# 14th European Cytogenomics Conference Montpellier - Program

## Saturday 1 July

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| 14:30-17:30 | **Workshops and discussion meetings of Permanent Working Groups** |
| 14:30-16:30 | **PWG Quality Issues, Training and Cytogenomics and ISCN Workshop, Room Barthez, Coordinators: Martine Doco-Fenzy, Jean-Michel Dupont** |
| 14:30 | **Martine Doco-Fenzy**: Introduction and news about the update of ISO 15189 |
| 14:45 | **Melody Tabiner:** External quality assessments, contribution to quality improvement |
| 15:15 | **Ros Hastings**: Genome Mapping (optical and electronic) nomenclature and ISCN 2024 (P1003) |
| 15:45 | **Jean-Michel Dupont**: Training on ISCN: Quiz |
| 16:15 | **Marie Bérengère Troadec**: Towards a decision-making tool for the identification of chromosome structural abnormalities in conventional cytogenetics: Development of a prototype for the detection of del(5q) deletion based on artificial intelligence (P1053) |
| 16:30 | **Closing** |
| 14:30-16:30 | **PWG Neoplasia, Room Sully 2, Coordinators: Paola Caria, Harald Rieder, Roberta Vanni** |
| 14:35 | **Francesco Pasquali**: Donor cell acute myeloid leukaemia after haematopoietic stem cell transplantation for chronic granulomatous disease (P1067) |
| 14:48 | **Kalliopi Manola:** Ring chromosomes in hematological malignancies are mainly associated with myeloid malignancies and complex karyotypes (P1023) |
| 15:01 | **Uliana Karnaukhova:** Cytogenetic groups of pediatric acute myeloid leukemia from Ukraine (P1120) |
| 15:14 | **Victoria Marcu**: Validation of the OGM for cytogenomic testing in hemato-oncology – Sheba Medical Center experience (P1052) |
| 15:27 | **Bob Argiropoulos:** Laboratory Validation and Clinical Implementation of an RNA sequencing-Based Prognostic Assay for Multiple Myeloma (P1155) |
| 15:40 | **Break** |
| 15:50 | **Gulsim Smagulova:** Mutation of the PIK3CA gene in breast cancer (P1069) |
| 16:03 | **Halka Lhotska**: Detection of promoter methylation as well as deletion of MGMT gene in patients with glioblastoma using methodologically different approaches (P1097) |
| 16:16 | **Tadeusz Kałużewski**: Report on the implementation of an early cancer identification and prevention program among the population of central Poland (P1128) |
| 16:30 | **Closing** |
| 14:30-15:30 | **PWG Prenatal Diagnosis, Room Sully 3**<br>**Coordinators: Rosário Carvalho Pinto Leite, Jean-Michel Dupont** |
| 14:30 | **Jean-Michel Dupont**: Presentation of the guidelines of Microarrays in Prenatal Diagnosis |
| 14:40 | **Rosário Carvalho Pinto Leite**: Presentation of the results of the survey |
| 14:50 | **Celine Dupont**: The ever-changing face of Cytogenetics Units: Use and contribution of Whole Exome Sequencing in prenatal diagnosis (P1029) |
| 15:05 | **Joris Vermeesch**: A larger European network about cancers in pregnancy |
| 15:30 | **Closing** |
| 15:30-17:30 | **PWG Clinical and Molecular Approaches to Cytogenetic Syndromes & Cytogenomics, Room Rondollet, Coordinators: Anna Lindstrand, Damien Sanlaville, Joris Vermeesch** |
| 15:30 | **Vasheghani Farahani Faezeh**: Optical Genome Mapping: Comparing OGM with other Cytogenomics technologies. Experience on 60 individuals with developmental or fertility disorders (P1032) |
| 15:42 | **Anna Lokchine**: Primary Ovarian Insufficiency: don't neglect intragenic CNVs (P1062) |
| 15:54 | **Vladimíra Vallová**: Different strategies for the detection of copy-number variations from exome sequencing data (P1037) |

