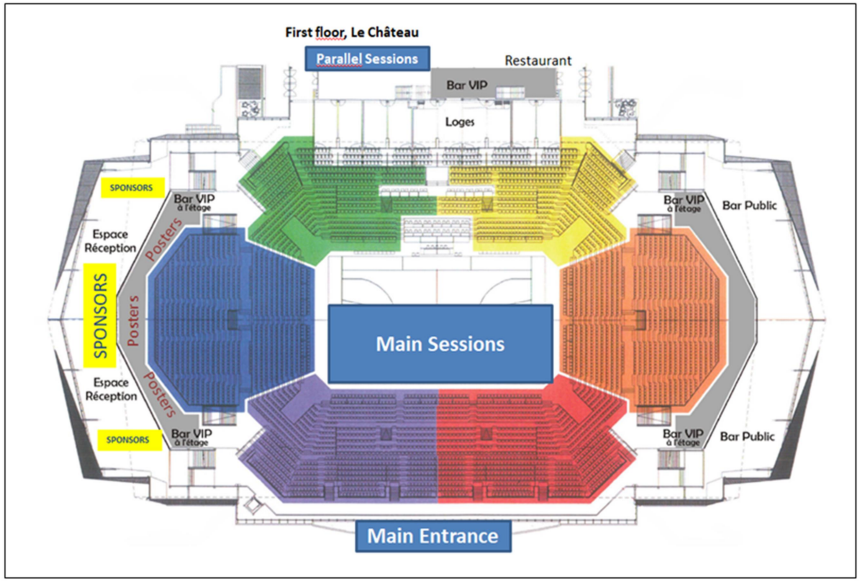
17 March 2023

BeSHG Annual Meeting

To DNA and beyond

AstraZeneca illumina® Roche

Dôme Plan



Sponsors, catering, coffee breaks & party area
Espace Réception

Main Sessions
On the parquet floor

Parallel Sessions
On first floor, Room Le Château
Main sessions room (parquet floor)

Posters
VIP Bar (1st floor of the Espace Réception)

Shuttle bus
Waiting in front of the main entrance. Will not be available after 9.00pm.

Dear all, members of the Belgian Society for Human Genetics,

It is my pleasure to welcome you in Charleroi!

This year's programme headlines the evolution and embryology of sex determination and the legal aspects of early surgical interventions on intersex children. The second theme is on the challenge of diagnosing and treating methylation disorders. We sincerely thank the invited speakers for joining us and for sharing their latest insights and results with us.

These are this year's highlights indeed, but note that our annual meeting covers all aspects of human genetics, including clinics, research, diagnostics, society and ethics. We sincerely thank everyone who contributes to the wealth of this meeting with an oral or a poster presentation.

We thank the sponsors for their essential and generous financial contribution to the organisation of the meeting. We invite especially the young attendants and trainees in the labs to interact with the companies' representatives to hear about their latest innovations.

We continue to live the heydays of genetics and genomics. Our professional society has a role in promoting scientific research, in its broadest sense, but also in the swift and appropriate introduction of genomic medicine in public health, and in the discussions on societal and ethical challenges. I want to call upon our members to engage in community activity, to participate in working groups, to be present in the public forums, etc. Some things within the realm of genetics are turning weird. Activism is opportune when genetics is being misused to discriminate citizens, when care models are developed that do not seem to serve the patients' needs in the first place, and when prices of orphan drugs are called 'marked-conform' while wickedly high…

Our colleagues from the Centre de Génétique at the Institut de Pathologie et de Génétique (IPG) have brought us to the Spiroudome, the home arena of the basketball team Spirou Charleroi. Named after Spirou, the famous comic character. Spirou is globe-trotter, an honest and brave young man of indeterminate age, he tries to fight injustice around him and help people.

It looks like he is a bit of a community activist. So let's be inspired.

