Reproductive Genetics

08.00-09.00 REGISTRATION

09.00-09.15 WELCOME ADDRESS BY THE PRESIDENT OF THE BESHG PROF. G. MATTHIJS

09.15-10.30 PLENARY SESSION 1

Chairs: Frederik Hes & Willem Verpoest

- **Prof. Guido de Wert**

  Expanded carrier screening: Ethical aspects

- **Prof. Claudia Spits**

  Mitochondrial DNA and Reproduction

10.30-11.00 BESHG ANNUAL GENERAL ASSEMBLY

11.00-11.30 COFFEE BREAK / MEET YOUR SPONSOR / POSTER VIEWING

11.30-13.00 SELECTED ORAL PRESENTATIONS

- **Parallel Session I:**

  Chairs: Julie Soblet & Bert Callewaert

11.30 - 11.45 Dr Loverix
"HRD and GIS in ovarian cancer: does one size really fit all?"
Astra Zeneca - Diamond Sponsor

11.45 - 12.00 Lukas Nollet, Matthias Van Gils, Andy Willaert, Paul J. Coucke & Olivier M. Vanakker
Center for Medical Genetics, Ghent University Hospital, Belgium

Minocycline attenuates excessive DNA damage response and reduces ectopic calcification in pseudoxanthoma elasticum

12.00 - 12.15 Bieke Bekaert[1], Annekatrien Boel[1], Lisa De Witte[2], Mina Popovic[1], Panagiotis Stamatiadis[1], Ramesh R. Gugalli[1], Swati Mishra[1], Gwenny Cosemans[1], Lise Tordeurs[1], Susana M. Chuva de Sousa Lopes [3], Petra De Sutter[1], Björn Menten[2], Dominic Stoop[1], Paul Coucke[2] & Björn Heindryckx[1]
[1] Ghent-Fertility And Stem cell Team (G-FaST), Department for Reproductive Medicine, Ghent University Hospital, Corneel Heymanslaan 10, 9000 Ghent, Belgium.
[2] Center for Medical Genetics Ghent, Ghent University, Department of Biomolecular Medicine, Corneel Heymanslaan 10, 9000 Ghent, Belgium.
[3] Department of Anatomy and Embryology, Leiden University Medical Center, Leiden, the Netherlands.

A proof-of-concept study to investigate DNA repair mechanisms in the human embryo after the application of CRISPR/Cas9 gene editing in the germ line

12.15 - 12.30 Laurenz De Cock[1], Joline Ingels[2], Rupert Mayer[1], Saskia Desmet[3], Kelly Heyns[4], Nele Lootens[3], Marieke Brusseel[3], Francis Impens[1], Karim Vermaelen[4], Björn Menten[1] & Bart Vandekerckhove[2]
[1] Department of Biomolecular Medicine, Ghent University
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[3] GMP Unit Cell Therapy, Department of Regenerative Medicine, Ghent University Hospital
[4] Department of Respiratory Medicine, Ghent University Hospital

Interim results of a phase i neoantigen-directed personalized cancer vaccine trial

12.30 - 12.45 Charlotte Fieuws[1], Bram Parton[1], Olivier De Wever [2], Hannelore Denys[3], Koen Van De Vijver[4] & Kathleen Claes[1]
[1] Center for Medical Genetics, Ghent University, Ghent, Belgium
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[4] Department of Anatomical Pathology, Ghent University Hospital, Ghent, Belgium

Heading towards an in vivo predictive test for personalized ovarian cancer treatment: application of novel therapies in zebrafish patient derived xenografts

12.45 - 13.00 Jonas Demeulemeester[1], Stefan C Dentro[2], Moritz Gerstung[2] & Peter Van Loo[3]
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Biallelic mutations in cancer genomes reveal local mutational determinants

- **Parallel Session II:**

  Chairs: Nisha Limaye ,Saskia Bulk

11:30 - 11.45: Koen Weenink
"MagnisDx, your solution for an urgent workflow"
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11.45 - 12.00 Elia Fernandez Gallardo[1,*], Alejandro Sifrim[1,*], Joel Chappell[2], Jonas Demeulemeester[1], Jenniffer Clara Herrmann[1], Robin Vermotte[1], Alison Kerremans[1], Michiel Van der Haegen[1], Daniel Brown[1], Koen Theunis[1], Jens Van Herck[1], Katy Vandereyken[1], Chris Ponting[1], Joris Vermeesch[4], Karen Peeraer[3], Sophie Debrock[3], Vincent Pasque[2,#] and Thierry Voet[1,4,5#].

