European Cytogeneticists Association

15th EUROPEAN CYTOGENOMICS CONFERENCE

June 29-July 1, 2025

LEUVEN, BELGIUM

www.eca2025.org

FINAL PROGRAMME BOOK

WELCOME MESSAGE

Dear Members of the ECA Community, Colleagues, and Friends,

It is my great pleasure to welcome you in Leuven, Belgium for the 15th European Cytogenomics Conference, the flagship event of the ECA, bringing our community together in every two years to share knowledge, foster collaborations, and celebrate progress in our field.

This year, we continue to build a bridge between the foundational principles of classical cytogenetics and the latest technological innovations, including high-throughput, high-fidelity sequencing. We are also pleased to introduce a new component to the programme: hands-on workshops taking place on the pre-conference day, just before the permanent working group meetings.

The scientific programme features a diverse range of sessions covering cutting-edge research, including the role of structural variation and nuclear organization in disease. These sessions are complemented by updates on Clinical Cytogenomics, Cancer Genomics, Prenatal Diagnosis, Animal and Plant Cytogenomics, and more.

As always, we aim to promote the exchange of ideas and encourage interaction across disciplines. Many of you have contributed abstracts, and selected posters are being presented as oral talks or as short, focused presentations within the Permanent Working Groups — providing a valuable opportunity to share and discuss your work with peers.

We are especially proud to host this conference at KU Leuven, an institution at the heart of European academia for six centuries. As KU Leuven marks its 600th anniversary in 2025, the city is alive with celebrations — making this a particularly special time to be here.

We are truly happy to be here in Leuven together with all of you.

Once again, welcome to ECA 2025!

Mariano Rocchi
President of the European Cytogeneticists Association (E.C.A.)

COMMITTEES

E.C.A. COMMITTEE

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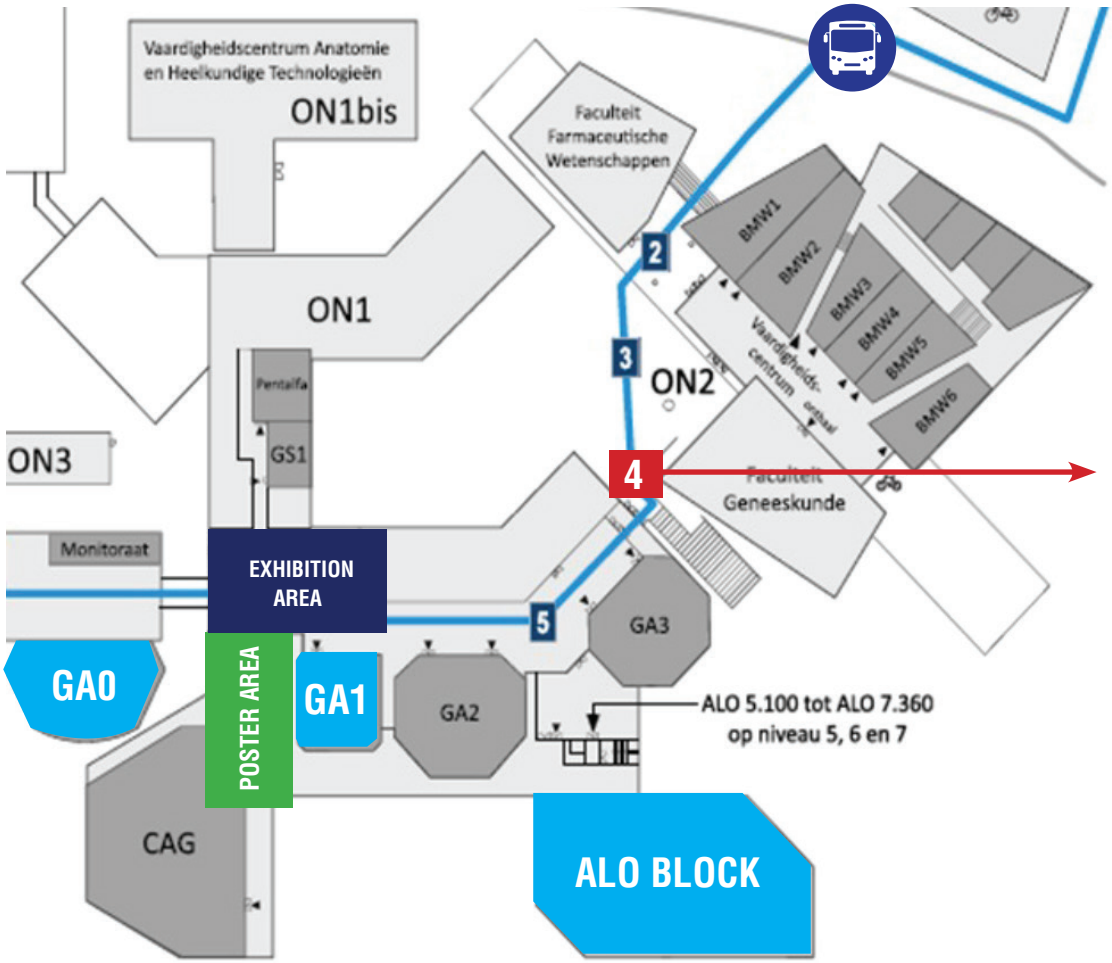
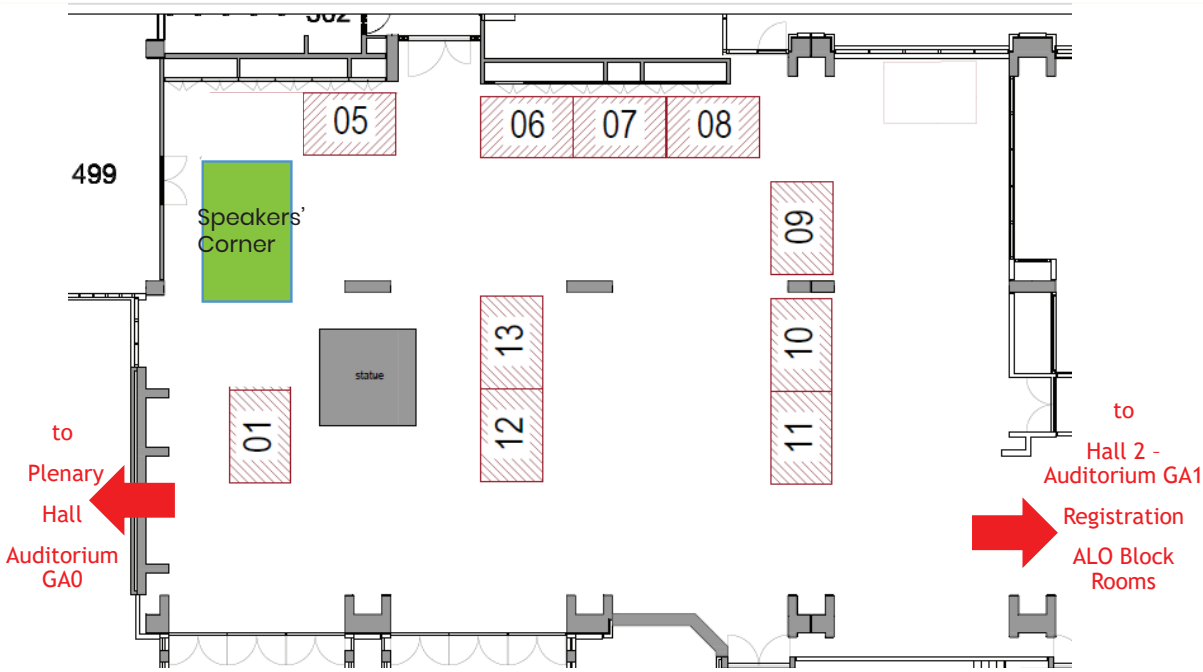
LAYOUT



