

The 11th European Cytogenetics Conference

Florence, 1-4 July 2017

Permanent Working Groups Scientific Programme

SATURDAY, 1 July 2017

14:00-16:00 ANIMAL, PLANT AND COMPARATIVE CYTOGENETICS

Coordinators: J.S. (Pat) HESLOP-HARRISON, Leopoldo IANNUZZI, Mariano ROCCHI

Affari Room 1

- 14:00-14:05 Introduction Pat Heslop-Harrison, Marino Rocchi and others
- 14:05-14:13 **Alessio Iannucci:** Here be dragons: chromosomes flow sorting of *Varanus komodoensis* for sequencing and genome anchoring
- 14:13-14:21 **Ming Chen:** Cytogenetic characterization, genome size estimation and restriction pattern analysis of two *Kurixalus* tree frogs (Anura: Rhacophoridae) with diverse reproductive behavior
- 14:21-14:29 **Artem Lisachov:** The toad-headed agamas (*Phrynocephalus*) are champions in crossing over rate
- 14:29-14:37 **Andreas Houben:** Structure and regulation of holocentric chromosomes
- 14:37-14:45 **Claudia Rita Catacchio:** Chromosome structural variation in grape genome: unlocking the genetic potential of table grape varieties towards the design of new water conservation programs in sustainable viticulture
- 14:45-14:53 **Paulina Tomaszewska:** Discrimination of the C genome and two closely related forms of the A genome in endosperm of an *Avena magna* x *A. longiglumis* amphiploid
- 14:53-15:01 **Anna Zlotina:** Lampbrush chromosome microdissection as an approach to comprehensive investigation of heterochromatic chromosome regions
- 15:01-15:09 **Vânia Techio:** Construction of physical maps of chromosomes in *Brachiaria* spp.: beginning a journey
- 15:09-15:17 **Katarzyna Kowalska:** Evaluation of chromosome micro-rearrangements of a horse applying array CGH
- 15:17-15:25 **Maria A. Biscotti:** The duplication events have traced the evolution of vitellogenin gene family
- 15:25-15:33 **Martina Johnson Pokorná:** The evolution of sex chromosomes in reptiles
- 15:33-15:41 **Halina Cernohorska:** Cytogenetic studies of the genus *Raphicerus* (Bovidae)
- 15:41-15:49 **Rebecca Jennings:** A Cross-Species Bioinformatics and FISH approach to physical mapping of Mammalian Genomes
- 15:49-15:57 **Darren Griffin:** A novel approach for the physical mapping of avian genome assemblies to a chromosomal level

14:00-15:00 CANCER CYTOGENETICS, SOLID TUMOR STUDIES

Coordinators: Roberta VANNI, David GISSELSSON NORD

Affari Room 2

- 14:00-14:05 **Roberta Vanni:** Welcome and commemoration
- 14:05-14:15 **David Gisselsson Nord:** Introduction to cancer cell evolution
- 15:15-15:30 **Sabrina Tosi:** HLXB9 expression is independent from its nuclear gene positioning in breast cancer cells
- 15:30-15:45 **Ruby Banerjee:** Characterisation of chromosomal aberrations in mouse pancreatic cancer using multiplex-FISH (M-FISH)
- 15:45-16:00 **Tiziana Storlazzi:** Myc-containing double minute chromosomes: origin, structure and impact upon transcriptome in acute myeloid leukemia

14:00-15:00 CLINICAL AND MOLECULAR APPROACHES TO CYTOGENETIC SYNDROMES

Coordinators: Conny van RAVENSWAALJ, Cristina SKRYPNYK, Nicole de LEEUW

Affari Room 3

- 14:00-14:05 **Nicole de Leeuw :** Welcome
- 14:05-14:20 **Niels Tommerup:** NGS mapped breakpoints in balanced chromosomal Copenhagen (DK) rearrangements including the first large cohort of healthy carriers
- 14:20-14:35 **Mana M. Mehrjouy:** Enhancer swapping dual loss and gains of enhancers by Copenhagen (DK) balanced translocations and inversions between highly conserved regulatory domains
- 14:35-14:50 **Beike Leegte:** The power of social media for karyotype-phenotype analysis Groningen (NL) in rare chromosome disorders
- 14:50-15:00 **Nicole de Leeuw:** Summary and closing comments