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A multi-omics genome-plus-transcriptome single-cell atlas of human preimplantation development reveals the impact of chromosome instability on cell function within the embryo

12.00 - 12.15: Yassmine Ayeb[1], Tatjana Jatsenko[1], Cindy Melotte[2], Olga Tsuiko[2], Joke Allemeersch[2], Sophie Debrock[3], Ellen Denayer[2], Eric Legius[2], Hilde Brems[2], Joris Robert Vermeesch[1] & Eftychia Dimitriadou[2]

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[3] Leuven University Fertility Center, University Hospitals Leuven, Leuven, Belgium

Comprehensive PGT for patients with de novo pathogenic variants following single-molecule long read amplicon sequencing based haplotyping

12.15 - 12.30: Simon Boutry[1], Raphael Helaers[2], Tom Lenaerts[3] & Miikka Vikkula[4]

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Walloon Excellence in Lifesciences and Biotechnology (WELBIO), Université catholique de Louvain, Brussels, Belgium

A novel tool to analyse oligogenic diseases

12:30 - 12.45: Victor Lopez Soriano, Stijn Van de Sompele, Alfredo Dueñas Rey, Eva D'haene, Miriam Bauwens & Elfride De Baere
Ghent University

Mapping the cis-regulatory landscape of ABCA4 in adult human retina

12.45 - 13.00: Dale John Annear[1], Geert Vandeweyer[1], Ellen Elinck[1], Alba Sanchis-Juan[2], Courtney E French[2], Lucy Raymond[2] & Frank Kooy[1]
[1] University of Antwerp
[2] University of Cambridge

Inheritance Patterns of the CGG Short Tandem Repeat

13.00-14.15 LUNCH BREAK / MEET YOUR SPONSOR / POSTER VIEWING

14.15-15.30 SELECTED ORAL PRESENTATIONS

- **Parallel Session III**

Chairs: Katrien Stouffs & Damien Lederer

14.15 - 14.30 Munevver Burcu Cicekdal[1], Fabiola Ceroni [2], Richard Holt[2], Dorine Bax[2], Julie Plaisancie [3], Nicolas Chassaing[3], Patrick Calvas[3], Alfredo Duenas Rey [4], Elfride De Baere[4], Nicola K Ragge[2] & Kris Vleminckx [5]
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[5] Department of Biomedical Molecular Biology, Ghent University, Belgium
Generation of mab21l2 crispants in Xenopus tropicalis reveals the importance of non-genic elements in human developmental eye anomalies

14.30 - 14.45 Stijn Van de Sompele[1], Caroline Van Cauwenbergh[2], Marjolein Carron[3], Alfredo D Rey[1], Hong T Tran[4], Toon Rosseel[1], Frauke Coppieters[1], Robrecht Cannoodt[5], Thalia Van Laethem[1], Brecht Guillemyn[1], Dimitri Roels[2], Helena Flipts[1], Erik Fransen[6], Marleen Praet[7], Thomas Langmann[8], Katharina Dannhausen[8], Karin Dahan[9], Kris Vleminckx[3], Wendy Chung[10], Andrew R Webster[11], Bart P Leroy[12] & Elfride De Baere[1]
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Dual molecular effects of dominant SF3B2 variants cause a novel spliceosomopathy displaying retinitis pigmentosa or developmental skeletal anomalies

14.45 - 15.00 Brecht Guillemyn[1], Hanna De Saffel[1], Jan Willem Bek[1], Piyanoot Tapaneeyaphan[1], Adelbert De Clercq[1], Tamara Jarayseh[1], Andy Willaert[1], Peter Byers[2], Paul Coucke[1], Bettina Blaumeiser[3], Delfien Syx[1], Fransiska Malfait[1] & Sofie Symoens[1]
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[3] University of Antwerp, Department of Medical Genetics, Antwerp, Belgium

Identification of a genetic defect in human STX18 and modelling in zebrafish unravel a key role of syntaxin-18 in chondrogenesis

15.00 - 15.15 Elsa Khoury[1], Hiba Maalouf[1], Antonella Mendola[2], Sylvain Choquet[3], Caroline Besson[4], Judith Landman-Parker[5], Vincenzo D'Angiolella[6], Hélène Antoine Poirel[7] & Nisha Limaye[1]
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[3] Service d'Hématologie, CHU La Pitié Salpétrière, France – French registry of familial lymphoid neoplasms
[4] Unité d'Hémato-Oncologie, Centre Hospitalier de Versailles, Le Chesnay, France – Centre for Research in Epidemiology and Population Health (CESP) INSERM Unit 1018, Villejuif, France
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[6] Department of Oncology, Medical Research Council Institute for Radiation Oncology, University of Oxford, Oxford, UK
[7] Belgian Cancer Registry, Brussels, Belgium