EXHIBITION FLOOR PLAN



Exhibition Floorplan

01- Bionano
05- Genome Diagnostics
06- Thermo Fisher Scientific
07- Agilent Technologies
08- Sinochrome
09- ASI
10- Bioview
11- ADS Biotec
12- Metasystems
13- Metasystems

GENERAL INFORMATION

VENUE

KU Leuven
Gasthuisberg Campus
Onderwijs & Navorsing 1 (O&N 1) Building

HOW TO ARRIVE

https://eca2025.org/files/downloads/ECA2025-How-to-arrive-the-conference-venue.pdf

CME INFORMATION

European Accreditation Council for Continuing Medical Education (EACCME)
An institution of U.E.M.S.

The **15th EUROPEAN CYTOGENOMICS CONFERENCE**, Leuven, Belgium 29/06/2025 - 01/07/2025, has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME®) with **16.5 European CME credits (ECMEC®s)**. Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

Through an agreement between the Union Européenne des Médecins Spécialistes and the American Medical Association, physicians may convert EACCME® credits to an equivalent number of AMA PRA Category 1 Credits™. Information on the process to convert EACCME® credit to AMA credit can be found at https://edhub.ama-assn.org/pages/applications.

Live educational activities, occurring outside of Canada, recognised by the UEMS-EACCME® for ECMEC®s are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.

Information regarding the conversion of EACCME® credits

Credit will be converted based on one (1) hour of participation equalling one credit for all activities. Physicians wishing to convert EACCME® credits to AMA PRA Category 1 Credit™ will be required to access the AMA website at https://edhub.amaassn.org/pages/applications to obtain the necessary paperwork and instructions. Physicians and other health care professionals will be required to pay a processing fee to the AMA.
For other countries, please contact the relevant national/regional accreditation authority.

CERTIFICATES OF ATTENDANCE & CME CERTIFICATES

After the ECA 2025 conference concludes, attendees will be able to access their Certificate of Attendance and CME (Continuing Medical Education) certificate directly through the registration platform.

PROGRAM

Sunday, June 29		
9:30-11:00	**Hands-on workshops / 1st Slots**	
	Analysis of clinical and haemotological samples, in group of three (the computer connects directly to the Bionano US server in US), ***Dana JABER***	**ALO 05.100**
	Bridging cytogenetics workflows: Hands-on analysis of arrays to NGS, ***Eithan ZAND, Marco RUSSMAN***	**ALO 05.200**
	The use of online resources to annotate/map clinical CNVs, ***Robert KUHN***	**ALO 07.100**
	Nanopore based sequencing for structural variant detection, from sample to report, ***"WET LAB", Erwin REILING, Erika SOUCHE***	**Meet. Point: Registration**
11:15-12:45	**Hands-on workshops / 2nd Slots**	
	Analysis of clinical and haemotological samples, in group of three (the computer connects directly to the Bionano US server in US), ***Dana JABER***	**ALO 05.100**
	Bridging cytogenetics workflows: Hands-on analysis of arrays to NGS, ***Eithan ZAND, Marco RUSSMAN***	**ALO 05.200**
	The use of online resources to annotate/map clinical CNVs, ***Robert KUHN***	**ALO 07.100**
	Nanopore based sequencing for structural variant detection, from sample to report, ***"DRY LAB", Erwin REILING, Erika SOUCHE***	**ALO 07.200**

PROGRAM

14:30-17:30 ***Permanent Working Group Meetings***

14:30-16:30	**Animal, plant, and comparative cytogenetics** PWG Coordinators: **Pat HESLOP-HARRISON**		
14:30-14:35	**Coordinators**	Welcome and foreword by PWG Coordinators	**ALO 05.100**
14:35-14:45	Andreas HOUBEN	Does chromoanagenesis play a role in the origin of B chromosomes?	
14:45-14:55	Alla KRASIKOVA	Retrotransposable elements drive transcription of tandem repeats	
14:55-15:05	Alessandra IANNUZZI	Exploring Telomere Length as a Biomarker of Well-Being in Aglianico Grapevines	
15:05-15:15	Liliana BURIBASA	Bivalent marker dynamics in protamine expression: unraveling chromatin compaction mechanisms	
15:15-15:25	Ioana NICOLAE	Cytogenetic investigations in Romanian Black and White Spotted cattle	
15:25-15:35	Lyubov MALINOVSKAYA	Germline-restricted chromosome during embryogenesis in sand martin (riparia riparia)	
15:35-15:45	Ahmet L. TEK	A novel model for functional centromere composition in soybean and Glycine soja	
15:45-15:55	Alessia DAPONTE	Unraveling the genetic architecture of centromeres with CENdetectHOR	
15:55-16:10	Paulina TOMASZEWSKA	Repetitive DNA sequences mark genome boundaries in the terrestrial orchid epipactis zinn	
16:10	**Coordinators**	General discussion and conclusive remarks by PWG Coordinators	

