

12th EUROPEAN CYTOGENOMICS CONFERENCE

6 – 9 JULY 2019 • SALZBURG CONGRESS • SALZBURG, AUSTRIA

SCIENTIFIC PROGRAMME

SATURDAY, 6 July 2019

14:00-17:00 **Permanent Working Groups Programme**

Mozart 2-3 Hall at Ground Floor

14:00-15:00 PWG Cytogenomics

15:00-16:00 PWG Clinical and Molecular Approaches to Cytogenetic Syndromes

16:00-17:00 PWG Prenatal Diagnosis

16.00-16.05 Welcome and Introduction

16.05-16.20 CVS :European practices

Rosário PINTO LEITE

16.20-16.35 "Back for the future" - Lessons from the past for an updated management of the trophoblast

Aurélie COUSSEMENT

16.35-16.50 "Risk estimation of uniparental disomy of chromosome 14 or 15 in a fetus with a parent carrying a non-homologous Robertsonian translocation"

Kamran MORADKHANI

16.50-17.00 Discussion

Mozart 4+5 Hall at Ground Floor

14:00-15:00 PWG Marker Chromosomes

15:00-16:00 PWG Cytogenetics of Haematological Malignancies

16:00-17:00 PWG Cancer Cytogenetics, Solid Tumor Studies

Trakl Hall at 3rd Floor:

14:00-15:00 PWG Cytogenetic Toxicology and Mutagenesis

15:00-17:00 PWG Animal, Plant and Comparative Cytogenetics

Doppler Hall at 4th Floor:

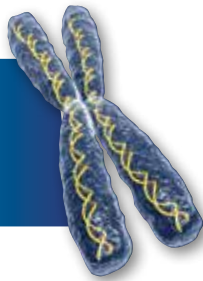
16:00-17:00 PWG Quality Issues And Training In Cytogenetics

17:00-17:50 **Symposium:** Molecular Cytogenetics (BMC-Springer/Nature)
symposium dedicated to the memory of
Prof. Yuri Yurov

Europa Hall

18:00-19:00 **Opening Lecture**
Chairs: Mariano Rocchi - Dieter Kotzot
Joris Vermeesch: Somatic chromosomal mosaicism

Europa Hall

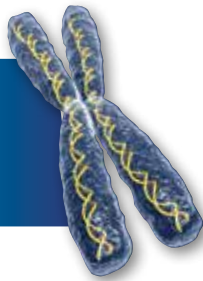


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SUNDAY, 7 July 2019

08:30-10:15	Plenary Session 1 - Recent advances in cytogenomics Chairs: Mariano Rocchi - Thierry Lavabre-Bertrand	<i>Europa Hall</i>
08:30-09:00	Claudia Haferlach: The future of cytogenomics in the diagnostics	
09:00-09:30	Michael Speicher: Liquid biopsies in patients with cancer	
09:30-10:15	Selected Abstracts	<i>Europa Hall</i>
09:30-09:45	Pascal Chambon: A simple, universal and cost-efficient dPCR method for the targeted analysis of copy number variations	
09:45-10:00	Laïla El Khattabi: Next Generation Mapping, a novel approach that enables the detection of unbalanced as well as balanced structural variants	
10:00-10:15	Paolo Reho: Low-coverage whole genome sequencing in plasma circulating cell-free DNA analysis: the Turner syndrome experience	
10:15-10:45	Coffee break	
10:45-11:45	Plenary Session 2 - 50 Years of chromosome banding Chairs: Kamlesh Madan - José Garcia-Sagredo	<i>Europa Hall</i>
10:45-11:15	Felix Mitelman: Chromosome banding: the end of the Dark Ages	
11:15-11:45	Darío G. Lupiáñez: Structural variation in the 3D genomic era: Implications for disease and evolution	
12:00-14:30	Poster Session	<i>Exhibition Area</i>
12:30-13:30	Agilent Satellite Symposia Advantages of CGH+SNP Microarrays in Clinical Cytogenetics Do not let a technology define your needs, choose a technology that fits your needs Aurélie Mouka & Aneta Mikulasova	<i>Europa Hall</i>
14:30-15:45	Concurrent Sessions	
	Concurrent Session 1 - 3D chromatin organization and dynamics Chairs: Jean-Michel Dupont - Darío G. Lupiáñez	<i>Europa Hall</i>
14:40-15:05	Alexandre Reymond: Genome architecture and diseases: the 16p11.2 paradigm	
15:05-15:30	Mario Nicodemi: Polymer physics predicts the impact on chromatin 3D structure of disease associated structural variants	
15:30-15:45	Selected Abstract Mireia Solé: Chromosome radial positioning in spermatogenic germ cells from <i>Mus musculus</i>	<i>Europa Hall</i>



