


European Cytogeneticists Association

14th EUROPEAN CYTOGENOMICS CONFERENCE

1 - 4 July 2023

MONTPELLIER - FRANCE

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PROGRAMME BOOK

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SCIENTIFIC PROGRAMME

14th European Cytogenomics Conference – ECA 2023 has been granted 16 European CME credits (ECMEC®s) by the European Accreditation Council for Continuing Medical Education (EACCME®).

1 JULY 2023, SATURDAY

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 Introduction and news about the update of ISO 15189
Martine Doco-Fenzy

14:45 External quality assessments, contribution to quality improvement
Melody Tabiner

15:15 Genome Mapping (optical and electronic) nomenclature and ISCN 2024 (P1003)
Ros Hastings

15:45 Training on ISCN: Quiz
Jean-Michel Dupont

16:15 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)
Marie Bérengère Troadec

16:30 **Closing**

14:30-16:30 **PWG NEOPLASIA** **Room Sully 2**
Coordinators: *Paola Caria, Harald Rieder, Roberta Vanni*

14:35 Donor cell acute myeloid leukaemia after haematopoietic stem cell transplantation for chronic granulomatous disease (P1067)
Francesco Pasquali

14:48 Ring chromosomes in hematological malignancies are mainly associated with myeloid malignancies and complex karyotypes (P1023)
Kalliopi Manola

15:01 Cytogenetic groups of pediatric acute myeloid leukemia from Ukraine (P1120)
Uliana Karnaukhova

15:14 Validation of the OGM for cytogenomic testing in hemato-oncology – Sheba Medical Center experience (P1052)
Victoria Marcu

15:27 Laboratory Validation and Clinical Implementation of an RNA sequencing-Based Prognostic Assay for Multiple Myeloma (P1155)
Bob Argiropoulos

15:40 **Break**

15:50 Mutation of the PIK3CA gene in breast cancer (P1069)
Gulsim Smagulova

16:03 Detection of promoter methylation as well as deletion of MGMT gene in patients with glioblastoma using methodologically different approaches (P1097)
Halka Lhotska

16:16 Report on the implementation of an early cancer identification and prevention program among the population of central Poland (P1128)
Tadeusz Kałużewski

16:30 **Closing**

14:30-15:30 **PWG PRENATAL DIAGNOSIS** **Room Sully 3**
Coordinators: *Rosário Carvalho Pinto Leite, Jean-Michel Dupont*

14:30 Presentation of the guidelines of Microarrays in Prenatal Diagnosis
Jean-Michel Dupont

14:40 Presentation of the results of the survey
Rosário Carvalho Pinto Leite

14:50 The ever-changing face of Cytogenetics Units: Use and contribution of Whole Exome Sequencing in prenatal diagnosis (P1029)
Celine Dupont

15:05 A larger European network about cancers in pregnancy
Joris Vermeesch

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 Optical Genome Mapping: Comparing OGM with other Cytogenomics technologies. Experience on 60 individuals with developmental or fertility disorders (P1032)
Vasheghani Farahani Faezeh

15:42 Primary Ovarian Insufficiency: don't neglect intragenic CNVs (P1062)
Anna Lokchine

15:54 Different strategies for the detection of copy-number variations from exome sequencing data (P1037)
Vladimíra Vallová

16:06 Breastfeeding promotes persistence of the mother's chimeric cells in their offspring (P1157)
Vincent Gatinois

16:18 Streamlining cytogenetics analysis of genome sequencing data: a comprehensive guide for Balanced Structural Variants (P1139)
Nicolas Chatron

16:30 Copy number detection in exome sequencing data for patients with neurodevelopmental disorders: an effective approach (P1154)
Annelies Dheedene

16:42 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)
Leona Morožin Pohovski

17:04 Expanding the phenotype of 14q11.2 microdeletions encompassing CHD8 and SUPT16H genes (P1065)
Anna Lengyel

17:30 **Closing**

15:30-17:30 **PWG ANIMAL, PLANT, AND COMPARATIVE CYTOGENETICS** **Room Sully 3**
Coordinators: *Pat Heslop-Harrison, Trude Schwarzacher*

15:30 Introduction
Valérie Fillon, J.S. (Pat) Heslop-Harrison

15:40 Gametogenesis in hybridogenetic frogs – tracking cellular events of genome elimination and endoreduplication (P1012)
Magdalena Chmielewska

15:55 Cytogenetics of the hybrid frog Pelophylax grafi and its parental species Pelophylax perezi (P1021)
Anna Dudzik

16:10 CAP-A satellite DNAs probe mapping on Sapajus cay paraguay and S.macrocephalus by FISH (Platyrrhini, Primates) (P1035)
Francesca Dumas

16:25 Chromosomal evolution and genome expansion in diploid oats
Trude Schwarzacher