PROGRAM

14:30-16:30	**Neoplasia** PWG Coordinators: **Harald RIEDERE, Paola CARIA**		
14:30-14:35	**Coordinators**	Welcome and foreword by PWG Coordinators	**ALO 07.100**
14:35-14:45	Tadeusz KALUZEWSKI	Evaluation of the Utility of TERT Promoter Mutations in the Early Detection of Urothelial Cancer	
14:45-14:55	Marija DENCIC FEKETE	Distribution of gene aberrations in chronic lymphocytic leukemia by NGS testing in a Serbian patient cohort	
14:55-15:05	Marie-Bérengère TROADEC	What is wrong with the deletion of chromosome region 5q in myelodysplastic syndrome? Identification of a novel actor of the sensitivity to lenalidomide of MDS with del(5q)	
15:05-15:15	Uliana LYKHOVA	Beyond t(12;21): unveiling the hidden layers in all karyotypes	
15:15-15:25	Laura YISSEL RENGIFO	Dynamic Follow Up Of Tumor Burden In Multiple Myeloma Through Analysis Of Ccfdna Markers	
15:25-15:35	Seon Y KIM	Detection of measurable residual disease using fluorescence in situ hybridization compared with multiparametric flow cytometry in patients with B-lymphoblastic leukemia	
15:35-15:45	Hila LEDERMAN NACHMIAS (pending)	New Insights Affecting Classification, Prognosis and Treatment of Multiple Myeloma Using Optical Genome Mapping	
15:45-15:55	Soumaya MAUGOU-ZERELLI (pending)	Mapping Cancer Risk in Constitutional Chromosomal Deletions: A Cytogenetic Analysis	
15:55-16:30	**Coordinators**	General discussion and conclusive remarks by PWG Coordinators	

PROGRAM

15:30-17:30	Clinical and molecular approaches to cytogenetic syndromes & cytogenomics PWG Coordinators: **Joris VERMEESCH, Anna LINDSTRAND, Damien SANLAVILLE**		**Auditorium GA1**
15:30-15:35	**Coordinators**	Welcome and foreword by PWG Coordinators	
15:35-15:45	Paola EVANGELIDOU	A rare and complex case of a male patient with DiGeorge – like phenotype, carrying three different mosaic copy number variants on chromosome 22	
15:45-15:55	Caroline SCHLUTH-BOLARD	FGF14 disruption by constitutional chromoanagenesis as a cause of spinocerebellar ataxia	
15:55-16:05	Leslie KULIKOWSKI	Resolving the Unresolved: Epigenomic Profiling as a Diagnostic Tool for Copy Number Variants of Uncertain Significance	
16:05-16:15	Martine DOCO-FENZY	Invdupdel Or Duptrp Rearrangements Revisited Using Array-CGH And Optical Genome Mapping	
16:15-16:25	Lusine NAZARYAN-PETERSEN	Detection Of Structural Variants By Short Read Whole Genome Sequencing And Interpretation For Genetic Diagnosis	
16:25-16:35	Marlene EK	Long Read Genome Sequencing Enhances Diagnosis Of Pediatric Neurological Disorders	
16:35-16:45	Igor LEBEDEV	X Chromosome Cnv Reclassification Integrating X Inactivation Status For Improved Pathogenicity Assessment	
16:45-16:55	Esmee TEN BERK DE BOER	Investigating X Chromosome Inactivation Patterns In X Autosome Translocations Using Long Read Sequencing And The T2t Genome Assembly	
16:55-17:30	**Coordinators**	General discussion and conclusive remarks by PWG Coordinators	

PROGRAM

16:30-17:30	**Chromosomes' integrity, stability, and dynamics** PWG Coordinators: **José M. GARCIA-SAGREDO, Emanuela VOLPI** ***Exploring new chromosomal paradigms for precision medicine and early disease detection***		
16:30-16:40	**Coordinators**	Welcome and foreword by PWG Coordinators	
16:40-16:50	Ulrike MAU-HOLZMANN	Multiple Variable Chromosomal Aberrations in Primary Fibroblasts: Further Hints to Chromosomal Instability as a Long-Term Effect Even Years After Irradiation	
16:50-17:00	Claudia OLIVEIRA	The DEB Test Beyond Fanconi anaemia: A new look into chromosome instability	**ALO 05.100**
17:00-17:10	Zuzanna GRACZIK	Impact of sperm fractioning on chromosome positioning, chromatin integrity and DNA methylation level	
17:10-17:20	Radhia M'KACHER	Telomere Dysfunction, DNA Breaks, Chromosomal Aberration Formation and the Dark Side of the Centromere	
17:20	**Coordinators**	Conclusive remarks and new initiative announcement by PWG Coordinators	
16:30-17:30	**Prenatal diagnosis** PWG Coordinators: **Rosário Carvalho PINTO LEITE, Jean-Michel DUPONT**		
16:30 -16:35	**Rosário Carvalho PINTO LEITE**	Introduction	
16:35 -16:45	A.VARDANYAN	Retrospective analysis of cytogenetic findings in pregnant women at risk following first-trimester screening: insights from NIPT in Armenia	
16:45 - 16:55	M.A.CARO MIRO	Circuit of prenatal screening with free circulating fetal DNA in the balearic islands	**ALO 07.100**
16:55 - 17:10	Rosário Carvalho PINTO LEITE	NIPT in Europe, Result of the PWG survey	
17:10 - 17:20	K.CASSINARI	First Prenatal Case of Jumping-like Translocations: Unraveling Complex Chromosomal Rearrangements	
17:20 - 17:30	N.CHATRON	Comparative Efficacy of cfDNA and aCGH in Detecting Chromosomal Aberrations Post-Miscarriage	
18:00-19:00	**Opening Lecture** **Evan E. EICHLER "Complete chromosomes and complex genomes"** Chaired by **Mariano ROCCHI, Joris VERMEESCH**		**GAO**
19:00-20:30	**ECA 2025 Kick-off party**		**Exhibition Area**

