



23rd Annual Meeting
March 17, 2023
Dôme, Charleroi

Programme

9h00-9h25: Registration

9h25-9h30: Welcome

9h30-11h00: **PLENARY SESSION 1 : Sex Determination**

- 9h30: Yann Guiguen : **Diversity and evolution of genetic sex-determination in fish.**
- 10h00: Jana Henck : **Single cell, whole embryo phenotyping of pleiotropic disorders of mammalian development.**
- 10h30: Kristof Van Assche : **Early surgical interventions on intersex children: Legal considerations and the human rights debate.**

11h00-11h30: Coffee break/Meet the sponsors/Poster viewing

11h30 – 12h45: Selected oral presentations

Parallel session 1 :

11h30: David Schröder : **Diamond AstraZeneca: Use of PARP inhibitor in early breast cancer as adjuvant therapy: new indication for BRCA1 and BRCA2 testing.**

11h45: Michiel Vanneste : **Multivariate GWAS on achondroplasia-like craniofacial shape variation in healthy human individuals.**

12h00: Michiel Vanhooydonck : **The establishment of the first reported zebrafish model for thoracic aortic dissection and rupture.**

12h15: Machteld Baetens : **Preimplantation genetic testing (PGT) in Belgium: national recommendations for genetic centers.**

12h30: Kristine Hovhannesyanyan : **BabyDetect project: molecular newborn screening in Belgium.**

Parallel session 2 :

11h30: Lisa Dangreau : ***The pseudoxanthoma elasticum zebrafish model contributes to novel pathophysiological insights and therapeutic strategies in ectopic mineralization.***

11h45: Bieke Bekaert : ***Retained chromosomal integrity following CRISPR/Cas9-based mutational correction in human embryos.***

12h00: Lucia Bucciolli : ***Striking phenotypical differences between Ipo8 knock-out mouse models on different genetic backgrounds explored by RNA sequencing.***

12h15: 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.***

12h30: Hamide Yildirim : ***Malfunctional CTRP1 impairs the cell cycle implying its crucial role in brain development.***

12h45-14h00: Lunch break and poster viewing

14h00-15h15: Selected oral presentations

Parallel session 3 :

14h00: Senne Meynants : ***Multiple recombination mechanisms drive the high incidence of 22q11.2 Deletion Syndrome.***

14h15: Dale Annear : ***CGG Short Tandem Repeat Expansions are Overrepresented in Neurodevelopmental Disorders.***

14h30: Marlies Colman : ***Kyphoscoliotic Ehlers-Danlos syndrome due to pathogenic variants in PLOD1 and FKBP14: further insights into the pathophysiology and comparison of clinical characteristics.***

14h45: Margot van Riel : ***Single-cell dissection of cervix and placenta reveal both novel and overlapping cell types.***

15h00: Liene Thys : ***To test or not to test: the importance of genetics in the diagnostic workup of cerebral palsy.***

Parallel session 4 :

14h00: Richard Capper : ***Diamond Illumina: Advancements in Whole-Genome Sequencing – Achieving a complete and comprehensive genome at scale.***

14h15: Miriam Bauwens : ***Whole genome sequencing sheds light on the dark matter of the genome in patients with inherited retinal diseases.***

14h30: Laurens Hannes : **Differential alternative splicing analysis links variation in ZRSR2 to a novel oral-facial-digital syndrome.**

14h45: Eline Van Vooren : **An in vitro enzymatic assay to elucidate the VUS problem in RPE65, a target for retinal gene therapy.**

15h00: Benjamin Huremagic : **Sample Catalog: A Federated Platform for Identification and sharing of biological samples in rare disorders.**

15h15 – 15h45: Coffee break/Meet the sponsors/Poster viewing

15h45 – 16h15: General assembly

16h15 – 17h30: PLENARY SESSION 2 : Methylation

- 16h15: Deborah Mackay : **It takes two: genetics and epigenetics of imprinting disorders.**
- 16h45 : Ulrich Zechner : **Derepression of imprinted gene alleles as a new approach to treat imprinting disorders.**
- 17h15: Closing remarks and best poster / oral presentation awards

17h30 – 23h: Reception and party