16:40 Cytogenetic screening of Romanian bovine breeds (P1098)
Ioana Ncolae

16:55 A glimpse of the karyotype reshuffling from human to Myotis blythii (Vespertillionidae, Chiroptera) (P1125)
Verónica Mestre

17:10 Evolution of gametogenic pathways in reproduction of hybrid males from Pelophylax esculentus complex (P1044)
Eleonora Pustovalova

17:25 Summary/general discussion

17:30 **Closing**

16:30-17:30 **PWG CHROMOSOMES' INTEGRITY, STABILITY AND DYNAMICS** **Room Barthez**
Coordinators: *Jose Garcia-Sagredo, Emanuela Volpi*

16:40 Molecular Mapping of Two Replication Stress-Induced Hotspots of Breakage at the Common Fragile Site FRA11D Harboring Cancer and Neurological Genes (P1068)
Eliane El Achkar

16:48 Chromosomal Instability in Mesenchymal Stromal Cells From Acute Myeloid Leukemia Patients (P1057)
Mateus de Oliveira Lisboa

16:56 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)
Anna Schachner

17:04 Cytogenetic analysis of induced pluripotent stem cell (iPSC) cultures derived from dermal fibroblasts (1046)
Nicoletta Selenti

17:12 Screening of biomarkers for chromosomal instability in the cytogenetic clinic: Present status on technological advances and their implementation into routine screening programs (P1151)
Radhia M'Kacher

17:30 **Closing**

16:30-17:30 **PWG MARKER CHROMOSOMES** **Room Sully 2**
Coordinators: *Thomas Liehr, Isabel Marques-Carreira*

16:30 Introdution on Small Supernumerary Marker Chromosomes (sSMC)
Thomas Liehr

16:40 Optical Genome Mapping (OGM): Validation and characterization of marker chromosomes (P1095)
Christina Pérez

16:50 Importance of arrayCGH for sSMC detection and characterization
Joana Melot

17:00 Classical genetic techniques are still in use: a case with low mosaicism (P1010)
Esther Cuatrecasas

17:10 The surprises that can come with the characterization of a sSMC
Isabel Carreira

17:30 **Closing**

18:00 – 19:00 **CONFERENCE OPENING LECTURE** **Room Pasteur**
Chair: *Mariano Rocchi and Franck Pellestor*

Aneuploidy in the Maternal Germline. ***Eva R. Hoffmann***

2 JULY 2023, SUNDAY

8.30- 10.15 PLENARY SESSION 1: Mosaicism: from Preimplantation Embryos to Aging **Room Pasteur**

Chairs: *Joris Vermeesch and Elisabeth Syk Lundberg*

8:30-9:00 Mosaicism in Preimplantation Embryos
Antonio Capalbo

9:00-9:30 Mosaicism in Prenatal Diagnosis: from NIPT to Amniocytes Investigation
Malgorzata I. Srebniak

9:30-10:00 Hematopietic Loss of Chromosome Y and Higher Mortality in Men
Lars A. Forsberg

Selected Abstract

10:00-10:15 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
Cornelia Daumer-Haas

10:15-10:45 **Coffee break**

10:45-12:00 PLENARY SESSION 2: Cancer Cytogenomics **Room Pasteur**
Chairs: *Felix Mitelman and Roberta Vanni*

10:45-11:15 Replication Stress Generates Distinctive Landscapes of DNA Copy Number Alterations and Chromosome Scale Losses in Cancer
Sarah McClelland

11:15-11:45 Whole-Genome Duplication Shapes the Aneuploidy Landscape of Human Cancers
Uri Ben-David

Selected Abstract

11:45-12:00 Optical Genome Mapping for Multiple Myeloma: Evaluation of The Technology in a Clinical Laboratory.
Christina Srouji

12:00-12:45 **MetaSystems Satellite Symposium** **Room Pasteur**
Intelligent Case Management – A Look at Three Scenarios
Pradipta Mandal

12:45-14:30 **POSTER SESSION AND LUNCH BREAK**

14:30-15:45 **CONCURRENT SESSION 1: Recent Advances in Cytogenomics** **Room Pasteur**
Chairs: *Franck Pellestor and Harald Rieder*

14:30-15:00 Optical Mapping to Karyotype
Alex Hoischen

15:00-15:30 Artificial Intelligence in Cytogenetics
Antonio Rausell

Selected Abstract

15:30-15:45 Dam Assisted Fluorescent Tagging of Chromatin Accessibility (DAFCA) for Optical Genome Mapping in Nano-Channel Arrays
Gil Nifker

14:30-15:45 **CONCURRENT SESSION 2: Beyond Genome Sequencing: the Epigenetic Signature** **Room Antigone 3**
Chairs: *O. Zuffardi and Joan Blanco*

14:30-15:00 DNA Methylation Episignatures Associated with Large Structural Copy Number Variants: Clinical Implications
Bekim Sadikovic

