

CONFERENCE PROGRAMME

Saturday, 3 July 2021

(Central European Summer Time, UTC +2)

CEST **Permanent Working Groups, Satellite Meetings and Online Poster Area**

09:00-09:05 **Introduction by the President**
Mariano Rocchi

09:05-10:00 **Permanent Working Group Animal & Plant Genomics**
Session Coordinator: Pat Heslop-Harrison

Poster Presentations:

Alla Krasikova: Spatial organization of topologically associated chromatin domains in chicken somatic cell types and meiotic lampbrush chromosomes revealed by FISH

Alsu Saifitdinova: Avian lampbrush chromosomes reflect the complexity of their genome organization at meiotic reprogramming stage

Tatiana Bikchurina: Hybrid sterility in gray voles

Alessandra Iannuzzi: Bovine telomere association between age and breed

Alexandr Sember: Cytogenomics of annual killifish: sex determination and repetitive DNA dynamics in closely related *Nothobranchius furzeri* and *N. kadleci*

Sebastian Pita: Satellitome analysis unravels the karyotypic evolution in *Triatoma* kissing bugs (Hemiptera: Reduviidae)

Pablo Mora: Comparative analysis of the satellitome in ladybirds (Coleoptera, Coccinellidae)

10:05-10:35 **Permanent Working Group Cytogenetic Toxicology and Mutagenesis**
Session Coordinator: José M. Garcia-Sagredo

Poster Presentations:

Sophia Zachaki: Chromosome instability and gene rearrangements in ectopic endometriotic tissue of women with endometriosis

Moonisah Usman: Predictors of genomic instability in childhood obesity: a study of multiple variables

Roberto Valli: Interstitial deletion of chromosome 20 and isochromosome i(7)(q10) in the bone marrow of patients with Shwachman-Diamond syndrome: novel evidence of karyotype instability related to somatic rescue mechanisms

10:40-11:35 **Industry-sponsored Satellite Symposium**

11:40-12:15 **Permanent Working Group Prenatal Diagnosis**
Session Coordinator: Jean-Michel Dupont

Tommaso Cavazza: Parental genome unification is highly error-prone in mammalian embryos

Poster Presentation:

Laurence Lohmann: Whole genome NIPT: confirmation of the high performance as a screening test

12:20-12:50 **Permanent Working Group Quality Issues, Training and Cytogenomics**
Session Coordinator: Ron Hochstenbach

Rosalind Hastings: ISCN 2020 - What's new?

Poster Presentation:

Thomas Liehr: An international collaboration for human ring chromosomes (ICHRC) towards developing standards and guidelines for best practice in laboratory diagnosis, clinical management and collaborative research

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- 12:55-13:50 **Industry-sponsored Satellite Symposium**
- 13:55-14:25 **Permanent Working Group Cytogenomics of Hematological Malignancy**
Session Coordinator: Harald Rieder
- Poster Presentations:
Dolors Costa: Balanced and unbalanced translocations in a multicentric series of 2,843 patients with chronic lymphocytic leukemia
- Catherine Menten: The use of optical genome mapping (Bionano) to understand a complex mechanism of KMT2A rearrangement in an AML case
- 14:30-15:25 **Permanent Working Group Cytogenomics**
Session Coordinator: Joris Vermeesch
- Poster Presentations:
Flavia Maggolini: Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution
- Veniamin Fishman: Precise detection of balanced chromosomal rearrangements and single nucleotide variations using chromosome conformation capture
- Tine De Coster: Non-canonical zygotical divisions are characterized by parental genome segregation errors
- Thomas Guignard: AnnotSV and knotAnnotSV: a web server for human structural variations annotations, ranking and analysis
- Jakob Schuy: A case of ring chromosome 21 with hemifacial microsomia, developmental and speech delay analysed with WGS and patient-derived iPSCs
- 15:30-16:25 **Industry-sponsored Satellite Symposium**
- 16:30-17:00 **Permanent Working Group Cancer Cytogenomics and Solid Tumor Studies**
Session Coordinator: David Gisselsson Nord
- Paola dal Cin: Next-Generation technologies in old generation laboratories
- 17:05-18:00 **Opening Ceremony**
Chair: Mariano Rocchi
President's Address
Mariano Rocchi
- Opening Lecture
Terry Hassold: Aneuploidy: Where do we go from here?

