interview analysis in parents - ***Sophie Van Hoyweghen***
P6	A	Pathogenic variants in HGF give rise to childhood-to-late onset primary lymphoedema by loss of function - ***Pascal Brouillard***
P7	A	Role of a rare variant in the NT5E gene in the pathogenesis of Systemic Sclerosis - ***Gaëlle Tilman***
P8	A	Cell-free DNA methylation analysis reveals tissue of origin dynamics in health - ***Mio Aerden***
P9	A	Constitutional Mismatch Repair Deficiency (CMMRD) syndrome: A Case Report of a Patient with biallelic germline PMS2 Mutations - ***Emeline Bollaert***
P10	A	UMOD Genotype and Determinants of Urinary Uromodulin in African Populations - ***Eric Olinger***
P11	A	Towards patient-specific aorta-on-a-chip models for thoracic aortic aneurysm and dissection - ***Amira Bousbaa***
P12	A	Finding the missing piece: Cowden syndrome - a case report - ***Joelle EL HAJJ***
P13	A	Investigation of ROS blocking strategy to prevent aneurysm formation in Marfan Syndrome - ***Lucia Buccioli***
P14	A	Variant of uncertain significance testing in zebrafish: a proof of concept using a COL1A2 variant as example - ***Michiel Vanhooydonck***
P15	A	Elucidating the pathomechanisms of Meester-Loeys syndrome: Insights from a Bgn-/0 mouse model - ***Anne Hebert***
P16	A	Zebrafish as a model for Myhre syndrome: growth deficits and vascular narrowing - ***Eva Vanbelleghem***
P17	A	In Vivo Functional Evaluation of BRCA2 Variants via CRISPR-mediated Knock-In in Zebrafish - ***Elyne De Neef***
P18	A	Biases in the Parsortix® system observed with pancreatic cancer cell lines - ***Nele Vandenbussche***
P19	A	Diagnostic Yield and Clinical Impact of Prenatal Whole-Exome Sequencing (WES) – Four-Year Single Center Experience - ***Katleen Janssens***
P20	A	Cancer risk in RASopathies - ***Katleen Janssens***

Poster presentations

Poster	*Area*	
P21	A	A large deletion responsible for ADTKD-UMOD - ***Sorya Fagnoul***
P22	B	ATRX syndrome diagnosed through methylation and RNA analysis: A case highlighting the benefits of an integromic approach - ***Sebastian Neuens***
P23	B	Uncovering transcriptomic signatures associated with resistance to anti-EGFR therapy in colorectal cancer - ***Ana Regina de Abreu***
P24	B	Shared genetic architecture of brain shape and IBD is specifically enriched for inflammatory response genes - ***Seppe Goovaerts***
P25	B	First report of a homozygous SYCP2 variant implicated in female infertility due to embryo developmental arrest - ***Annelore Van Der Kelen***
P26	B	Assessment of gene–disease associations and recommendations for genetic testing for somatic variants in vascular anomalies by VASCERN-VASCA - ***Nicole Revencu***
P27	B	Altered collagen receptors and ECM organisation in dermal fibroblast cultures from patients with classical Ehlers-Danlos Syndrome - ***Ruben Vanlerberghe***
P28	B	A PDGFRB splice site variant associated with familial infantile myofibromatosis and resistance to imatinib - ***Boutaina Boulouadnine***
P29	B	Decreased local TGF-beta sequestration in a mutant fibrillin-1 mouse model leads to thoracic aortic wall damage - ***Maarten Dhaese***
P30	B	Understanding human linkeropathies: generation of knock-in models to study of the consequences of defective proteoglycan biosynthesis - ***Jana De Troyer***
P31	B	Characterization of endothelial cell subtypes in Venous Malformations including the Blue Rubber Bleb Nevus syndrome by snRNA-seq - ***Nicolas Huyghe***
P32	B	Assessing the eGect of aberrant FOXG1 expression in neural organoids - ***Nore Van Loon***
P33	B	Incidental finding of X-linked periventricular heterotopia in a male presenting joint hypermobility - ***Sybille Collin***
P34	B	A first case of desmin-related myofibrillar myopathy due to inheritance from a confirmed mosaic asymptomatic carrier - ***Sophie Uyttebroeck***
P35	B	PDGFRB Variants in Hereditary Progressive Mucinous Histiocytosis - ***Camille Leducq***
P36	B	RNA-sequencing unveils novel FLT4 splice site variants in isolated CHD - ***Maxim Verlee***
P37	B	Distinct genetic patterns involving multilocus inheritance and unique and novel variants drive a high diagnostic yield in an understudied consanguineous retinopathy cohort - ***Lieselot Vincke***
P38	B	Unexpected STK11 Gene Deletion in a Breast Cancer Patient: Implications for Cancer Predisposition Panels - ***Edith Sepulchre***
P39	B	Germline mutational landscape in pediatric cancers and disease relevance: : Insights from a Canadian Experience - ***Edith Sepulchre***
P40	B	Cutis Laxa type 2E: report of a new case highlighting hypotonia as a major feature - ***Claire Fouquet***