15:00-15:30 Multi-locus imprinting disorders
Karen Temple

Selected Abstract

15:30-15:45 Long Read Whole Genome Sequencing for The Detection of Structural and Epigenetic Variation in Developmental Disorders
Mathilde Geysens

15.45-16.15 **Coffee break**

16:15-17:30 **PLENARY SESSION 3: Newly Emerged Technologies in Cytogenomics** **Room Pasteur**
Chairs: *Pat Heslop-Harrison and Emanuela Volpi*

16:15-16:45 Paint-SHOP; Genome-Scale Oligonucleotide FISH Experiments
Brian Beliveau

16:45-17:30 Interactive Discussion; Massive Oligonucleotide Pools to Track Organization and Evolution of Chromosomes and Genomes
Pat Heslop-Harrison and Emanuela Volpi

17:30-18:30 **POSTER SESSION**

3 JULY 2023, MONDAY

8:30-10:30 **PLENARY SESSION 4: Clinical Cytogenomics I** **Room Pasteur**
Chairs: *Damien Sanlaville and José Garcia-Sagredo*

8:30-9:00 Complex Genomic Rearrangements: an Underestimated Cause of Rare Diseases
Anna Lindstrand

9:00-9:30 Distal Germ-Line Deletions in Mosaic With Copy-Neutral Loss of Heterozygosity: Something to Be Considered in Genetic Counselling
Orsetta Zuffardi

9:30-10:00 From Gene Disruption to Missense Variants: how Different Types of Variants Influence the X-Linked Inheritance Model
Brunella Franco

Selected Abstracts

10:00-10:15 Burden of Long Range Position Effects in Balanced Chromosomal Rearrangements
Niels Tommerup

10:15-10:30 Multiomic Profiling Unravels Disease Mechanisms in Complex Chromosomal Rearrangements and Marker Chromosome Carriers
Marlene Ek

10:30-11:00 **Coffee break**

11:00-12:15 **CONCURRENT SESSION 3: Clinical Cytogenomics II** **Room Pasteur**
Chairs: *Orsetta Zuffardi and Martine Doco-Fenzy*

11:00-11:30 Structural Variants in Clinical Practice Using Genome Sequencing
Nicolas Chatron

11:30-12:00 Constitutional Chromoanagenesis: From Diagnosis to Genetic Counselling
Caroline Schluth-Bolard

Selected Abstract

12:00-12:15 Systematic X-Inactivation Studies of Sequence Resolved Balanced X Chromosomal Rearrangements
Sanam Khan

11:00-12:15 **CONCURRENT SESSION 4: Animal and Plant Cytogenomics I** **Room Antigone 3**
Chairs: *Tony Heitkam and Trude Schwarzacher*

11:00-11:30 Comparative Genomics and Tools for Studying Chromosome Evolution
Mathieu Rouard

11:30-12:00 Coleopteran Satellite Profiles: Chromosomal and Sequence Organization
Brankica Mravinac

Selected Abstract

12:00-12:15 A physical map of repetitive elements in the genomes of Iberian Peninsula chiropteran species
Maria Filomena Lopes Adega

12:15-13:00 **Bionano Satellite Symposium** **Room Pasteur**
Revolutionizing Cytogenomics with Optical Genome Mapping sample-to-answer workflow: Now powered by VIA software
"Introduction to Bionano sample-to-answer workflow" - ***Dana Jaber (Bionano)***
"OGM for Hematological Malignancies - Sheba Experience" - ***Victoria Marcu (Sheba Medical Center, Israel)***
"OGM contribution to the Etiological Diagnosis of Developmental disorders, experience of CHU Nantes Genetic Department" - ***Martine Doco-Fenzy (CHU de Nantes, France)***

13:00-14:30 **POSTER SESSION AND LUNCH BREAK**

14:30 - 15:45 **PLENARY SESSION 5: Nuclear Organization and Diseases** **Room Pasteur**
Chairs: *Jean-Michel Dupont and Emanuela Volpi*

14:30-14:55 The 3D genome organization into TADs and chromatin nanodomains
Frédéric Bantignies

15:00-15:30 Spatial Organization of Transcribed Eukaryotic Genes
Irina Solovei

Selected Abstract

15:30-15:45 3D Nuclear Architecture Distinguishes Thyroid Cancer Histotypes
Paola Caria

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

16:15-17:30 **CONCURRENT SESSION 5: Animal and Plant Cytogenomics II** **Room Pasteur**
Chairs: *Trude Schwarzacher and Brankica Mravinac*

16:15-16:45 Adding a Chromosome Perspective to Plant Genomics: Making Sense of Retained Retroviruses, Moving Retrotransposons and Expanding Satellite DNAs
Tony Heitkam

16:45-17:15 Plasticity in centromere organization: A few megabased-sized centromere units can form a holocentromere
Yi Tzu Kuo