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Concurrent Session 2 - Clinical cytogenomics

Karajan Hall

Chairs: Damien Sanlaville - Orsetta Zuffardi

14:40-15:05 **Nicole de Leeuw:** CNV and diseases: an overview in constitutional diagnostics

15:05-15:30 **Malte Spielmann:** The effect of structural variation in the three-dimensional genome

15:30-15:45 **Selected Abstract**

Karajan Hall

Jesper Eisfeldt: From cytogenetics to cytogenomics, whole genome sequencing as a comprehensive genetic test in rare disease diagnostics

15:45-16:15 **Coffee break**

16:15-17:20 **Concurrent Sessions**

Concurrent Session 3 - Structural organization of the human genome

Europa Hall

Chairs: Joris Vermeesch - Nicole de Leeuw

16:15-16:40 **Megan Y. Dennis:** The role of duplicated genes in human brain evolution and disease

16:40-17:05 **Francesca Antonacci:** Inversion variants in the human genome

17:05-17:20 **Selected Abstract**

Europa Hall

Anna Lindstrand: Cytogenetically visible inversions are formed by multiple molecular mechanisms

Concurrent Session 4 - Human infertility

Karajan Hall

Chairs: Elisabeth Syk Lundberg - Sevilhan Artan

16:15-16:40 **Pierre Ray:** Male infertility in humans, interest of whole exome sequencing

16:40-17:05 **Terry Hassold:** Aneuploidy in humans: new insights into an age-old problem

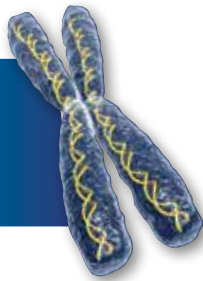
17:05-17:20 **Selected Abstract**

Karajan Hall

Harita Ghevaria: Next generation sequencing detects premeiotic errors in human oocytes and provides evidence of genetic influence

17:20-18:30 **Poster Session**

Exhibition Area

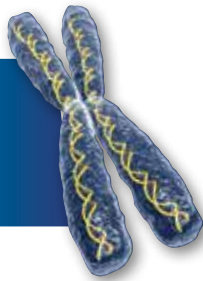


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MONDAY, 8 July 2019

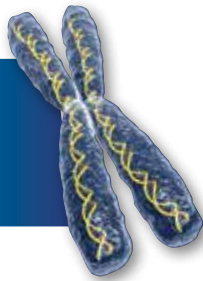
08:30-10:30	Plenary session 3 - Tumor Cytogenomics I Chairs: Felix Mitelman – Roberta Vanni	<i>Europa Hall</i>
08:30-09:00	Fredrik Mertens: Clonal evolution among different sarcoma subtypes	
09:00-09:30	David Gisselsson: Treatment resilience of cancer through clonal evolution	
09:30-10:30	Selected Abstracts	<i>Europa Hall</i>
09:30-09:45	Roberto Valli: Shwachman-Diamond Syndrome, expression arrays, clonal chromosome anomalies	
09:45-10:00	Paola Caria: Three-Dimensional Telomere Organization in papillary thyroid cancers	
10:00-10:15	Jordi Camps: Patterns of acquired uniparental disomy reveal biallelic inactivation of tumor suppressor genes in gastrointestinal cancers and in colorectal advanced adenomas	
10:15-10:30	Sabrina Haslinger: ZNF384 gene fusions in B-ALL: A report of fifteen Austrian cases secured by systematic FISH and array screening	
10:30-11:00	Coffee break	
11:00-12:15	Concurrent Sessions	
	Concurrent Session 5 - Tumor cytogenomics II Chairs: Claudia Haferlach - Harald Rieder	<i>Europa Hall</i>
11:00-11:30	Liran Shlush: Clonal evolution and risk factors - from age-related clonal hematopoiesis to AML	
11:30-12:00	Floris Foijer: Single cell DNA sequencing to quantify karyotype heterogeneity in cancer	
12:00-12:15	Selected Abstract Karla Svobodova: Identification of cryptic aberrations allows more accurate prognostic classification of patients with myelodysplastic syndromes and clonal evolution	<i>Europa Hall</i>
	Concurrent Session 6 - Animal and plant cytogenomics I Chairs: Pat Heslop-Harrison - Valerie Fillon	<i>Karajan Hall</i>
11:00-11:30	Alain Pinton: Chromosome rearrangements and meiosis in pig	
11:30-12:00	Ilya Kirov: Plant repeatome: cytogenetic, transcriptomic and proteomic aspects	
12:00-12:15	Selected Abstract Alessandra Iannuzzi: Cytogenetic and genomic assays in river buffalo (<i>Bubalus bubalis</i> , 2n=50) cows raised in urban and rural areas	<i>Karajan Hall</i>