I thought that Spirou could be derived from 'inspiration' indeed. It is not, or at least not according to Wikipedia:

Spirou (Walloon pronunciation: [spiˈʀu], [spiˈʀø:]; French: [spiʁu]; Walloon for "squirrel", "mischievous"; Dutch: Robbedoes) is a Belgian comic strip character and protagonist in the comic strip series Spirou & Fantasio and Le Petit Spirou, and the eponymous character of the Belgian comic strip magazine Spirou.

Spirou was originally an elevator operator and bell-boy at the fictional Moustique Hotel. At some point he became a reporter though he remained dressed in his trademark red uniform.

He is usually more level-headed than Fantasio, a journalist who always accompanies him, along with the pet squirrel Spip and the character and fictional animal species Marsupilami.

Spirou addicts shall now yell: 'Houba!'

'Houba!' to our colleagues from the Centre de Génétique at IPG for the compilation of the scientific program and for dealing with all the practical aspects of organising the meeting. We gratefully acknowledge the tremendous effort they have put into the preparation of BeSHG 2023.

Welcome in Charleroi! Enjoy the meeting.

Gert Matthijs
President of BeSHG

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General information

Abstracts
All Abstracts can be found on: https://beshg.be/meeting/abstracts

For the selected oral presentations and posters of the meeting you can use these QR Codes:

Oral presentations

Posters

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

Contact
General Email: damien.lederer@ipg.be

If you have any queries or comments, please do not hesitate to contact a member of staff who will be pleased to help you. You can find the staff in the plenary rooms and at the registration desk.

Covid measurements
The activity takes place with respect for the corona measures in force.

Covid Save Ticket (CST) – Not necessary for this event.

Internet access
Wireless internet is freely available throughout the Dôme. Connect to the Dôme network and access will be granted automatically.

Scientific Session Protocol
Photography, audio or video recording is not permitted.

Presentation
Staff members will be available before the parallel sessions for any questions regarding your presentation.

Poster sessions
Posters will be displayed throughout the conference. Please display your poster in the VIP Bar (1st floor – Espace Réception). A number and place will be assigned for each poster.
Posters must be removed before 4pm.

Content

Program

09:00-09:30 Registration & welcome
Entrance hall

09:30-11:00 PLENARY SESSION 1: SEX DETERMINATION
Parquet floor (Main sessions room)

09:30 **Yann Guigen**: Diversity and evolution of genetic sex-determination in fish.

10:00 **Jana Henck**: Single cell, whole embryo phenotyping of pleiotropic disorders of mammalian development.

10:30 **Kristof Van Assche**: Early surgical interventions on intersex children: Legal considerations and the human rights debate.

11:00-11:30 Coffee break/Meet the sponsors/Poster viewing
Espace Réception & VIP Bar (1st floor - Espace Réception)

11:30-12:45 Selected oral presentations

PARALLEL SESSION 1
Parquet Floor (Main sessions room)

11:30 **David Schröder**: Diamond: AstraZeneca: Use of PARP inhibitor in early breast cancer as adjuvant therapy: new indication for BRCA1 and BRCA2 testing.

11:45 **Michiel Vanneste**: Multivariate GWAS on achondroplasia-like craniofacial shape variation in healthy human individuals.

12:00 **Michiel Vanhooydonck:** The establishment of the first reported zebrafish model for thoracic aortic dissection and rupture.

12:15 **Machteld Baetens**: Preimplantation genetic testing (PGT) in Belgium: national recommendations for genetic centers.

12:30 **Kristine Hovhannesyan**: BabyDetect project: molecular newborn screening in Belgium.

PARALLEL SESSION 2

Le Château room

11:30 **Lisa Dangreau**: The pseudoxanthoma elasticum zebrafish model contributes to novel pathophysiological insights and therapeutic strategies in ectopic mineralization.

11:45 **Bieke Bekaert**: Retained chromosomal integrity following CRISPR/Cas9-based mutational correction in human embryos.