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| 16:06 | **Vincent Gatinois**: Breastfeeding promotes persistence of the mother's chimeric cells in their offspring (P1157) |
| 16:18 | **Nicolas Chatron**: Streamlining cytogenetics analysis of genome sequencing data: a comprehensive guide for Balanced Structural Variants (P1139) |
| 16:30 | **Annelies Dheedene**: Copy number detection in exome sequencing data for patients with neurodevelopmental disorders: an effective approach (P1154) |
| 16:42 | **Leona Morožin Pohovski**: First case report of a patient with three copies of distal 16p12.1p11.2 (BP1-BP3 region) and four copies of proximal 16p11.2 (BP4-BP5 region) inherited from both parents (P1154) |
| 17:04 | **Anna Lengyel**: Expanding the phenotype of 14q11.2 microdeletions encompassing CHD8 and SUPT16H genes (P1065) |
| 17:30 | **Closing** |
| 15:30-17:30 | **PWG Animal, Plant, and Comparative Cytogenetics, Room Sully 3**<br>**Coordinators; Pat Heslop-Harrison, Trude Schwarzacher** |
| 15:30 | **Pat Heslop-Harrison**: Introduction |
| 15:40 | **Magdalena Chmielewska**: Gametogenesis in hybridogenetic frogs – tracking cellular events of genome elimination and endoreduplication (P1012) |
| 15:55 | **Anna Dudzik**: Cytogenetics of the hybrid frog Pelophylax grafi and its parental species Pelophylax perezi (P1021) |
| 16:10 | **Francesca Dumas**: CAP-A satellite DNAs probe mapping on Sapajus cay paraguay and S. macrocephalus by FISH (Platyrrhini, Primates) (P1035) |
| 16:25 | **Trude Schwarzacher**: Chromosomal evolution and genome expansion in diploid oats |
| 16:40 | **Ioana Nicolae**: Cytogenetic screening of Romanian bovine breeds (P1098) |
| 16:55 | **Verónica Mestre**: A glimpse of the karyotype reshuffling from human to Myotis blythii (Vespertillionidae, Chiroptera) (P1125) |
| 17:10 | **Eleonora Pustovalova** Evolution of gametogenic pathways in reproduction of hybrid males from Pelophylax esculentus complex (P1044) |
| 17:25 | Summary/general discussion |
| 17:30 | **Closing** |
| 16:30-17:30 | **PWG Chromosomes' Integrity, Stability and Dynamics, Room Barthez**<br>**Coordinators: Jose Garcia-Sagredo, Emanuela Volpi** |
| 16:40 | **Eliane El Achkar**: Molecular Mapping of Two Replication Stress-Induced Hotspots of Breakage at the Common Fragile Site FRA11D Harboring Cancer and Neurological Genes (P1068) |
| 16:48 | **Mateus de Oliveira Lisboa**: Chromosomal Instability in Mesenchymal Stromal Cells From Acute Myeloid Leukemia Patients (P1057) |
| 17:56 | **Anna Schachner**: Monitoring of long-term cultured induced pluripotent stem cells by Optical Genome Mapping (OGM) confirms sustained fine-structural genomic stability across more than 60 in vitro passages (P1101) |
| 17:04 | **Nicoletta Selenti**: Cytogenetic analysis of induced pluripotent stem cell (iPSC) cultures derived from dermal fibroblasts (1046) |
| 17:12 | **Radhia M'Kacher**: Screening of biomarkers for chromosomal instability in the cytogenetic clinic: Present status on technological advances and their implementation into routine screening programs (P1151) |
| 17:30 | **Closing** |
| 16:30-17:30 | **PWG Marker Chromosomes, Room Sully 2**<br>**Coordinators: Thomas Liehr, Isabel Marques-Carreira** |
| 16:30 | **Thomas Liehr**: Introdution on Small Supernumerary Marker Chromosomes (sSMC) |
| 16:40 | **Christina Pérez**: Optical Genome Mapping (OGM): Validation and characterization of marker chromosomes (P1095) |
| 16:50 | **Joana Melot**: Importance of arrayCGH for sSMC detection and characterization |
| 17:00 | **Esther Cuatrecasas**: Classical genetic techniques are still in use: a case with low mosaicism (P1010) |
| 17:10 | **Thomas Liehr**: Most complex sSMC ever, as yet! |
| 17:30 | **Closing** |

## Saturday 1 July

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| 18:00-19:00 | **Conference Opening lecture**<br>**Chair: Mariano Rocchi and Franck Pellestor** |
| | **Eva R. Hoffmann**: Aneuploidy in the Maternal Germline |