PROGRAM

Monday, June 30			
8:30-10:15	**Plenary session 1** **Structural variation in health and disease** Chaired by: **Joris VERMEESCH, Anna LINDSTRAND**		
8:30-9:00	**Alexander REYMOND**	The pleiotropic spectrum of proximal 16p11.2 CNVs	**GAO**
9:00-9:30	**David PORUBSKY**	Structural variation of 22q11.2 region in normal and diseased human population	
9:30-10:00	**Tobias MARSCHALL**	Mapping structural variation in the pangenome	
10:00-10:15	**Nivin MOUSTAFA-HAWASH**	Optical genome mapping in the clinic reveals germline and somatic findings that may influence the treatment approach	
10:15-10:45	**COFFEE BREAK**		**Exhibition Area**
10:45-12:15	**Plenary session 2** **Complexity of cancer genomes** Chaired by **Roberta VANNI, Barbara DEWAELE**		
10:45-11:15	**Jonas DEMEULEMEESTER**	Multiomic long-read sequencing to improve diagnosis and care of genomically complex sarcomas	**GAO**
11:15-11:45	**Stefano SANTAGUIDA**	Mechanistic insights into the consequences of chromosome segregation errors on cell physiology	
11:45-12:00	**Amber VERHASSELT**	Optical genome mapping is a powerful diagnostic tool in non Hodgkin lymphoma	
12:00-12:15	**Şule ALTINER**	Cytogenetic profile of hematological malignancies with complex karyotype a single center study from Turkey	
12:15-14:30	**POSTER SESSION**		**Poster Area**
12:30-14:30	**Sponsored Workshop: Next-gen cytogenetics: Applications of AI and digital FISH in diagnostics** Speakers: **Jana BUKOLSKÁ, Yarin HADID, Lee KAPLAN**		**ALO 05.100**
14:30-15:30	**Concurrent Session 1** **Meiosis and Mitosis** Chaired by **Jean-Michel DUPONT, Elisabeth SYK LUNDBERG**		
14:30-15:00	**Marta DE RUIJTER VILLANI**	Meiosis/mitosis transition	**GAO**
15:00-15:30	**Carolina VILLARROYA-BELTRI**	Mosaic variegated aneuploidy in development, ageing and cancer	

PROGRAM

14:30-15:30	**Concurrent Session 2** **Automation and AI in Clinical Genetics** Chaired by **Barbara DEWAEKE, Franck PELLESTOR**		**GA1**
14:30-15:00	**Claudia HAFERLACH**	Application of AI in hematological diagnostics	
15:00-15:30	**Robert KUHN**	Online resources at UCSC	
15:30-15:45	**COFFEE BREAK**		**Exhibition Area**
15:45-17:30	**Plenary Session 3** **Clinical Cytogenomics** Chaired by **Damien SANLAVILLE, Orsetta ZUFFARDI**		**GA0**
15:45-16:15	**Thomas BOURGERON**	The genetic architecture of autism: from medicine to neurodiversity	
16:15-16:45	**Jesper EISFELDT**	Long read genome sequencing in clinical cytogenomics	
16:45-17:15	**Andrea CIOLFI**	DNA methylation profiling as a diagnostic tool	
17:15-17:30	**Dominik REZNY**	Precision approaches in clinical cytogenomics the role of optical genome mapping and long read sequencing in structural variant detection	
17:30-20:00	**POSTER SESSION**		**Poster Area**
18:00-19:00	***ECA General Assembly***		**GA0**

PROGRAM

Tuesday, July 1			
8:30-10:30	**Plenary Session 4** **Animal, Plant and Comparative Cytogenomics** Chaired by **Pat HESLOP-HARRISON, Mariano ROCCHI**		
8:30-9:00	**Aurora RUIZ-HERRERA**	Evolution and function of 3D chromatin folding	**GA0**
9:00-9:30	**Julie SARDOS**	Diversity and diversification in banana: how in silico chromosome painting opens new perspectives for the conservation and use of an iconic fruit	
9:30-10:00	**Pat HESLOP-HARRISON**	What cytogenomics has done, and is doing, for agriculture in our world	
10:00-10:15	**Simon MALLET**	Interstitial telomeric sequences and accumulation of dna damage hallmarks of genomic ınstability ın cancer resistant wild vertebrates	
10:15-10:30	**Fengtang YANG**	Genomic complexity and evolutionary plasticity ın dugesia japonica revealed by multi ploidy chromosome level assemblies	
10:30-11:00	**COFFEE BREAK**		**Exhibition Area**
11:00-12:15	**Concurrent Session 3** **Nuclear organisation and disease** Chaired by **Emanuela VOLPI, Pat HESLOP-HARRISON**		**GA1**
11:00-11:30	**Martin MENSAH**	Nucleolar dysfunction in rare genetic diseases	
11:30-12:00	**Cristina CARDOSO**	Epigenetic reprogramming and disease	
12:00-12:15	**Lusine NAZARYAN-PETERSEN**	Detection of structural variants by short read whole genome sequencing and interpretation for genetic diagnosis	
11:00-12:15	**Concurrent Session 4** **Clonal correction of constitutional chromosome imbalances** Chaired by **Damien SANLAVILLE, Orsetta ZUFFARDI**		**GA0**
11:00-11:30	**Diane VAN OPSTAL**	Placental cytogenetic studies provide a glimpse into the black box of early embryogenesis	
11:30-12:00	**Alfredo BRUSCO**	Somatic recombination and the removal of the structural variant: any phenotypic outcome?	
12:00-12:15	**Anikó UJFALUSI**	Evaluation of X-ınactivation pattern ın carriers of X chromosome aberrations and DMD gene mutations	