Selected Abstract

17:15-17:30 The first nuclear and cytoplasmic whole transcriptome profile of chicken oocytes at the lampbrush chromosome stage
Alla Krasikova

16:15-17:30 **CONCURRENT SESSION 6: Accreditation, Quality Control and Education** **Room Antigone 3**
Chairs: *Konstantin Miller and Martine Doco-Fenzy*

16:15-16:45 The New ISO 15189 Standard Medical Laboratories
Folker Spitzenberger

16:45-17:15 Sequence-based Nomenclature and the Novelties to Come in the Next ISCN Version
Johan den Dunnen

Selected Abstract

17:15-17:30 Educational Benefits of Analysing Highly Complex Chromosomal Rearrangements Such as Chromoanagenesis by Long Read Approaches
Mathilde Quibeuf

17:30-18:30 **POSTER SESSION**

4 JULY 2023, TUESDAY

8:30-10:30 **PLENARY SESSION 6: Prenatal Diagnosis and Preimplantation** **Room Pasteur**
Chairs: *Jean-Michel Dupont and Rosário Pinto Leite*

8:30-9:00 Genome-Wide Noninvasive Prenatal Testing: Follow-Up Results of the TRIDENT-2 Study
Robert-Jan H. Galjaard

9:00-9:30 Fragmentomics and Non Invasive Prenatal Screening (NIPS)
Joris Vermeesch

9:30-10:00 Prenatal Diagnostic Yield and Pitfalls Through Arrays, Exome, and NIPT
Lyn Chitty

Selected Abstracts

10:00-10.15 Multicentric Longitudinal Performance Monitoring of Different non-Invasive Prenatal Screening Technologies Used in Belgium
Armelle Duquenne

10:15-10.30 Triploid Conceptions Are Predominantly Caused by Female Meiosis II Errors and Their Risk Increases with Advancing Maternal Age
Ludovica Picchetta

10:30-11.00 **Coffee break**

11:00-11:50 **CLOSING KEYNOTE**
Chairs: *Mariano Rocchi and Thierry Lavabre-Bertrand*

The Landscape of Structural Variation Across Diverse Global Populations and Developmental Disorders.
Michael E.Talkowski

12:00 **CLOSING CEREMONY**

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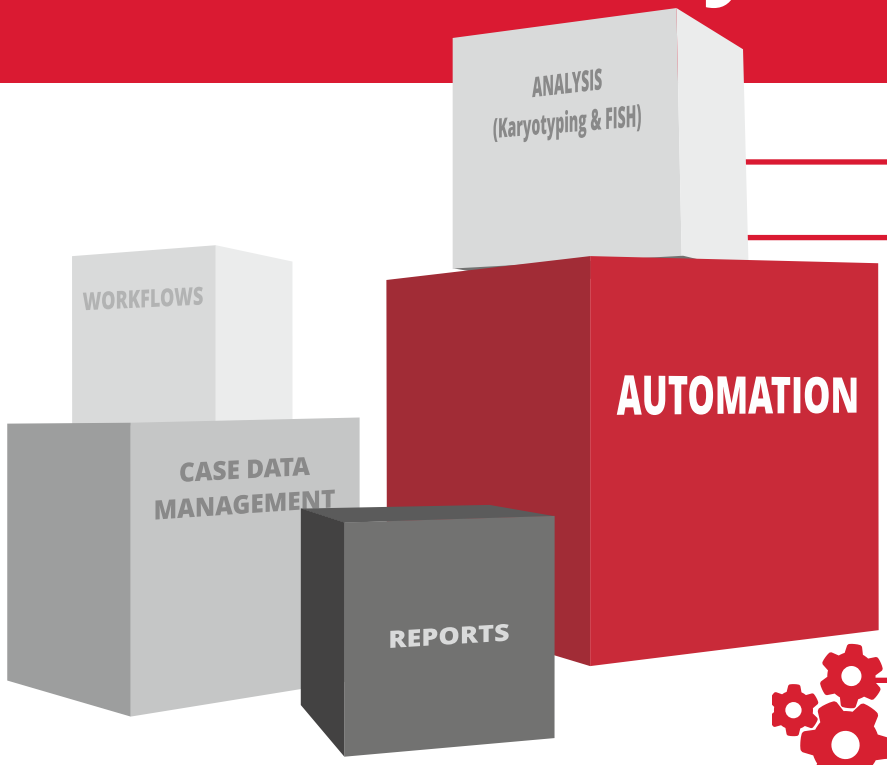

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POSTERS

1. Accreditation Quality Control Education

P1003 Genome Mapping (optical and electronic) nomenclature and ISCN 2024
Ros Hastings

P1032 Optical Genome Mapping Comparing OGM with other Cytogenomics technologies. Experience on 60 individuals with developmental or fertility disorders.
Doco-Fenzy Martine