12:00 **Lucia Buccioli**: Striking phenotypical differences between Ipo8 knock- out mouse models on different genetic backgrounds explored by RNA sequencing.

12:15 **Hannes Syryn**: WES advances a genetic diagnosis in patients with differences of sex development and corroborates the role of RXFP2 in autosomal recessive bilateral cryptorchidism and infertility.

12:30 **Hamide Yildirim**: Malfunctional CTDP1 impairs the cell cycle implying its crucial role in brain development

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12:45-14:00 Lunch break and poster viewing

Espace Réception & VIP Bar (1st floor – Espace Réception)

14:00-15:15 Selected oral presentations

PARALLEL SESSION 3

Parquet Floor (Main sessions room)

14:00 **Senne Meynants**: Multiple recombination mechanisms drive the high incidence of 22q11.2 Deletion Syndrome.

14:15 **Dale Annear**: CGG short tandem repeat expansions are overrepresented in neurodevelopmental disorders.

14:30 **Marlies Colman**: Kyphoscoliotic Ehlers-Danlos syndrome due to pathogenic variants in PLOD1 and FKBP14: further insights into the pathophysiology and comparison of clinical characteristics.

14:45 **Margot van Riel**: Single-cell dissection of cervix and placenta reveal both novel and overlapping cell types.

15:00 **Liene Thys**: To test or not to test: the importance of genetics in the diagnostic workup of cerebral palsy.

PARALLEL SESSION 4

Le Château room

14:00 **Richard Capper**: Diamond: Illumina: Advancements in Whole-Genome Sequencing – Achieving a complete and comprehensive genome at scale.

14:15 **Miriam Bauwens**: Whole genome sequencing sheds light on the dark matter of the genome in patients with inherited retinal diseases.

14:30 **Laurens Hannes**: Differential alternative splicing analysis links variation in ZRSR2 to a novel oral-facial-digital syndrome.

14:45 **Eline Van Vooren**: An in vitro enzymatic assay to elucidate the VUS problem in RPE65, a target for retinal gene therapy.

15:00 **Benjamin Huremagic**: Sample Catalog: A Federated Platform for Identification and sharing of biological samples in rare disorders.

15:15-15:45 Coffee break/Meet the sponsors/Poster viewing
Espace Réception & VIP Bar (1st floor – Espace Réception)

15:45-16:15 General Assembly

16:15-17:30 PLENARY SESSION 2: METHYLATION

Parquet floor (Main sessions room)

16:15 **Deborah Mackay**: It takes two: genetics and epigenetics of imprinting disorders.

16:45 **Ulrich Zechner**: Derepression of imprinted gene alleles as a new approach to treat imprinting disorders.

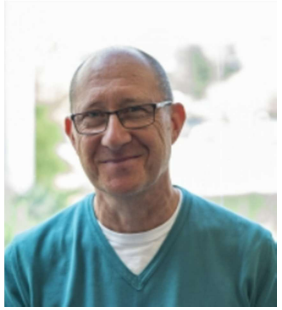
17:15 Closing remarks and best poster/oral presentations awards

17:30-23:00 Reception and party
Espace Réception

Abstracts can be found on: https://beshg.be/meeting/abstracts

QR Code:

Invited speakers



Diversity and evolution of genetic sex-determination in fish

Yann Guiguen, Directeur de Recherche, INRAE, Rennes, France

Despite the quite conserved and strict genetic sex determination (GSD) systems of most mammals and birds, other vertebrates display a great variety of sex determination mechanisms ranging from a complete control of the environment (Environmental Sex Determination = ESD) to a strict genetic control (GSD).