PROGRAM

12:15-14:00	**POSTER SESSION**		**Poster Area**
12:30-13:15	**Sponsored Session:**		
12:30-12:45	**Drew ELLERSHAW**	Introduction	**GA1**
12:45-13:05	**Inga NAGEL**	Addressing clinical challenges in rare diseases through long-range insights by a novel genome sequencing technology	
13:05-13:15		Q&A	
14:00-15:45	**Concurrent Session 5** **Accreditation and workshop on ISCN** Chaired by **Franck PELLESTOR, Harald RIEDER**		**GA0**
14:00-14:30	**Konstantin MILLER**	ISO15189 and cytogenetic laboratories	
14:30-15:45	**Jean-Michel DUPONT**	Workshop on ISCN 2024	
14:00-15:15	**Concurrent Session 6** **Applied Cytogenotoxicity** Chaired by **José GARCIA SAGREDO, Joan BLANCO**		**GA1**
14:00-14:30	**Alba HERNADEZ BONILLA**	Genotoxicity and carcinogenicity of long-term micro- & nano-plastics exposure: current understanding and future directions	
14:30-15:00	**Ans BAEYENS**	Chromosomal radiosensitivity testing for inborn errors of immunity	
15:00-15:15	**Marlene EK**	Long-read genome sequencing enhances diagnosis of pediatric neurological disorders	
15:45-16:15	**COFFEE BREAK**		**Exhibition Area**
16:15-17:30	**Plenary Session 5** **Prenatal Diagnosis and Preimplantation** Chaired by **Elisabeth SYK LUNDBERG, Rosario PINTO LEITE**		**GA0**
16:15-16:45	**Alan HANDYSIDE**	PGT, with a focus on aneuploidies	
16:45-17:15	**Nathalie JANEL**	Prenatal treatment of chromosomal anomalies	
17:15-17:30	**Charlotte TARDY**	Transforming prenatal cytogenetics rapid chromosomal rearrangement characterization with Nanopore sequencing	
17:30-18:30	**Closing Keynote** **Joris VERMEESCH "Cytogenomics, where we are and where we are heading"** Chaired by **Mariano ROCCHI, Jean-Michel DUPONT**		**GA0**
18:30	**Closing Ceremony**		**GA0**

****Rooms**** The rooms of plenary and concurrent sessions (GA0 and GA1) are on the same floor as the reception area, posters, and company booths. Due to the building's underground levels, this is considered the 4th floor. Consequently, rooms AL0 0**5**.xxx, and AL0 **7**.xxx are located one, and three floors above the GA0/GA1 rooms, respectively.

POSTER LIST

Animal and Plant Cytogenomics

P-1006	Andreas Houben	Does chromoanagenesis play a role in the origin of B chromosomes?
P-1009	Alla Krasikova	Retrotransposable elements drive transcription of tandem repeats
P-1051	Liliana Burlibasa	Bivalent marker dynamics in protamine expression: unraveling chromatin compaction mechanisms
P-1054	Ioana NICOLAE	Cytogenetic investigations in Romanian Black and White Spotted cattle
P-1063	Lyubov Malinovskaya	germline-restricted chromosome during embryogenesis in sand martin (riparia riparia)
P-1093	Alessia Daponte	Unraveling the genetic architecture of centromeres with CENdetectHOR
P-1137	Paulina Tomaszewska	repetitive dna sequences mark genome boundaries in the terrestrial orchid epipactis zinn

Clinical Cytogenomics

P-1007	Eva Pinti	Duplication of 13q – A rare chromosomal abnormality
P-1008	Nadja Kokalj Vokac	Copy number variations in males with unexplained azoospermia
P-1011	Eleana Rraku	Developing Del2Phen: a novel phenotype description tool for chromosome deletions
P-1016	Valentyna Kurakova	Cytogenetic analysis of a complex ring chromosome 5 with 5p15.2 duplication in a child with congenital developmental delay
P-1017	Sinem YALCINTEPE	A case with both SMARCA2 pathogenic variant and 14q24.1-q32.33 deletion with the only finding hypocalciuric hypercalcemia
P-1021	Eusebiu Vlad Gorduza	a rare case of carrier of two different balanced chromosomal rearrangements
P-1024	Hara Tsimela	Investigation of 30 couples with infertility with Fluorescent in situ hybridization (fish) and Correlation with their karyotypic findings
P-1025	Marta Olszewska	Optical genomic mapping (OGM) and whole-genome sequencing (WGS) reveal a double reciprocal chromosomal translocation (RCT) and a chromosome 13 rearrangement in an infertile male with normozoospermia
P-1028	Anita SY Kan	Ring chromosomes in Hong Kong: 173 cases diagnosed through a 23 years period in two centers
P-1030	Armelle Duquenne	Prenatal Sex Discrepancy Between NIPS and Phenotype: A Rare Y Chromosome Rearrangement with Neocentromere Formation
P-1031	Mui Li Tan	Utility of Optical Genome Mapping in routine cytogenetics laboratory workflow: a presentation of two cases
P-1036	Társis Paiva Vieira	molecular characterization of human ring chromosomes and complex genomic rearrangements using optical genome mapping and short-read genome sequencing
P-1038	Cristina Candeias	Developmental Delay and Dysmorphic Features unravel a Marker Chromosome with a Neocentromere Derived from Chromosome 8