P1063 Reciprocal translocations and 3:1 segregation reminder to think of potential effects concerning viability or UPD
Ulrike A. Mau-Holzmann

2. Animal and Plant Cytogenomics

P1012 Gametogenesis in hybridogenetic frogs – tracking cellular events of genome elimination and endoreduplication
Magdalena Chmielewska

P1021 Cytogenetics of the hybrid frog Pelophylax grafi and its parental species Pelophylax perezi
Anna Dudzik

P1028 Drive of chromosomes and programmed chromosome elimination – different sites of the same coin
Andreas Houben

P1035 CAP A satellite DNAs probe mapping on Sapajus cay paraguay and S. macrocephalus by FISH (Platyrrhini Primates)
Francesca Dumas

P1044 Evolution of gametogenic pathways in reproduction of hybrid males from Pelophylax esculentus complex
Eleonora Pustovalova

P1098 Cytogenetic screening of Romanian bovine breeds
Ioana Nicolae

P1125 A glimpse of the karyotype reshuffling from human to Myotis blythii (Vespertillionidae Chiroptera)
Verónica F. Mestre

3. Clinical Cytogenomics

P1002 A very rare double chromosome 9 mosaicism a case report
Andreja Zagorac

P1007 the finnish national collection of balanced translocations and inversions facilitates gene mapping
Teppo Varilo

P1008 Genetic hearing loss screening by MLPA in a cohort of Portuguese patients
Cristina Candeias

P1009 Chromosomal abnormalities in donor gamete candidates in a Public Bank: retrospective analysis 2013-2022
Manuela Mota Freitas

P1010 CLASSICAL GENETIC TECHNIQUES ARE STILL IN USE a case with low mosaicism
Esther Cuatrecasas

P1016 CACNA1B GENE AND AUTISM CORRELATION A CASE REPORT
Silvia Motta

P1026 Balanced complex chromosomal rearrangement of chromosome 2 in an infertile male
Tea Mladenić

P1030 Chromosomal abnormalities in male partners of infertile couples
Elisavet Kouvidi

P1037 Different strategies for the detection of copy number variations from exome sequencing data
Vladimíra Vallová

P1041 A case report of the interstitial duplication the short arm of chromosome 7
Valentyna Kurakova

P1043 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
Leona Morožin Pohovski

P1046 Cytogenetic analysis of induced pluripotent stem cell (iPSC) cultures derived from dermal fibroblasts
Nikoletta Selenti

P1048 Diagnosis in Emanuel syndrome a challenge for geneticists
Eusebiu Vlad Gorduza

P1049 familial case of X;9 translocation
Natalia Oparina

P1050 Cytogenetic and molecular profile of azoospermia in
Algeria Anissa Nini

P1054 Cytogenetic and molecular characterization of a mosaic ring chromosome 13 mechanism of formation and instability
Mar Xunclà

P1055 Currarino syndrome in two Moroccan siblings with inherited 7q36 deletion due to maternal t(7;21)(q36;p11) a case report
Zhour El Amrani

P1056 Prader Willi syndrome as a result of a de novo unbalanced translocation 15q;19p
Jasenka Wagner

P1062 Primary Ovarian Insufficiency don't neglect intragenic CNVs
Anna Lokchine

P1064 Study of CNVs small rearrangements by CGH array on dedicated oligonucleotide chip in the setting of molecular diagnosis of Marfan syndrome related syndromes and non syndromic hereditary thoracic aortic aneurysms dissections (hTAAD).
Thierry Lavabre-Bertrand

P1065 Expanding the phenotype of 14q11.2 microdeletions encompassing CHD8 and SUPT16H genes
Anna Lengyel

P1071 Interesting case studies in patients suffering from anorectal malformations
Drahuse Novotna

P1072 Synergy of various diagnostic methods in MCADD Slovenian patient
Maja Ficko

P1078 A case report of a child with 46 XX del(5)(q21.1q31.1) derived from a maternal insertion 46 XX ins(14;5)(q24.3;q21.1q31.1)
Tiiu Roovere

P1079 De novo intrachromosomal insertion with 1q32.1q32.2 duplication in a patient with neurodevelopmental delay
Isaltina Silva

P1080 Abnormal features of DiGeorge syndrome
Tereza Kutilova

P1085 A case of mosaicism with a complex unbalanced reciprocal translocation and a normal cell line in a male patient with fertility problem. A challenging cytogenomic diagnosis.
Eva-Maria Krimmel

P1088 Intrachromosomal insertion or paracentric inversion A classic trap in chromosome analysis
Samira Ahmed-Elie

P1089 Optical mapping characterization of a very complex chromosomal insertion
Alberto Plaja

P1092 Rare genomic imbalances encompassing kinase genes in a group of patients with autism spectrum disorders
Magdalena Budisteanu