Fishes are a very good examples of this high diversity with ESD, GSD and mixed systems quickly evolving in many closely related species. In species with GSD, frequent changes of sex chromosomes (the high turnover model) are found in fishes, with switches from male-heterogametic (XX/XY) to female-heterogametic (ZZ/ZW) GSD systems (or the opposite) evolving on short evolutionary periods, and sometime even within different populations of the same species.
Even more complicated genetic systems can be found with polygenic sex determination relying on a single pair or multiple pairs of sex chromosomes, or species relying for sex determination on a supernumerary and accessory B-type chromosome. Finally, fish master-sex determining genes (MSD) acting at the top of the sex determination pathway also exhibit frequent changes with a high diversity of MSD genes.

Single cell, whole embryo phenotyping of pleiotropic genetic disorders of mammalian development

Jana Henck, PhD student, Max Planck Institute, Berlin, Germany

Mouse models represent a critical tool to study human genetic diseases, as well as for advancing our general understanding of developmental biology. However, current methods lack the necessary throughput and resolution for measuring phenotype at the scale of whole organisms. In our study, we set out to establish single cell RNA sequencing as a tool for large scale standardized and comprehensive phenotypic analysis of whole mouse mutant embryos. In a multiplexed experiment, we applied single cell RNA sequencing to profile 104 whole mouse embryos from 22 different mutants and 4 wildtype strains at embryonic stage E13.5. The mouse mutants range from established multisystem disorders to single enhancer deletions. To analyze the resulting dataset of 1.9 million cells we developed and applied several analytical frameworks, successfully detecting differences in composition and gene expression changes across all cell types and trajectories. Some mutants exhibited changes in dozens of trajectories (e.g., the pleiotropic consequences of altering the Sox9 regulatory landscape) whereas the impact of others was considerably more restricted. We further identified differences between widely used wildtype strains, compared gain vs. loss of function mutants, and identified changes that are shared by heretofore unrelated models.
Overall, our findings show how single cell profiling of whole embryos can enable the systematic molecular and cellular phenotypic characterization of mouse mutants with unprecedented breadth and resolution.

Early surgical interventions on intersex children: legal considerations and the human rights debate

Kristof Van Assche, Research Professor in health law & Professor of philosophy of law, University of Antwerp, Belgium

So-called “intersex” children or, more neutral, children with variations of sex development are born with atypical sex characteristics. Some of these children suffer from conditions that require urgent medical intervention, but the great majority are physically healthy. Yet, until recently, the prevailing view and practice was that, despite the absence of medical urgency, these children should as early as possible be subjected to surgical and/or hormonal intervention to reinforce the gender that they are assigned at birth by the parents and medical team. Following evidence of high levels of gender dysphoria and other severe mental health problems in persons whose actual gender identity at a later age differed from the gender they had surgically been assigned as an infant, international human rights organisations are calling for a prohibition of these interventions until the child can give valid consent. As a result, since 2015, countries such as France, Germany, Iceland, and Malta have adopted legislation to that effect, albeit with a considerable variety of approaches, and a legislative proposal is currently also under discussion in Belgium. In this presentation, the ethical and legal issues involved in allowing or prohibiting early surgical interventions in children with variations of sex development will be examined.

It takes two: an update on the genetics and epigenetics of clinical imprinting disorders

Deborah Mackay, Professor of Medical Epigenetics, Univeristy of Southampton, UK

Imprinting disorders are a group of rare congenital disorders with clinical features impacting growth, development, metabolism and behaviour; they are caused by altered expression of imprinted genes, which are normally expressed from only one parental allele, while the other parental allele remains silent. Imprinting disorders can be caused by coding mutations or copy number changes, chromosomal changes, DNA methylation disturbance altering imprinted gene expression in *cis*, or multi-locus imprinting disturbance (MLID) associated with environmental insults or trans-acting genetic variants.

Ongoing research continues to widen the clinical heterogeneity of imprinting disorders, posing ongoing challenges for diagnosis, family counselling, and the targeted management that minimises long-term morbidities. By the same token, the increasing diversity of recognised epigenetic and genetic causes presents a constant challenge for molecular testing, interpretation and reporting.

I will explore recent findings and outstanding questions in genetic and epigenetic causes of imprinting disorders, and current approaches for clinical and molecular diagnosis.

