The 11th European Cytogenetics Conference  
Florence, 1-4 July 2017  
Scientific Programme

SATURDAY, 1 July 2017

14:00-17:00  Permanent Working Groups
17:00-17:50  Satellite Symposium
18:00-19:00  **Opening lecture.** Chairs: Elisabeth Syk Lundberg - Lidia Larizza  
Dennis Lo: From Chromosomes to circulating DNA

SUNDAY, 2 July 2017

08:30-10:25  **Plenary session 1 - Chromosomal Imbalances and Rearrangements**  
Chairs: Orsetta Zuffardi - Damien Sanlaville
08:30-09:05  **Michael Talkowski:** Chromosomal rearrangements and sequencing
09:05-09:40  **Wigard Kloosterman:** Genomic and functional overlap between somatic and germline chromosomal rearrangements
09:40-10:25  Selected abstracts
  **Caroline Schluth-Bolard:** Unravelling structural chromosomal rearrangements by whole genome sequencing: results of the ANI project
  **Ivan Iourov:** Chromothripsis as a mechanism driving genomic instability mediating brain diseases
  **Anna Lindstrand:** Whole genome characterization of array defined clustered CNVs reveals two distinct complex rearrangement subclasses generated through either non-homologous repair or template switching

11:00-12:00  **Plenary session 2: 100 Years ago: T. C. Hsu and Mammalian Cytogenetics**  
Chairs: Mariano Rocchi - Vladimir Trivonof
11:00-11:30  **Malcolm Ferguson-Smith:** Mammalian cytogenetics on the centenary of T.C. Hsu
11:30-12:00  **Pat Heslop-Harrison:** Comparative cytogenomics
12:00-14:40  Poster session and Satellite Symposia
14:40 - 15:45  Concurrent Sessions

  **Concurrent Session 1 - Genome Plasticity**  
  Chairs: Evan Eichler – Mariano Rocchi
14:40-15:05  **Mario Ventura:** Evolutionary instability and disease susceptibility of the chromosome 8p23.1 region
15:05-15:30  **Scott E. Devine:** L1 and cancer
15:30-15:45  Selected Abstract
Sandra Louzada: Defining haplotypes of complex structural variation using multicolour fibre-FISH: amylase CNV study

Concurrent Session 2 - Clinical Cytogenetics
Chairs: Albert Schinzel - Sabrina Giglio

14:40-15:05 Orsetta Zuffardi: A fil rouge links numerical to structural chromosome abnormalities via chromothripsis

15:05-15:30 Guy Froyen: Intellectual disability in female patients with a skewed X-inactivation pattern

15:30-15:45 Selected Abstract
Mariana Moyses-Oliveira: Balanced X-autosome translocation suggests association of AMMCR1 disruption with hearing loss and growth, bone and heart alterations

Coffee break

Concurrent Sessions

16:15-17:20 Concurrent Session 3 - CRISPR-Cas9
Chairs: Nicole de Leeuw - Juan Cigudosa

16:15-16:40 Giovanni Perini: The CRISPR-Cas9 world

16:40-17:05 Nissim Benvenisty: Mapping the human genome using human haploid embryonic stem cells

17:05-17:20 Selected Abstract
Isabell Pechtl: Monitoring guide RNA synthesis for CRISPR-Cas9 genome editing workflow

Concurrent Session 4 - Chromatin Organization and Dynamics
Chairs: Lidia Larizza - Thomas Cremer

16:15-16:40 Stefan Mundlos: Chromatin domains and diseases

16:40-17:05 Maria Pia Cosma: The nanoscale structure of chromatin fibers correlates with cellular state

17:05-17:20 Selected Abstract
Mireia Solé: Chromosome territories in mice spermatogenesis: a new three-dimensional methodology

17:20-18:30 Poster session

20:00 Conference Dinner 20 Years of E.C.A.