## Sunday 2 July

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| 8:30-10:15 | **Plenary session 1 - Mosaicism: from Preimplantation Embryos to Aging**<br>**Chairs: Joris Vermeesch and Elisabeth Syk Lundberg** |
| 8:30-9:00 | **Antonio Capalbo**: Mosaicism in Preimplantation Embryos |
| 9:00-9:30 | **Malgorzata I. Srebniak**: Mosaicism in Prenatal Diagnosis: from NIPT to Amniocytes Investigation |
| 9:30-10:00 | **Lars A. Forsberg**: Hematopietic Loss of Chromosome Y and Higher Mortality in Men |
| | Selected abstract |
| 10:00-10:15 | **Cornelia Daumer-Haas**: Normal Array-CGH Results in a Patient With Short Stature and Global Developmental Delay Carrying a de novo Ring Chromosome 2p and a Chromosome 2q Derivative With a Neocentromere |
| 10:15-10:45 | Coffee break |
| 10:45-12:00 | **Plenary session 2 - Cancer Cytogenomics**<br>**Chairs: Felix Mitelman and Roberta Vanni** |
| 10:45-11:15 | **Sarah McClelland**: Replication Stress Generates Distinctive Landscapes of DNA Copy Number Alterations and Chromosome Scale Losses in Cancer |
| 11:15-11:45 | **Uri Ben-David**: Whole-Genome Duplication Shapes the Aneuploidy Landscape of Human Cancers |
| | Selected abstract |
| 11:45-12:00 | **Christina Srouji**: Optical Genome Mapping for Multiple Myeloma: Evaluation of The Technology in a Clinical Laboratory. |
| 12:00-14:30 | **Poster session and Satellite Symposia** |
| 14:30-15:45 | **Concurrent Session 1 - Recent Advances in Cytogenomics**<br>**Chairs: Franck Pellestor and Harald Rieder** |
| 14:30-15:00 | **Alex Hoischen**: Optical Mapping to Karyotype |
| 15:00-15:30 | **Antonio Rausell**: Artificial Intelligence in Cytogenetics |
| | Selected abstract |
| 15:30-15:45 | **Gil Nifker**: Dam Assisted Fluorescent Tagging of Chromatin Accessibility (DAFCA) for Optical Genome Mapping in Nano-Channel Arrays |
| 14:30-15:45 | **Concurrent Session 2 - Beyond Genome Sequencing: the Epigenetic Signature**<br>**Chairs: Orsetta Zuffardi and Joan Blanco** |
| 14:30-15:00 | **Bekim Sadikovic**: DNA Methylation Episignatures Associated with Large Structural Copy Number Variants: Clinical Implications |
| 15:00-15:30 | **Karen Temple**: Multi-locus imprinting disorders |
| | Selected abstract |
| 15:30-15:45 | **Mathilde Geysens**: Long Read Whole Genome Sequencing for The Detection of Structural and Epigenetic Variation in Developmental Disorders |
| 15.45-16.15 | Coffee break |
| 16:15-17:30 | **Plenary session 3 - Newly Emerged Technologies in Cytogenomics**<br>**Chairs: Pat Heslop-Harrison and Emanuela Volpi** |
| 16:15-16:45 | **Brian Beliveau**: Paint-SHOP; Genome-Scale Oligonucleotide FISH Experiments |
| 16:45-17:30 | **Pat Heslop-Harrison and Emanuela Volpi**: Interactive Discussion; Massive Oligonucleotide Pools to Track Organization and Evolution of Chromosomes and Genomes |
| 17:30-18:30 | **Poster session** |