Derepression of imprinted gene alleles as a new approach to treat imprinting disorders

Prof. Dr. Ulrich Zechner, Johannes Gutenberg University, Mainz, Germany

Current treatment strategies of imprinting disorders mainly focus on the alleviation of some of the symptoms like behavioural and endocrine manifestations. There are no treatments specifically for clinical signs like intellectual disability which is an overarching finding associated with several imprinting disorders such as Angelman syndrome (AS) and Birk-Barel syndrome (BIBARS). Imprinting disorders with loss or mutation of the active copy of an imprinted gene offer unique possibilities of causal therapeutic intervention by derepression of the other silent allele. Novel derepression-based therapeutic approaches using epigenetic drugs, antisense-oligonucleotides (ASOs) or epigenome editing, their preclinical application in mouse models of AS and BIBARS as well as recently started first-in-humans studies with Angelman-ASOs will be presented.

Selected oral presentations

No	Name	Title
Parallel session 1:		
	David Schröder	Diamond: AstraZeneca: Use of PARP inhibitor in early breast cancer as adjuvant therapy: new indication for BRCA1 and BRCA2 testing
O1	Michiel Vanneste	Multivariate GWAS on achondroplasia-like craniofacial shape variation in healthy human individuals
O2	Michiel Vanhooy-donck	The establishment of the first reported zebrafish model for thoracic aortic dissection and rupture
O3	Machteld Baetens	Preimplantation genetic testing (PGT) in Belgium: national recommendations for genetic centers
O4	Kristine Hovhannes-yan	BabyDetect project: molecular newborn screening in Belgium
Parallel session 2:		
O5	Lisa Dangreau	The pseudoxanthoma elasticum zebrafish model contributes to novel pathophysiological insights and therapeutic strategies in ectopic mineralization
O6	Bieke Bekaert	Retained chromosomal integrity following CRISPR/Cas9-based mutational correction in human embryos.
O7	Lucia Buccioli	Striking phenotypical differences between Ipo8 knock-out mouse models on different genetic backgrounds explored by RNA sequencing
O8	Hannes Syryn	WES advances a genetic diagnosis in patients with differences of sex development and corroborates the role of RXFP2 in autosomal recessive bilateral cryptorchidism and infertility
O9	Hamide Yildrim	Malfunctional CTDP1 impairs the cell cycle implying its crucial role in brain development
Parallel session 3:		
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O12	Marlies Colman	Kyphoscoliotic Ehlers-Danlos syndrome due to pathogenic variants in PLOD1 and FKBP14: further insights into the pathophysiology and comparison of clinical characteristics
O13	Margot van Riel	Single-cell dissection of cervix and placenta reveal both novel and overlapping cell types
O14	Liene Thys	To test or not to test: the importance of genetics in the diagnostic workup of cerebral palsy

No	Name	Title
Parallel session 4:		
	Richard Capper	Diamond: Illumina: Advancements in Whole-Genome Sequencing – Achieving a complete and comprehensive genome at scale
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O16	Laurens Hannes	Differential alternative splicing analysis links variation in ZRSR2 to a novel oral-facial-digital syndrome
O17	Eline Van Vooren	An in vitro enzymatic assay to elucidate the VUS problem in RPE65, a target for retinal gene therapy
O18	Benjamin Huremagic	Sample Catalog: A Federated Platform for Identification and sharing of biological samples in rare disorders