MONDAY, 3 July 2017

08:30-10:30 Plenary session 3 - Tumor Cytogenetics I
Chairs: Felix Mitelman - Claudia Haferlach

08:30-09:00 Peter Lichter: Mechanistic aspects of chromothripsis

09:00-09:30 Francesco Lo Coco: Chromosome translocations in cancer: from genetics to therapy – APL as a paradigm

09:30-10:30 Selected Abstracts
David Gisselsson: A geography of clones: mapping the tumour genome over anatomic space in children with cancer

Emanuela Maserati: Novel recurrent chromosome anomalies in Shwachman-Diamond syndrome

Nicole Chia: Deletion 13q characterised by SNP microarray profiling of a large cohort of CLL patients

Gemma Macchia: The transcriptome plasticity of genome amplification in cancer

coffee break

11:00-12:15 Concurrent Sessions

**Concurrent Session 5 - Tumor Cytogenetics II**

Chairs: Roberta Vanni - Thierry Lavabre Bertrand

11:00-11:30 Ellen Heitzer: Circulating tumor DNA as a liquid biopsy for cancer

11:30-12:00 Vassiliou George: Ageing and leukemogenesis

12:00-12:15 Selected Abstract

**Karin Nebral**: Prospective diagnostic evaluation of genetic abnormalities in childhood acute lymphoblastic leukemia with hub gene FISH screening and array analyses

**Concurrent Session 6 - Animal and Plant Cytogenetics**

Chairs: Pat Heslop-Harrison - Roscoe Stanyon

11:00-11:30 **Vladimir Trivonof**: Whole genome duplications in vertebrate evolution

11:30-12:00 **Martin Lysak**: Reconstruction of ancestral karyotypes and identification of chromosome evolution

12:00-12:15 Selected Abstract

**Giorgia Chiatante**: Apparently identical heterozygous neocentromeres in two closely related Cercopithecini species

12:15-14:30 Poster session and Satellite Symposia

14:30-15:45 **Plenary session 4 - Meiosis and Non-Disjunction**

Chairs: Harald Rieder - Trude Schwarzacher

14:30-15:00 **Agata Zielinska**: Chromosome segregation defects in human oocytes

15:00-15:30 **Eva Hoffmann**: Meiosis and chromosome segregation

15:30-15:45 Selected Abstract

**Joy Delhanty**: Meiotic outcome in two carriers of Y autosome reciprocal translocations: selective elimination of certain segregants

coffee break

16:15-17:15 Concurrent Sessions

**Concurrent Session 7 - Epigenetics**

Chairs: Joris Vermesch - Juan C. Cigudosa

16:15-16:45 **Mafalda Barbosa**: Epimutations as a novel cause of congenital disorders

16:45-17:15 **Yuri Dubrova**: Transgenerational epigenetics effects
17:15-17:30 Selected Abstract

Neus Baena: Deletion encompassing DLK1 gene at 14q32 imprinted region in two new Temple syndrome cases

Concurrent Session 8 - Accreditation and Quality Control

Chairs: Konstantin Miller - Jose Garcia Sagredo

16:15-16:45 Harald Rieder: Ten years external quality assessment in leukemia cytogenetics in Germany – what did we learn?

16:45-17:15 Martine Doco-Fenzy: External quality assessment in constitutionnal array-CGH and consequences for routine practice, the French experience

17:15-17:30 Selected Abstract

Ros Hastings: External Quality Assessment of clinical genetics: experiences with the pilot assessments

17:30-18:30 Poster session

18:30 E.C.A. General Assembly

Tuesday, 4 July 2017

09:00-10:30 Plenary session 5 - Prenatal Diagnosis

Chairs: The-Hung Bui - Jean-Michel Dupont

09:00-09:25 Mark I Evans: Noninvasive prenatal screening or advanced diagnostic testing: caveat emptor

09:25-09:50 Vincenzo Cirigliano: Performance evaluation and first clinical application of a new paired-end sequencing approach for cfDNA based aneuploidy screening

09:50-10:15 Raoul Orvieto: Should preimplantation genetic screening (PGS) be implemented to routine IVF practice?

10:15-10:30 Selected Abstract

Kris Van Den Bogaert: The majority of uncommon chromosomal imbalances detected by NIPT are postzygotic (feto)placental mosaics

10:30-10.50 Coffee break

10:50-11.40 Satellite Symposia

11:50-12:20 Keynote lecture. Chairs: Mariano Rocchi - Elisabeth Syk-Lundberg

Evan E. Eichler: Duplications and Evolution

12:20 Closing ceremony. Elisabeth Syk Lundberg - Orsetta Zuffardi