15:00-16:00 CYTOGENETIC TOXICOLOGY AND MUTAGENESIS

Coordinators: Jose M. GARCIA-SAGREDO, Lucia MIGLIORE

Affari Room 2

- 15:00-15:10 **Jose M. Garcia-Sagredo, Lucia Migliore:** Welcome
- 15:10-15:20 **Tamara Borgonovo:** Genomic integrity of stem cells
- 15:20-15:30 **Vivi Miriam Srivastava:** Clonal chromosomal abnormalities in ataxia telangiectasia
- 15:30-15:40 **Vedran Mužinić:** Effect of α -cypermethrin on chromosome mis-segregation and aneuploidy induction in human peripheral blood lymphocytes as measured by FISH-coupled micronucleus assay

- 15:40-15:50 **Petra Musilova:** Chromosomal aberrations in lymphocytes of patients with newly diagnosed solid tumors and patients undergoing long-term chemotherapy detected by FISH
- 15:50-16:00 PWG Coordinators Conclusions and future directions

15:00-16:00 MARKER CHROMOSOMES

Coordinators: Isabel Marques CARREIRA, Joana Barbosa MELO

Affari Room 3

- 15:00-15:10 **Isabel M. Carreira:** Introduction – Marker Chromosomes Permanent Working Group
- 15:10-15:20 **Anette K. Eek:** Short Communications on markers, Karyotyping and arrayCGH do we still need both
- 15:20-15:30 **Carreira, Mascarenhas, Pinto, Pires, Paiva, Jardim, Melo:** The mysterious case of the ghost marker chromosome
- 15:30-15:50 **Niels Tommerup:** NGS and detection/characterization of Marker Chromosomes
- 15:50-16:00 **Joana B. Melo:** Supernumerary marker chromosomes webpage – Importance and New projects

15:00-16:00 Prenatal Diagnosis

Coordinators: Maria Do Rosário CARVALHO PINTO LEITE, Seher BARASAN

Adua Room 4

- 15:00-15:10 **Cindy Melotte:** Transfer of aneuploid embryos following preimplantation genetic diagnosis the added value of a haplotyping based genome wide approach
- 15:15-15:25 **Paolo Guanciali Franchi:** Sequential combined test, second trimester maternal serum markers, and circulating fetal cells to select women for invasive prenatal diagnosis
- 15:30-15:40 **Peter Benn:** Clinical Experience with a Single-Nucleotide Polymorphism-Based Non-Invasive Prenatal Test for Five Clinically Significant Microdeletions
- 15:45-15:55 **Ming Chen:** Prenatal diagnosis of fetal aneuploidy and de novo copy number variation by isolating extravillous trophoblasts and nucleated fetal red blood cells from maternal circulation: a proof-of-principle study using the automated Cell Reveal™ platform based on nanostructure and microfluidics

16:00-17:00 QUALITY ISSUES AND TRAINING IN CYTOGENETICS

Coordinators: Ros HASTINGS, Martine DOCO-FENZY, Marta RODRIGUEZ DE ALBA

Affari Room 1

- 16:00-16:20 **Ros Hastings:** Use of ISCN in reporting Cytogenomic results – common errors and how to prevent them
- 16:20-16:45 **Martine Doco-Fenzy:** Changes in ISCN 2016 + ISCN quiz
- 16:45-17:00 Update on the cytogenomic guidelines under review and those international guidelines currently available (TBC).

16:00-17:00 CYTOGENETICS OF HAEMATOLOGICAL MALIGNANCIES

Coordinators: Bertil JOHANSSON, Harald RIEDER

Affari Room 2

- 16:00-16:05 **Harald Rieder:** Welcome
- 16:05-16:30 **Harald Rieder:** Report of the meeting of the workpackage “Cytogenetics” of the European Leukemia Network, Feb. 2017, Mannheim, Germany
- 16:30-17:00 **TBA**

16:00-17:00 MOLECULAR CYTOGENETICS AND ARRAY CGH

Coordinators: Joris VERMEESCH, Abhijit KULKARNI

Affari Room 3

- 16:00-16:20 **Katrina Rack:** Prenatal CNV classification and reporting survey from 60 European centres
- 16:20-16:40 **Nicole de Leeuw:** Clinical added value of SNP arrays over CGH arrays
- 16:40-17:00 **Björn Menten:** Detection of copy number variation using shallow whole genome sequencing (CNVseq) as a valuable alternative to genomic microarrays