P1095 Optical Genome Mapping (OGM) Validation and characterization of marker chromosomes
Cristina Pérez

P1096 Detection of 12;13 reciprocal translocation with copy number losses detected by karyotype and array and characterized by Optical Genome Mapping.
Elisabet Lloveras

P1100 ICF Syndrome diagnosis conventional cytogenetics could be the key to disclose the syndrome
Eunice Matoso

P1103 Recurring phenotype in a family A coincidence or not
Eva Pinti

P1109 Complete paternal isodisomy of chromosome 15 in a patient with atypical presentation of Angelman syndrome
Oğuz Çilingir

P1111 A de novo small marker chromosome that causes Trisomy 9p in a patient with failure to develop, microcephaly and normal neuromotor development
Sevilhan Artan

P1121 Molecular confirmation of PHEX related hypophosphatemic rickets in a nine year old girl
Anette Eek

P1122 Ring chromosome 13 and translocation of 13q31.1 qter to 21p12 in a healthy female with medical history of eye cancer
Sylke Singer

P1126 Assessing variants of uncertain significance a retrospective analysis of aCGH cases
Inês Costa

P1132 Cytogenetic and flow cytometric findings in skin fibroblasts of patient with FANCI subtype of Fanconi anemia
Anna Repczynska

P1137 Investigation of Genetic Etiology of Short Stature
Birsen Karaman

P1142 Challenges detecting a MAGEL2 in frame deletion variant two clinically distinct families and sequencing issues
Sarah Delbaere

P1146 Interstitial 11q deletion in a patient with Sprengel's deformity case report and review of literature
Dhekra Ismail

P1154 Copy number detection in exome sequencing data for patients with neurodevelopmental disorders an effective approach
Dheedene Annelies

4. Genomics

P1005 Improvement of STR based approaches in the analysis of the genetic composition of the Tunisian population and its application in forensic identification.
Ali Saad

P1006 Case report demonstrating certain pitfalls and challenges in NGS data interpretation
Paola Evangelidou

P1106 Functional Characterization of a MLH1 missense variant identified in a Tunisian Turcot syndrome patient
Marwa Mahdouani

P1113 Molecular characterization of Porokeratosis in Tunisian patients
Haifa El Mabrouk

P1116 Telomere dysfunction leads to chromosomal aberrations in patients with disorders of sexual development
Haifaou Younoussa

P1138 Switching on sex Genetic findings in a Tunisian cohort with Disorders of sexual development (DSD)
Khouloud Rjiba

P1148 Somatic and Germline Variants in Ovarian Cancer Patients from a Unique Geographically Isolated Population A Comprehensive Molecular Analysis
Federica Cannas

P1154 Copy number detection in exome sequencing data for patients with neurodevelopmental disorders an effective approach
Dheedene Annelies

P1157 Breastfeeding promotes persistence of the mother's chimeric cells in their offspring
Vincent Gatinois

5. Prenatal Diagnosis

P1013 One Laboratory's Experience – Concordance between NIPT Karyotyping FISH and Prenatal CMA for Diagnosing Chromosomal Anomalies
Xin Yi Boo

P1029 The ever changing face of Cytogenetics Units Use and contribution of Whole Exome Sequencing in prenatal diagnosis.
Celine Dupont

P1031 A role of cytogenetic methods in prenatal diagnostics (case study)
Helena Peková

P1034 Prenatal diagnosis of Beckwith Wiedemann syndrome a case report
Cristina Ferreira

P1042 Inherited unbalanced reciprocal translocation with 18p tetrasomy and 9q34.3 trisomy in a foetus revealed by cell free foetal DNA (cffDNA) testing cytogenetic and cytogenomic characterization in prenatal diagnosis
Luigia De Falco

P1066 Anticipation of Sex Discrepancies in non invasive Prenatal Testing Due To Maternal Genetic Abnormalities
Nuria Balaguer

P1083 Prenatal diagnosis of a 15q24.1 microdeletion in a fetus with cerebral and urogenital abnormalities
Anaik Previdi

P1093 Expression of the syncytin 1 and syncytin 2 genes in the trophoblastic tissue of the early pregnancy losses with normal and abnormal karyotypes
Esra Tug

P1127 Have the objectives been met in the screening for aneuploidies in TPNI
Rosa Maria Lobo

P1133 Deletion of Exon 16 of the COL2A1 Gene in Prenatal Spondylo-Epiphyseal Dysplasia
Maria Rosa Martorell Riera

P1136 Significance of chromosome 18p duplication in prenatal diagnosis
Chariyawan Charalsawadi

P1143 Prenatal DiagnosisIAGNOSIS IN A CARRIER OF AN UNSOLVED CRYPTIC TRANSLOCATION
Ester Margarit

P1145 A challenge in prenatal diagnosis clinic significance of a CNV in DMD gene (Incidental finding)
Neus Castells-Sarret