CCNF (Cyclin F) as a candidate gene for familial Hodgkin lymphoma

15.15 - 15.30 Maaike Alaerts[1], Aleksandra Nijak[1], Eline Simons[1], Bert Vandendriessche[1], Dieter Van de Sande[2], Erik Fransen[3], Ewa Sieliwonczyk[1], Dorien Schepers[1], Dirk Snyders[2], Emeline Van Craenenbroeck[4], Johan Saenen[4], Alain J Labro[5] & Bart L Loeys[1]
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Functional characterization of iPSC-derived cardiomyocytes from Brugada syndrome patients of a genetically unresolved family reveals distinct underlying mechanisms

➢ Parallel Session IV

Chairs: Kim van Berkel & Arvid Suls

14.15 - 14.30 Joke Mertens[1], Marius Regin[1], Neelke De Munck[2], Edouard Couvreu de Deckersberg[1], Christophe Blockeel[2], Herman Tournaye[2], Hilde Van de Velde[2], Karen Sermon[1] & Claudia Spits[1]
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Mitochondrial DNA mosaicism in early human development

14.30 - 14.45 Marie De Borre[1], Qian Yu[2], Mika Van Den Ackerveken[2], Lore Lannoo[3], Leen Vancoillie[4], Jeroen Breckpot[4], Koenraad Devriendt[4], Joris Vermeesch[4], Kristel Van Calsteren[3] & Bernard Thienpont[2]
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[4] Center for Human Genetics, University hospital Leuven, University of Leuven, Leuven, Belgium

Methylome Analysis of cfDNA to diagnose preeclampsia presymptomatically
14.45 - 15.00 Kris Van Den Bogaert[1], Lore Lannoo[2], Nathalie Brison[1], Machteld Baetens[3], Bettina Blaumeiser[4], François Boemer[5], Laura Bourlard[6], Vincent Bours[5], Anne De Leener[7], Marjan De Rademaeker[4], Julie Désir[8], Annelies Dheedene[3], Armelle Duquenne[7], Nathalie Fieremans[9], Annelies Fieuw[9], Jean-Stéphane Gatot[5], Katrien Janssens[10], Sandra Janssens[3], Damien Lederer[8], Axel Marichal[8], Björn Menten[3], Colombine Meunier[8], Leonora Palmeira[5], Bruno Pichon[6], Eva Sammels[9], Guillaume Smits[6], Yves Sznajer[7], Elise Vantroys[9], Koen Devriendt[1] & Joris Vermeesch[11]
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Outcome of publicly funded nationwide first-tier noninvasive prenatal screening
15.00 - 15.15 Ilse Van Gucht[1], Josephina A.N. Meester[1], Jotte Rodrigues Bento[1], Maaike Bastiaansen[1], Jarl Bastianen[1], Ilse Luyckx[1], Lotte Van Den Heuvel[1], Cédric H.G. Neutel[2], Pieter-Jan Guns[2], Mandy Vermont[2], Erik Fransen[1], Melanie H.A.M. Perik[1], Joe Davis Velchev[1], Maaike Alaerts[1], Dorien Schepers[1], Silke Peeters[1], Isabel Pintelon[3], Lut Van Laer[1], Bart L. Loeys[1] & Aline Verstraeten[1]

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[3] Laboratory of Cell Biology & Histology, Department of Veterinary Sciences, University of Antwerp, Antwerp, Belgium

The first human importin-β-related disorder: syndromic thoracic aortic aneurysm caused by bi-allelic loss-of-function variants in IPO8

15.15 - 15.30 Martina Marangoni[1], Guillaume Smits[1], Bettina Blaumeiser[2], Elise Brischoux-Boucher[3], Saskia Bulk[4], Thomy De Ravel[5], Guillaume Debray[4], Boyan Dimitrov[5], Sandra Janssens[6], Kathelijn Keymolen[5], Marie Laterre[4], Kim van Berkel[5], Lionel Van Maldergem[3], Isabelle Vandernoot[1], Catheline Vilain[7], Catherine Donner[8], Laura Tecco[9], Dominique Thomas[10], Julie Désir[11], Marc Abramowicz[12] & Isabelle Migeotte[13]
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Implementation of fetal clinical exome sequencing: comparing prospective and retrospective cohorts

15.30-16.00 COFFEE BREAK / MEET YOUR SPONSOR / POSTER VIEWING

16.00-17.30 PLENARY SESSION 2

Chairs: Joris Vermeesch & Martine De Rycke

- **Prof. Eva Hoffmann**

  The origin of aneuploidy and its relevance for reproduction

- **Prof Stéphane Viville**

  Meiosis genes: the difference in meiosis between male and female

17.30-18.00 CLOSING REMARKS / BEST POSTER/ PRESENTATION AWARDS

18.00 RECEPTION