POSTER LIST

P-1041	Manuela Mota Freitas	Unravelling a Complex Case of del(9)(p24) and Subtelomeric del(4q) in a 46,XY Female Patient
P-1043	Caroline Schluth-Bolard	FGF14 disruption by constitutional chromoanagenesis as a cause of spinocerebellar ataxia
P-1044	LESLIE kULIKOWSKI	Resolving the Unresolved: Epigenomic Profiling as a Diagnostic Tool for Copy Number Variants of Uncertain Significance
P-1045	Dezso David	A personalized genomic medicine approach to rare genomic disorders associated with simple chromosomal structural variants
P-1048	Rim Khelifi	Comprehensive analysis of copy number variations in Tunisian patients with congenital heart defects
P-1049	Bochra Khadija	Copy number variations (CNV) found in Tunisian patients with corpus callosum malformations
P-1052	DOCO-FENZY Martine	Invdupdel or duptrp rearrangements revisited using Array-CGH and Optical Genome Mapping
P-1057	Afia Hasnain	characterization of complex chromosomal structural variation: a comparison of cytogenetic methods
P-1058	Irina Puppo	Chromosomal abnormalities in infertile Armenian couples: a cytogenetic analysis
P-1059	Aleksandra Lesniewska	Additional Y chromosome in 47,XYY – implications for male infertility
P-1060	Rezacova Hana	Optical Genome Mapping in patients with neurodevelopmental disorders, our first experience.
P-1061	TEA MLADENIC	De Novo Deletion of 18p in a Female with Short Stature and Premature Ovarian Failure: A Case Report
P-1064	Uwe Heinrich	Mosaic trisomy 14 in a child with multiple congenital anomalies due to an unstable Robertsonian translocation involving chromosomes 14 and 22
P-1065	Rashmi Shukla	Cytogenetic and molecular characterisation of chromoanasynthesis in a child with multiple congenital anomalies
P-1068	Annalaura Montanari	unraveling genetic variability in reproductive failure: a novel approach to infertility diagnosis
P-1069	Ilona Dietze-Armana	A rare Y-autosome reciprocal translocation t(Y;17) found in a patient with azoospermia
P-1070	Susanne Anders	Maternally inherited gain Xq24 encompassing the CUL4B gene causative for X-linked intellectual disability
P-1072	Bettina Pfütze	A complex chromosomal translocation in a young girl with mental retardation
P-1075	Nikoletta Selenti	Chromosomal abnormalities in reproductive health: data from cytogenetic analysis in a Greek cohort the last decade
P-1076	Anna Lengyel	A family with multiple recurrent copy number variations associated with increased risk for neurodevelopmenetal disorders
P-1077	Anouck Schneider	Decoding Complex Chromosomal Rearrangements and their genetic impacts

POSTER LIST

P-1078	Sylke Singer	Maternal cryptic inv(16)(p13.3q24.3) leading to opposite recombinant chromosomes 16 in two daughters
P-1079	Miroslav Stolfa	Comparative Evaluation of CNVs Detection: A Case Study of Optical Genome Mapping and Long-Read Whole-Genome Sequencing versus Chromosomal Microarray
P-1080	Bárbara Marques	MULTIPLE NON-CONTIGUOUS INTERSTITIAL DELETIONS IN 5Q21Q22.1, INCLUDING THE CHD1 GENE, IDENTIFIED IN A BOY WITH DEVELOPMENTAL DELAY AND SEVERE LANGUAGE IMPAIRMENT.
P-1081	Fanni Toth-Szumutku	Exome sequencing identifies copy number variants associated with 22q11.2 deletion syndrome-like phenotype
P-1090	Fatma Maazoun	Biological Implementation of Optical Genome Mapping in Recurrent Miscarriages and Implantation Failure: A Comprehensive Evaluation
P-1094	Simon Schabat	Enhancing Clinical Cytogenetics: Automated Detection of Chromosomal Aberrations Using Telomere and Centromere Staining
P-1095	Vincent Gatinois	Prenatal diagnosis of a recombinant chromosome from a parental insertion: from karyotype to whole genome sequencing, the cytogeneticist's eye remains necessary!
P-1100	BOUCHAHTA HICHAM	Maternally Derived Complex Small Supernumerary Marker Chromosome 22 Associated with Cat-Eye Syndrome Like Features
P-1101	Saad Alomar	A cytogenetic Study of Down's Syndrome in Iraqi Population
P-1107	Candice Saurin	Optical genome mapping as a first-tier tool for Y chromosome structural variations analysis?
P-1109	Omar A.Z. Tutakhel	RORB gene: A novel interstitial microdeletion characterized by speech delay and hypertelorism
P-1111	Wiem Ayed	Genetic Investigation Of Tunisian Infertile Men With Globozoospermia And A Report Of A New DPY19L2 Mutation
P-1117	Derya Karaer	Clinical and Cytogenetic Presentation of Warkany Syndrome2: A Case of Mosaic Trisomy 8
P-1120	Igor Lebedev	X-Chromosome CNV reclassification: Integrating X-inactivation status for improved pathogenicity assessment
P-1127	Yarin Hadid	High-Level Trisomy 8 Mosaicism in a Healthy Adult: A Rare Case with No Clinical Manifestations
P-1132	Matilde Loja	Recurrent Absence of Heterozygosity (AOH) in the Portuguese Population: Clinical and Genomic Implications
P-1136	Niels Tommerup	Translocation of FIRRE as the critical event in X;autosomal translocations associated with premature ovarian insufficiency: A hypothesis.

POSTER LIST

Genomics

P-1014	Ivana Škrlec	Genetic and environmental factors in Hashimoto's thyroiditis: study on the association of MTNR1B gene polymorphisms with body mass index in a Croatian population
P-1032	Paola Evangelidou	A rare and complex case of a male patient with DiGeorge – like phenotype, carrying three different mosaic copy number variants on chromosome 22
P-1033	Elizabeta Ivanovska	HRAS related Costello Syndrome caused by HRAS c.34G>A p.Gly12Ser variation in child suspected with achondroplasia
P-1119	Damien Sanlaville	BARACUDA (B-Allele RAtio Chromosomal Uniparental Disomy and Aneuploidies): a visual tool to improve the detection and interpretation of mosaic CNVs and uniparental disomies in rare disease genomics
P-1130	Esmee ten Berk de Boer	investigating x chromosome inactivation patterns in x-autosome translocations using long-read sequencing and the T2T genome assembly
P-1140	Ester Margarit	16p11.2p12.1 duplication in a 47,XYY child

Other Cytogenomics topics

P-1013	Cláudia Oliveira	The DEB Test Beyond Fanconi anemia: A new look into chromosome instability
P-1026	Zuzanna Graczyk	Impact of sperm fractioning on chromosome positioning, chromatin integrity and DNA methylation level
P-1053	Jana Limbergova	Preliminary experience with AI-based karyotyping in peripheral blood specimens
P-1056	Attila Pintér	Developing artificial intelligence supported karyotyping software, KAYRA
P-1092	Lavinia Caba	Nijmegen Syndrome from a Founder Mutation: A Genetic Convergence Across Generations
P-1114	Cahide Kartal	Immunofluorescent visualization of H3K79me2 and H3K27me3 histone modifications in metaphase and PCC-obtained interphase chromosomes
P-1118	Damien SANLAVILLE	The BANCCO+ Project: A National Initiative to Improve Clinical Interpretation and Epidemiological Knowledge of CNVs in Neurodevelopmental Disorders and Fetal Anomalies