P1152 UPD in the Prenatal setting The Benefit of Integration of Methods
Ayala Frumkin

6. Tumour Cytogenomics

P1004 The importance of "Complex Karyotype" (CK) diagnosis in the Chronic Lymphocytic Leukemia (CLL) patients with normal TP53 FISH (Fluorescence in situ hybridization) results
Mira Ziv

P1018 The role of cytogenetic analysis in patients receiving CAR T cell therapy
Anastasia Athanasiadou

P1019 Additional cytogenetic abnormalities in patients with core binding factor AML. Experience of a center.
Giorgos Papaioanno

P1020 Common ALL and pre B ALL with t(8;14) and t(14;18)
Maria Gkaitatzi

P1022 Optical Genome Mapping in Routine Cytogenetic Diagnosis of Acute Leukemia
Gwendoline Soler

P1023 Ring chromosomes in hematological malignancies are mainly associated with myeloid malignancies and complex karyotypes
Kalliopi Manola

P1024 Optical genome mapping in Leukemia demonstrates full concordance and new cytogenetic findings in an Israeli cohort
Nivin Moustafa-Hawash

P1027 The importance of using FISH (Fluorescent In Situ Hybridization) technology as an investigative tool in patients with malignant hemopathies in Albania
Dorina Roko

P1033 Cytogenetic profile of Core Binding Factor Acute Myeloid Leukemia in Tunisian patients
Wiem Ayed

P1039 Presence of JAK2 V617F MPL mutations and cytogenetic aberrations in patients with thrombocytosis
Milica Strnad

P1045 Chromosome 1 abnormalities in Childhood B Lymphoblastic Leukemia – An analysis with respect to clinical variables and survival outcome
Neelum Mansoor

P1051 Additional chromosomal abnormalites in Philadelphia chromosome positive chronic myeloid leukemia. Single center experience.
Arpad Szomor

P1052 Validation of the OGM for cytogenomic testing in hemato oncology – Sheba Medical Center experience.
Victoria Marcu

P1059 T cell receptor (TCR) loci rearrangements in children with T cell acute lymphoblastic leukemia
Libuse Lizcova

P1060 Optical genome mapping of bone-marrow in Hematological malignancies
Hila Nachmias

P1061 Newly detected TP53 gene deletion in repeatedly examined patients with multiple myeloma (MM)
Lenka Pavlistova

P1067 Donor cell acute myeloid leukaemia after haematopoietic stem cell transplantation for chronic granulomatous disease
Francesco Pasquali

P1069 mutation of the PIK3CA gene in breast cancer.
Gulsim Smagulova

P1075 Optical Genome Mapping use in the multiple myeloma diagnosis
Catherine Menten

P1077 NPM1(+) Acute Myeloid Leukemia with an abnormal karyotype
Chrysavgi Lalayanni

P1081 Hematological disorders in patients with constitutional chromosomal abnormality
Marie Valerianova

P1082 Significance of chromosome 7 aberrations in myeloid malignancies
Sarka Ransdorfova

P1087 In multiple myeloma a clear distinction between gain(1q) and amp(1q) is required
Ana Doplihar Kebe

P1097 Detection of promoter methylation as well as deletion of MGMT gene in patients with glioblastoma using methodologically different approaches
Halka Lhotska

P1107 Cytogenetic analysis in acute myeloid leukemia during the 2012 2022 period in Republic of Srpska Bosnia and Hercegovina
Marija Vukovic

P1108 Three paediatric cases of monosomy 7 with different further course of the disease
Pille Tammur

P1112 Impact of additional cytogenetics abnormalities in young chronic myeloid leukemia patients single centre experience.
Mehwish Imam Khushk

P1115 The role of variant t(9;22) in Chronic Myeloid Leukemia A report of 5 cases
Beyhan Durak Aras

P1117 Glioma polyploid stem cells are sensitive to the treatments of aurora kinase inhibitor
Martina Giambra

P1120 Cytogenetic groups of pediatric acute myeloid leukemia from Ukraine
Uliana Karnaukhova

P1124 Glioblastoma from cytogenomics and methylation profile to liquid biopsies and Biomarker Identification
Isabel M Carreira

P1128 Report on the implementation of an early cancer identification and prevention programme among the population of the central Poland
Tadeusz Kałużewski

P1129 SNP Array characterization of acute lymphoblastic leukemia samples
Joana Barbosa De Melo

P1140 Complex translocation leading to13q interstitial deletion in a Moroccan child with retinoblastoma and intellectual disability
Wafaa Jdioui

P1141 Genomic status of PTEN gene and AR expression in primary prostate carcinoma in a cohort of patients from Dobrogea
Georgeta Camelia Cozaru

P1155 Laboratory Validation and Clinical Implementation of an RNA sequencing Based Prognostic Assay for Multiple Myeloma
Bob Argiropoulos