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12:15-14:30	Poster Session	<i>Exhibition Area</i>
14:30-15:45	Plenary Session 4 - Chromosomal Imbalances Chairs: Mariano Rocchi - Konstantin Miller	<i>Europa Hall</i>
14:30-14:55	Orsetta Zuffardi: The trisomy legacy: from numerical to structural abnormalities	
15:00-15:30	Iben Bache: Long-term outcomes of prenatally detected de novo balanced chromosomal rearrangements	
15:30-15:45	Selected Abstract Aafke Engwerda: Phenotype-genotype analysis in a large cohort of 85 individuals with a terminal 6q deletion	<i>Europa Hall</i>
15:45-16:15	Coffee break	
16:15-17:30	Concurrent Sessions	
	Concurrent Session 7 - Animal and plant cytogenomics II Chairs: Trude Schwarzacher - Leopoldo Iannuzzi	<i>Europa Hall</i>
16:15-16:45	Raquel Chaves: Satellite evolution in Bovidae	
16:45-17:15	Vincent Colot: Transposable element mobilization: where, how and with what consequences?	
17:15-17:30	Selected Abstract Fengtang Yang: Characterisation of complex genomic structure and variation by high-resolution fibre-FISH: an overview	<i>Europa Hall</i>
	Concurrent Session 8 - Accreditation, quality control and education Chairs: Konstantin Miller - Albert Schinzel	<i>Karajan Hall</i>
16:15-16:45	Thomas Liehr: European Certification and continuous education of clinical laboratory geneticists working in cytogenetics	
16:45-17:15	Thomas Eggermann: Next generation sequencing and quality assurance: Challenges and opportunities	
17:15-17:30	Selected Abstract Ron Hochstenbach: What should laboratory specialists in clinical genetics know about chromosomes ten years from now?	<i>Karajan Hall</i>
17:30-18:30	Poster session	<i>Exhibition Area</i>
18:30	E.C.A. General Assembly	<i>Europa Hall</i>
20:00	Conference Dinner of E.C.A.	



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Tuesday, 9 July 2019

09:00-10:30	Plenary Session 5 - Prenatal diagnosis Chairs: Damien Sanlaville - Maria Rosario Pinto Leite	<i>Europa Hall</i>
09:00-09:25	Rossa Chiu: Cell-free DNA analysis as a tool for “non-invasive cytogenetics”	
09:25-09:50	Nathalie Brison: The landscape of pathogenic copy number variations in healthy, reproducing females	
09:50-10:20	Selected Abstracts	<i>Europa Hall</i>
09:50-10:05	Ming Chen: A silicon-based coral-like nanostructured microfluidics to isolate rare cells in human circulation: validation by SK-BR-3 cancer cell line and its utility in circulating fetal nucleated red blood cells	
10:05-10:20	Celine Dupont: Six years of molecular cytogenetic in prenatal diagnosis: Benefits, lessons and perspectives. A new approach according to observation of ultrasound abnormalities	
10:20-10:40	Coffee break	
10:40-11:25	Plenary Session 6 Chair: Jean-Michel Dupont	<i>Europa Hall</i>
10:40-11:10	Frank Pellestor: Chromoanagenesis: cataclysms behind complex chromosomal rearrangements	
11:10-11:25	Selected Abstract Adriana Di-Battista: Balanced X-autosome translocations and premature ovarian failure are associated with altered expression of growth factors, junction organization and immune pathways	<i>Europa Hall</i>
11:25-12:15	Keynote lecture Chair: Mariano Rocchi Stylianos Antonarakis: Chromatin and single cell genomics, to understand the gene dosage imbalance in aneuploidies	<i>Europa Hall</i>
12:15	Closing Ceremony: Mariano Rocchi	<i>Europa Hall</i>