## Monday 3 July

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| 8:30-10:30 | **Plenary session 4 - Clinical Cytogenomics I**<br>**Chairs: Damien Sanlaville and José Garcia-Sagredo** |
| 8:30-9:00 | **Anna Lindstrand**: Complex Genomic Rearrangements: an Underestimated Cause of Rare Diseases |
| 9:00-9:30 | **Orsetta Zuffardi**: Distal Germ-Line Deletions in Mosaic With Copy-Neutral Loss of Heterozygosity: Something to Be Considered in Genetic Counselling |
| 9:30-10:00 | **Brunella Franco**: From Gene Disruption to Missense Variants: how Different Types of Variants Influence the X-Linked Inheritance Model |
| | Selected abstracts |
| 10:00-10:15 | **Niels Tommerup**: Burden of Long Range Position Effects in Balanced Chromosomal Rearrangements |
| 10:15-10:30 | **Marlene Ek**: Multiomic Profiling Unravels Disease Mechanisms in Complex Chromosomal Rearrangements and Marker Chromosome Carriers |
| 10:30-11:00 | Coffee break |
| 11:00-12:15 | **Concurrent Session 3 - Clinical Cytogenomics II**<br>**Chairs: Orsetta Zuffardi and Martine Doco-Fenzy** |
| 11:00-11:30 | **Nicolas Chatron**: Structural Variants in Clinical Practice Using Genome Sequencing |
| 11:30-12:00 | **Caroline Schluth-Bolard**: Constitutional Chromoanagenesis: From Diagnosis to Genetic Counselling |
| | Selected abstract |
| 12:00-12:15 | **Sanam Khan**: Systematic X-Inactivation Studies of Sequence Resolved Balanced X Chromosomal Rearrangements |
| 11:00-12:15 | **Concurrent Session 4 - Animal and Plant Cytogenomics I**<br>**Chairs: Tony Heitkam and Trude Schwarzacher** |
| 11:00-11:30 | **Mathieu Rouard**: Comparative Genomics and Tools for Studying Chromosome Evolution |
| 11:30-12:00 | **Brankica Mravinac**: Coleopteran Satellite Profiles: Chromosomal and Sequence Organization |
| | Selected abstract |
| 12:00-12:15 | **Maria Filomena Lopes Adega**: A physical map of repetitive elements in the genomes of Iberian Peninsula chiropteran species |
| 12:15-14:30 | **Poster session and Satellite Symposia** |
| 14:30 - 15:45 | **Plenary session 5: Nuclear Organization and Diseases**<br>**Chairs: Jean-Michel Dupont and Emanuela Volpi** |
| 14:30 - 14:55 | **Giacomo Cavalli**: The Role of 3D Genome Organization in The Regulation of Gene Expression and Cell Fate (to be comfrmed) |
| 15:00-15:30 | **Irina Solovei**: Spatial Organization of Transcribed Eukaryotic Genes |
| | Selected abstract |
| 15:30-15:45 | **Paola Caria**: 3D Nuclear Architecture Distinguishes Thyroid Cancer Histotypes |
| 10:45-16:15 | Coffee break |
| 16:15-17:30 | **Concurrent Session 5 - Animal and Plant Cytogenomics II**<br>**Chairs: Trude Schwarzacher and Brankica Mravinac** |
| 16:15-16:45 | **Tony Heitkam**: Adding a Chromosome Perspective to Plant Genomics: Making Sense of Retained Retroviruses, Moving Retrotransposons and Expanding Satellite DNAs |

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| 16:45-17:15 | **Yi Tzu Kuo**: Plasticity in centromere organization: A few megabased-sized centromere units can form a holocentromere |
| | Selected abstract |
| 17:15-17:30 | **Alla Krasikova**: The first nuclear and cytoplasmic whole transcriptome profile of chicken oocytes at the lampbrush chromosome stage |
| 16:15-17:30 | **Concurrent Session 6 - Accreditation, Quality Control and Education**<br>**Chairs: Konstantin Miller and Martine Doco** |
| 16:15-16:45 | **Folker Spitzenberger**: The New ISO 15189 Standard Medical Laboratories |
| 16:45-17:15 | **Johan den Dunnen**: Sequence-based Nomenclature and the Novelties to Come in the Next ISCN Version |
| | Selected abstract |
| 17:15-17:30 | **Mathilde Quibeuf**: Educational Benefits of Analysing Highly Complex Chromosomal Rearrangements Such as Chromoanagenesis by Long Read Approaches |
| 17:30-18:30 | Poster session |

## Tuesday 4 July

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| 8:30-10:30 | **Plenary session 6 - Prenatal Diagnosis and Preimplantation**<br>**Chairs: Jean-Michel Dupont and Rosário Pinto Leite** |
| 8:30-9:00 | **Robert-Jan H. Galjaard**: Genome-Wide Noninvasive Prenatal Testing: Follow-Up Results of the TRIDENT-2 Study |
| 9:00-9:30 | **Joris Vermeesch**: Fragmentomics and Non Invasive Prenatal Screening (NIPS) |
| 9:30-10:00 | **Lyn Chitty**: Prenatal Diagnostic Yield and Pitfalls Through Arrays, Exome, and NIPT |
| | Selected abstracts |
| 10:00-10.15 | **Armelle Duquenne**: Multicentric Longitudinal Performance Monitoring of Different non-Invasive Prenatal Screening Technologies Used in Belgium |
| 10:15-10.30 | **Ludovica Picchetta**: Triploid Conceptions Are Predominantly Caused by Female Meiosis II Errors and Their Risk Increases with Advancing Maternal Age |
| 10:30-10.50 | Coffee break |
| 10:50-11.30 | Satellite Symposia |
| 11:30-12:20 | **Closing keynote**<br>**Chairs: Mariano Rocchi and Thierry Lavabre-Bertrand**<br>**Michael E.Talkowski**: The Landscape of Structural Variation Across Diverse Global Populations and Developmental Disorders |
| 12:20 | **Closing ceremony** |