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Sunday, 4 July 2021

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CEST ONLINE POSTER AREA

10:00-12:30 **Session 1 : Prenatal and Preimplantation Diagnosis**

Chair: Jean-Michel Dupont

10:00-10:35 Lyn Chitty: The impact of sequencing on prenatal diagnosis

10:35-11:10 Erik Sistermans: Non-invasive fetal aneuploidy screening

11:10-11:45 Carmen Rubio: Embryo aneuploidy testing, past, present and future

11:45-12:30 Selected Abstracts

Stanislav Vasilyev: Aneuploidy is associated with increased methylation of different LINE 1 retrotransposon families in placenta of first trimester miscarriages

Kris Van Den Bogaert: Outcome of publicly funded nationwide first tier noninvasive prenatal screening

Radhia M'Kacher: Direct inheritance of telomere shortening and aberrations detected during prenatal period

12:30-14:00 **ONLINE EXHIBITION & BREAK**

14:00-16:15 **Session 2 : Cancer Cytogenomics**

Chair: Roberta Vanni

14:00-14:35 Roland Schwarz: Pervasive chromosomal instability and karyotype order in tumour evolution

14:35-15:10 Florent Mouliere: Early detection of cancer by liquid biopsy

15:10-15:45 Lindsey Montefiori: Enhancer hijacking of BCL11B defines a subtype of lineage ambiguous acute leukemia

15:45-16:15 Selected Abstracts

Ming Chen: primary tumor and circulating tumor cells captured by microfluidics in epithelial ovarian cancer tumor heterogeneity or allele dropout?

Barbara Dewaele: Opportunities of Optical Genome Mapping for Genetic Diagnosis in Acute Lymphoblastic Leukemia

16:15-16:30 **ONLINE EXHIBITION & BREAK**

16:30-18:45 **Session 3 : Clinical Cytogenomics**

Chair: Elisabeth Syk-Lundberg

16:30-17:05 Chelsea Lowther: Computational and molecular dissection of long-range positional effects in human developmental disorders

17:05-17:40 Gulio Genovese: Mosaic variants and clonal selection in health and disease

17:40-18:15 Sébastien Jaquemont: Predicting CNV effects on brain structure, function and risk for psychiatric conditions

18:15-18:45 Selected Abstracts

Heleen Masset: Chimerism and mixoploidy in human preimplantation embryos disclosed by comprehensive PGT

Romain Nicolle: 16p13.11p11.2 triplication syndrome a new recognizable genomic disorder characterized by optical mapping

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Monday, 5 July 2021

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CEST	ONLINE POSTER AREA
10:00-12:15	Session 4 : Animal and Plant Cytogenomics <i>Chair: Pat Heslop-Harrison</i>
10:00-10:35	Tony Heitkam: Repetitive DNA, epigenetic variability, and genomic innovation in higher plants
10:35-11:10	Marta Farré-Belmonte: Genome evolution and genetic changes associated with individual and species differences
11:10-11:45	Aurora Ruiz-Herrera: Genome integrity and mechanisms driving karyotype evolution
11:45-12:15	Selected Abstracts Lyubov P. Malinovskaya: The meiotic behavior and transmission of germline restricted chromosome (GRC) in songbirds Cibele Sotero-Caio: Trends of chromosome rearrangements and evolution in bats highlights from comparative chromosome painting analyses
12:15 - 13:45	ONLINE EXHIBITION & BREAK
13:45-15:30	Session 5 : Widening Perspectives on Genome Variation <i>Chair: Joris Vermeesch</i>
13:45-14:20	Tom Misteli: Genome Organization: from basics to disease
14:20-14:55	Thierry Voet: Single-cell multi-omics to study genetic heterogeneity in health and disease
14:55-15:30	Segun Fatumo: The global genome/genomes sans frontiers
15:30-16:00	ONLINE EXHIBITION & BREAK
16:00-17:45	Session 6 : Disease Prevalence and Sex Differences <i>Chair: Kamlesh Madan</i>
16:00-16:35	Joris Veltman: (Cyto)genetics of male infertility
16:35-17:10	Darío Lupiáñez: Non-coding regulation of sexual features
17:10-17:45	Christine M Disteche: X-chromosome regulation and sex differences in brain anatomy
17:50-18:25	Farewell Session <i>Chair: Mariano Rocchi</i> Closing Keynote Evan E. Eichler: Long-read sequencing, disease variants and genome evolution
18:25-19:00	Closing Ceremony <i>Chair: Mariano Rocchi</i>