Poster Area

P41 B Atypical glutamic acid to lysine substitution in the triple helix of type III collagen manifests as overlap between classical and vascular Ehlers-Danlos syndrome - **Margot Callens**

P42 B Mutant zebrafish model in the search for new therapies for childhood chronic kidney disease - ***Pauline Van Wesemael***

P43 B Insights in COL4A1 and COL4A2 Variants through Comprehensive Genotype-Phenotype Mapping - ***Merlijn Nemegeer***

P44 B Filamin C associated cardiomyopathy in pediatric patients: a Belgian case series and literature review - ***Wannes Renders***

P45 B Immunogenomics solves missing heritability and ends the diagnostic odyssey in RAG1-SCID - ***Marieke De Bruyne***

P46 B Prevalence of intronic AAGGG repeat expansion in RFC1 in Belgian patients - ***Amélie Van Eesbeeck***

P47 B Introduction of a new diagnostic test for the detection of a GAA expansion in the FGF14 gene - ***Amélie Van Eesbeeck***

P48 B Nuclear metabolism in the control of DNA methylation in tumour hypoxia - ***Roxane Verdikt***

P49 B Unraveling the enhancer - promoter interaction networks that drive craniofacial development - ***Aikaterini Rapti***

P50 B Nanopore long read whole genome sequencing in developmental disorders - ***Mathilde Geysens***

P51 B GIPXplorePLUZ: a comprehensive ctDNA test using multi-omics to enhance cancer detection and diagnosis - ***Wouter Bossuyt***

P52 B Unraveling syndromic traits in dual genetic diagnoses with 3D facial phenotyping - ***Michiel Vanneste***

P53 B Genetic yield in a large ataxia and spastic paraplegia cohort in Flanders - ***Sien Van Daele***

P54 B Genetic risk stratification in the Leuven familial pulmonary fibrosis cohort - ***Nathalie Veyt***

P55 B Breaking the mold: exploring the impact of sex on phenotypical features and challenges in Marfan Syndrome - ***Lotte Van Den Heuvel***

P56 B Clinical and Molecular Characterization of a Novel Homozygous MTRFR Missense Variant Associated with Spastic Paraplegia, Peripheral Neuropathy and Optic Atrophy - ***Claire Fouquet*** (absent)

P57 B Implication of large heterozygous deletions in the GBA gene in Parkinson disease - ***Emilie Guerit***

P58 B Residual Protein Function Shapes Immune Dysregulation in Loss-of-Function RC3H1 Mutations - ***Simon Tavernier***

P59 B The role of the meiotic chromosome axis in the assembly of the DNA double-strand break machinery - ***Taniya Roy***

P60 B Insights in underlying pathophysiology of microcephaly with simplified gyral pattern associated with VRK1 pathogenic variants derived from fetal neuropathology - ***Essier Aude***

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BeSHG

Thank you for your attendance

Organizing committee

Emeline Bollaert, Armelle Duquenne, Joëlle El Hajj, Jinine Hatoum, Rodolphe Michiels, Magali Philippeau , Laïla Zahed

Scientific committee

Prof. Jean-Baptiste Demoulin, Prof. Nisha Limaye, Prof. Yves Sznajer, Prof. Miikka Vikkula, Prof. Laïla Zahed

UCLouvain

Save the Date
Joint NVHG/BeSHG meeting
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