Prenatal Diagnosis

P-1020	Alla Vardanyan	Retrospective analysis of cytogenetic findings in pregnant women at risk following first-trimester screening: insights from nipt in armenia
P-1040	Luiza Dimos	the power of comprehensive genetic testing in one family case
P-1046	L.T. KWONG	Prenatal diagnosis of unusual chromosomal aberrations presenting with non-immune hydrops fetalis
P-1066	Anca Pavel	Whole-exome sequencing: a useful tool in prenatal diagnosis
P-1102	Cristina Ferreira	Genetic Analysis of Early and Recurrent Pregnancy Loss: Challenges and Advances

POSTER LIST

P-1125	Sarra Dimassi	Rare prenatal complex chromosomal rearrangement with 11q21-q22.1 deletion
P-1126	Nicolas Chatron	Comparative Efficacy of cfDNA and aCGH in Detecting Chromosomal Aberrations Post-Miscarriage
P-1128	Kévin Cassinari	First Prenatal Case of Jumping-like Translocations: Unraveling Complex Chromosomal Rearrangements
P-1134	Laura Coninx	Prenatal diagnosis of mosaic 45,X/47,XXX Turner syndrome in a patient with recurrent spontaneous abortions: a case report
P-1138	Maria Rosa Martorell Riera	CIRCUIT OF PRENATAL SCREENING WITH FREE CIRCULATING FETAL DNA IN THE BALEARIC ISLANDS

Tumour Cytogenomics

P-1004	Yit Jun Ng	Improvement of fluorescence in-situ hybridization (FISH) test results in formalin-fixed paraffin embedded (FFPE) tissues
P-1005	Milica Strnad	JAK2 mutations and endogenous erythroid colony formation in patients with polycythemia vera
P-1018	Seon Young Kim	Detection of measurable residual disease using fluorescence in situ hybridization compared with multiparametric flow cytometry in patients with B-lymphoblastic leukemia
P-1019	Hila Lederman Nachmias	New Insights Affecting Classification, Prognosis and Treatment of Multiple Myeloma Using Optical Genome Mapping
P-1022	Georgios Papanikolaou	The prognostic significance of isochromosome 17q in myeloid neoplasms
P-1023	Maria Gkaitatzi	T-ACUTE LYMPHOBLASTIC LEUKEMIA WITH TRANSLOCATION t(10;14)
P-1027	Dorina Roko	Prognostic biomarkers for Chronic Lymphocytic Leukemia patient's outcome in Albania
P-1029	Alexandra Ivanova	From manual to digital fluorescence in situ hybridization (FISH) – a time saving experience
P-1034	Elisavet Kouvidi	Evaluation of conventional and molecular cytogenetic analysis in plasma cell dyscrasias
P-1037	Uliana Lykhova	Beyond t(12;21): unveiling the hidden layers in all karyotypes
P-1039	Nataliia Levkovych	Genomic landscape of pediatric AML in Ukraine
P-1042	Nádia Neto	Unusual Cytogenetic Profile in Myelodysplastic Neoplasm: A Case Report of Hyperdiploidy with TRB Gene Involvement
P-1047	Yvoonne Yam Fang Tan	Oligosecretory multiple myeloma with low plasma cell count: a case study with etection of prognostic genomic abnormalities in metaphase cells
P-1050	Helena Podgornik	Evaluation of AI Suported Digital Karyotyping In Routine Analysis Of Hematological Malignancies
P-1055	Soumaya Mougou-Zerelli	Mapping Cancer Risk in Constitutional Chromosomal Deletions: A Cytogenetic Analysis

P-1062	Libuse Lizcova	Intra- and Intertumoral Heterogeneity in Glioblastomas Revealed by Optical Genome Mapping
P-1067	Iveta Mendlikova	Neocentromeres in complex AML karyotypes as an indicator of chromosomal instability and disease progression
P-1071	Sarka Ransdorfova	Optical genome mapping improves detection of cryptic aberrations in acute myeloid leukemia
P-1082	Marie-Bérengère Troadec	Optical genome mapping is a powerful tool for detecting clinically significant variants and chromosome abnormalities in hematological diseases
P-1083	Marie-Bérengère Troadec	What is wrong in the deletion of chromosome region 5q in myelodysplastic syndrome ? Identification of a novel actor of the sensitivity to lenalidomide of MDS with del(5q)
P-1085	Ulrike A. Mau-Holzmann	Multiple variable chromosomal aberrations in primary fibroblasts: further hints to chromosomal instability as a long-term effect - even years after irradiation
P-1086	Gulsim Smagulova	Morphological features and mutational status of the FGFR gene in urothelial carcinoma
P-1087	Marija Dencic Fekete	Distribution of gene aberrations in chronic lymphocytic leukemia by ngs testing in a serbian patients cohort
P-1096	Radhia M'kacher	Telomere dysfunction, DNA breaks, chromosomal aberration formation, and the dark side of the centromere
P-1098	Syeda Zehra	The enigma of KMT2A gene in hematological disorders
P-1099	Barbara Dewaele	Optimization of the routine diagnostic workflow for Myeloid/Lymphoid Neoplasms with eosinophilia and Tyrosine Kinase gene fusions (MLN-TK) by optical genome mapping
P-1104	Laura Yissel Rengifo	dynamic follow-up of tumor burden in multiple myeloma through analysis of ccfdna markers
P-1106	Beyhan Durak Aras	What does atypical philadelphia fish signal pattern indicate in cml?
P-1108	Amber Verhasselt	Optical genome mapping is a powerful diagnostic tool in non-hodgkin lymphoma
P-1113	Wiem Ayed	Complex Karyotype Prognosis of Acute Myeloid Leukemia in Tunisian Patients
P-1115	Catherine Menten	Improved detection of cytogenetic abnormalities in Multiple Myeloma (MM) using Optical Genome Mapping (OGM)
P-1116	Yelda Tarkan Argüden	Immunofluorescent visualization of H3K27ac and H3K27me3 histone modifications in normal and cancer chromosomes
P-1121	Ilda P Ribeiro	Glioblastoma Cytogenomics: Patterns & Insights
P-1123	Isabel M Carreira	Rectal Adenocarcinoma: cytogenomic alterations and their implications in disease progression and treatment response
P-1133	Paola Caria	Linking genetic predisposition to telomere disfunction in familial non medullary thyroid cancer
P-1135	Tadeusz Kaluzewski	Evaluation of the Utility of TERT Promoter Mutations in the Early Detection of Urothelial Cancer
P-1139	Árpád Szomor	Occurence of myeloid and lymphoid disorders in a Hungarian family