Abstracts can be found on: https://beshg.be/meeting/abstracts, **QR CODE:**

Poster presentations

No.	Name	Title
P01	Randy Osei	Results From A Large Prospective Genetic Study In Children With Brugada Syndrome
P02	Sonia Van Dooren	The prognostic role of (predicted) null variants in Brugada syndrome
P03	Meng Yuan	Assessing univariate facial phenotyping approaches in GWAS
P04	Chatelain Camille	Case report: Missense variant c.593A>G p.(Lys198Arg) in CSNK2A1, a recurrent variant in Okur-Chung neurodevelopmental syndrome
P05	Candy Kumps	Report of a novel familial DNMT3A variant causing a variable clinical phenotype of Tatton-Brown-Rahman syndrome
P06	Sophie Uyttebroeck	Immunomodulatory therapy helps patient with biallelic FLG variants become less itchy
P07	Marie De Borre	A Case of Microphthalmia and Bilateral Papillary Coloboma Associated With a New Gain-of-Function Mutation of RBP4
P08	Anne Hebert	Expanding genotype-phenotype associations in BGN-related Meester-Loeys syndrome
P09	Ewa Hordyjewska-Kowalczyk	Update on the allelic heterogeneity and phenotypic diversity in CBFB-related cleidocranial dysplasiaLoeys-Dietz Syndrome type 3 patient with a SMAD3 p.Arg287Gln mutation
P10	Lena Kukor	Evolution of Ehlers-Danlos Syndrome consultations in the university hospital of Liege

P11	Laura Bourlard	MIRAGE syndrome caused by a pathogenic variant c.1376G>A (p.Arg459Gln) in the SAMD9 gene with presumed maternal gonadal mosaicism : report of a family with two affected sisters.
P12	Julie Crèvecoeur	How to adapt clinical practice for genetic testing when the index case is negative: a case report.
P13	Hanne Boen	Characterization of a Belgian pathogenic founder variant in MYBPC3
P14	Sien Van Daele	Genetic variability in known ALS genes in sporadic ALS patients
P15	Amina Ameli	Familial Mediterranean fever in Moroccan population
P16	Lore Pottie	The clinical use of exome sequencing to diagnose PCD patients
P17	Eleonore Pairet	PDGFC : new candidate gene for cleft lip and palate
P18	Ilse Parijs	Population screening for 15q11-q13 duplications: corroboration of the difference in impact between maternally and paternally inherited alleles
P19	Nicole Revencu	Two Mosaic ENG mutations in a patient with hereditary hemorrhagic telangiectasia
P20	Jo Sourbron	Genetics in Sunflower syndrome: GABRG2 as a culprit gene?
P21	Natacha Leroi	Discordancy in interpretation of a BRCA1 variant in the Belgian population.
P22	Elise Pelgrims	Triplications of chromosome 1p36.3, including the genes gabrd and ski, are associated with a developmental disorder and recurrent facial features

P23	Lieselot Vincke	Characterization of the genetic architecture of inherited retinal disease in a consanguineous Iranian cohort, an understudied population
P24	Sophie Van Hoyweghen	The psychological impact of genetic screening in childhood cancer: A systematic review
P25	Florence Arts	Unravelling a familial case of DOK7 congenital myasthenic syndrome by analyzing RNA in patient's cultured cells
P26	Mio Aerden	The neurodevelopmental and facial phenotype in individuals with a TRIP12 variant
P27	Maria Valeria Freire Chadrina	Coinheritance of pathogenic variants in ATM and BRCA2 in families with multiple cancers: a case series
P28	Tessier Aude	Bi-allelic variations in CRB2, encoding the Crumbs Cell Polarity Complex Component 2, lead to non-communicating hydrocephaly due to atresia of the Aqueduct of Sylvius and central canal
P29	Perrine Masson	Pre and postnatal follow-up of a boy affected with 3M syndrome diagnosed in utero by trio exome analysis
P30	Pascal Brouillard	Biallelic ANGPT2 loss-of-function causes severe early onset non-immune hydrops fetalis
P31	Julie Désir	Discordant fetal sex on NIPT and ultrasound. Monocentric retrospective study
P32	Colombine Meunier	Prenatal diagnosis of de novo RAC3 variant associated to severe cerebral malformations and bilateral clubfoot

P33	Benoit Parmentier	Unexpected 6 Mb terminal 10p balanced rearrangement in a man revealed by an abnormal Non Invasive Prenatal Testing (NIPT)
P34	Mariia Butynets	A ticking biological clock in men: age-related alterations in the human sperm methylome and risk for diseases in offspring
P35	Annelore Van Der Kelen	A systematic review of monogenic gene-disease relationships in human female infertility and differences in sex development
P36	Nathalie Veyt	Expanding the phenotype of copy number variations involving NR0B1 (DAX1)
P37	Nathalie Vanden Eynde	Prenatal diagnosis of a homozygous nonsense MPDZ variant in a foetus with severe congenital hydrocephalus
P38	Sonia Rombout	The IPG experience in maternal cancer detection by noninvasive prenatal testing (NIPT) – retrospective study.
P39	Elyne De Neef	Exploring synthetic lethality caused by novobiocin in homologous recombination-deficient zebrafish: a novel preclinical in vivo model for POL inhibitor development?
P40	Irene Valdivia	An optimised protocol for isolation of single cells from mouse aorta for single-cell RNA sequencing
P41	Dogan Akdeniz	Developing a dual-reporter iPSC-line using Genetically Encoded Calcium and Voltage Indicators (GEVI/GECI)
P42	Marc Terrones	Optimizing monoclonal establishment of CRISPR/Cas9-edited non-small cell lung cancer lines

P43	Tamara Jarayseh	Identification of modifier genes underlying intra-familial phenotypic variability in zebrafish OI models
P44	Andy Willaert	Zebrafish Facility Ghent – A new Ugent CORE Facility
P45	Sophie Debaenst	Proteomic analysis of the vertebral column in dominant OI zebrafish models reveals biomarkers for phenotypic outcome
P46	Tessi Beyltjens	Artificial intelligence-assisted exome analysis for hearing loss
P47	Erika D'haenens	The added value of RNA-sequencing in exome variant interpretation
P48	Erika Souche	Comparison of accuracy of short and long read sequencing technologies
P49	Sandrine Mary	Use of Oxford Nanopore Long Read Sequencing to solve complex genetic cases
P50	Griet De Clercq	Long-read sequencing resolves (complex) cryptic structural variation in patients with syndromic intellectual disability
P51	Seppe Goovaerts	GWAS conditioned by SNP-specific shape effects identifies 11 loci underlying shape covariation of the cerebral cortex and cranial vault
P52	Lisa Hamerlinck	An optimized workflow for CRISPR/Cas9-mediated generation of indels and large deletions in induced pluripotent stem cells and neural stem cells
P53	Nishkala Sattanathan	WiNGS - Federated approach for genomics data sharing and analysis
P54	Dhanya Sudhakaran	Methylation-based deconvolution of cell-free DNA allows for non-invasive multi-cancer typing

P55	Ana Regina de Abreu	Genome-wide DNA methylation sequencing methods for cancer biomarker discovery: their potential and limitations
P56	Mathilde Geysens	Long read whole genome sequencing for the detection of structural and epigenetic variation in developmental disorders
P57	Olivier Monestier	Digital droplet PCR: an efficient, rapid, and simple methodology for CNV confirmation
P58	Olivier Monestier	Our Whole Exome Sequencing analysis at IPG: Outcomes of the first 1100 patients
P59	Emilie Guerit	Study of nonsense-mediated decay triggered by splicing variants: interest of purromycin use?
P60	Nathalie Lannoy	The Belgian Genetic Tests Database: a centralized and accessible tool for genetic testing allowing transparent access to comprehensive and quality information
P61	Sarah Delbaere	Challenges with a MAGEL2 inframe deletion variant: two clinically distinct families and sequencing issues

Abstracts can be found on: https://beshg.be/meeting/abstracts, **QR CODE:**

Notes

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Notes

We thank you for your numerous attendance
and look forward to see you again next year
for the:

BeSHG annual meeting 2024

12 April 2024
At
KULeuven

For the Belgian Society of Human Genetics

The Board:

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