SPONSORED WORKSHOPS & SESSIONS

PRE-CONGRESS WORKSHOPS

Analysis of clinical and haemotological samples, in group of three (the computer connects directly to the Bionano US server in US)
5th Floor (1 floor up), ALO 05.100

Bridging cytogenetics workflows: Hands-on analysis of arrays to NGS
5th Floor (1 floor up), ALO 05.200

The use of online resources to annotate/map clinical CNVs
7th Floor (3 floors up), ALO 07.100

Nanopore based sequencing for structural variant detection, from sample to report
Wet Lab: Laboratory (Meeting point Registration desk)
Dry Lab: 7th Floor (3 floors up), ALO 07.200

SPONSORED SESSIONS

Next-gen cytogenetics: Applications of AI and digital FISH in diagnostics
June 30th, 12:30-14:30 / 5th Floor (1 floor up), ALO 05.100

Addressing clinical challenges in rare diseases through long-range insights by a novel genome sequencing technology
July 1st, 12:30-13:15 / 4th Floor (Ground), Auditorium GA1

European Cytogeneticists Association

15th EUROPEAN CYTOGENOMICS CONFERENCE

June 29-July 1, 2025

LEUVEN, BELGIUM

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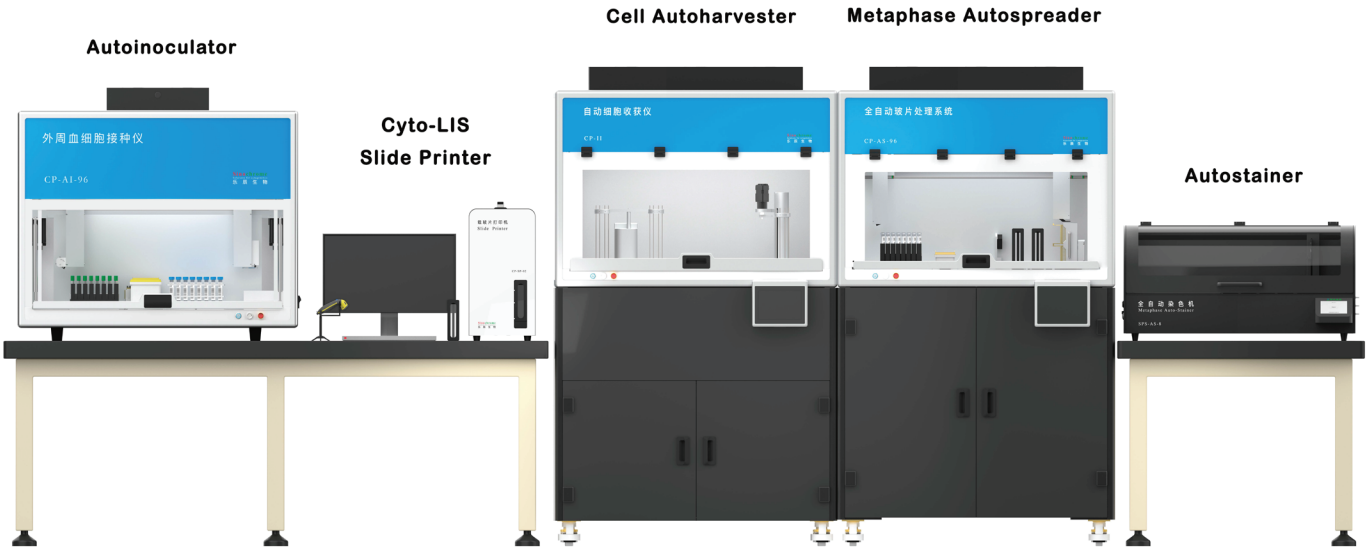

Inoculation → Cell Culture → Harvesting → Spreading → Banding

The sample preparation for cytogenetic test involves a series of sequential steps: sample inoculation, cell culture, cell harvesting, metaphase spreading and G-banding. Numerous factors can influence both the chromosome preparation process and the quality of metaphase. Sinochrome provides a perfect solution for the complex sample preparation process, which require proficient skills and meticulous operation. This comprehensive solution is composed of Autoinoculator, Cell Autoharvester, Metaphase Spreader, Autostainer, and Cytogenegtic Lab Information System. By integrating these components, Sinochrome's solutions achieve full automation, standardization, informatization of sample processing. There are 100+ well-known users of Sinochrome's solution.

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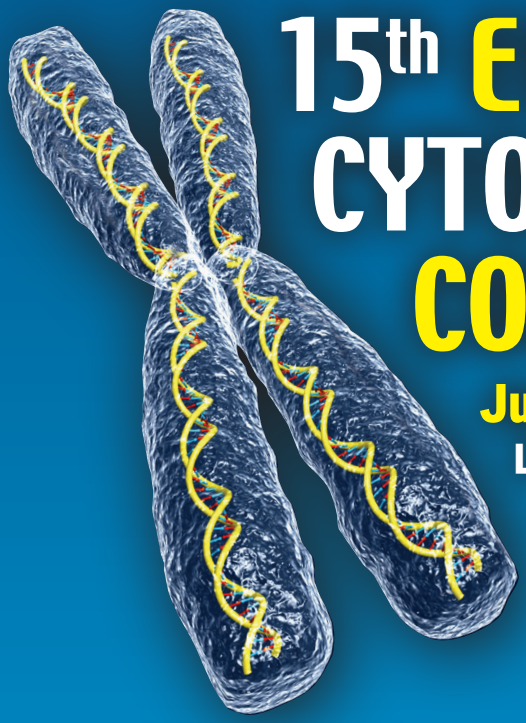

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