P1156 Trisomy 14 a rare event in acute myeloid leukemia
Rosário Pinto Leite

P1053 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.
Mariebérengère Troadec

7. Other Cytogenomic Topics

P1057 Chromosomal Instability in Mesenchymal Stromal Cells From Acute Myeloid Leukemia Patients
Mateus De Oliveira Lisboa

P1068 Molecular Mapping of Two Replication Stress Induced Hotspots of Breakage at the Common Fragile Site FRA11D Harboring Cancer and Neurological Genes
Eliane El Achkar

P1074 CNV Hub a computational tool to classify Copy Number Variations of Unknown Significance using an artificial intelligence based method
Vignesh Guru Pillay

P1101 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
Anna Schachner

P1110 Placental activator and inhibitor miRNAs efficiency on abortion development; an epigenetic alteration on true fetal tissues
Meral Yirmibes Karaoguz

P1147 Clinical Impact of RNA sequencing in Diagnostics
Erika D'haenens

P1151 Screening of biomarkers for chromosomal instability in the cytogenetic clinic Presentstatus on technological advances and their implementation into routine screening programmes.
Radhia M'Kacher

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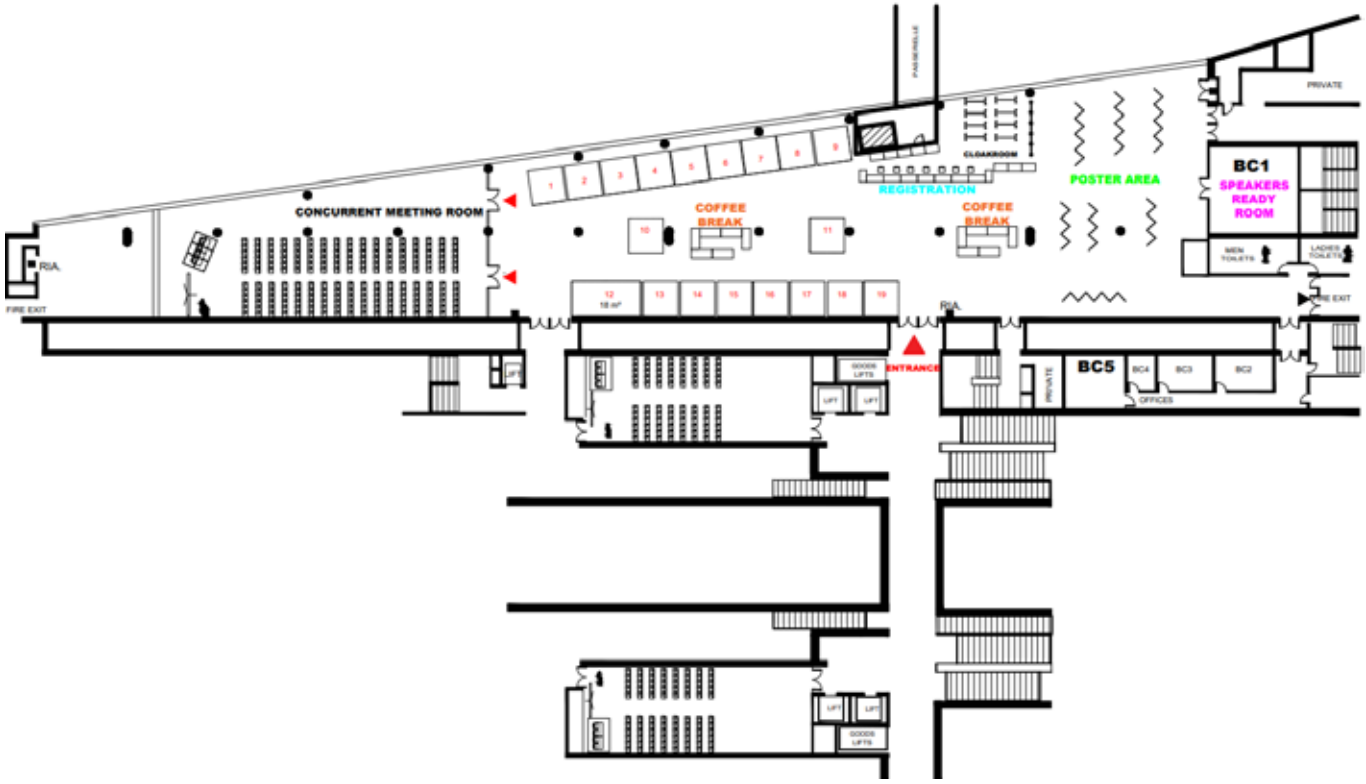
Exhibition visiting hours

1st July, Saturday	14:00 – 19:00
2nd July, Sunday	08:30 – 18:30
3rd July, Monday	08:30 – 18:30
4th July, Tuesday	08:30 – 12:00

EXHIBITION FLOOR PLAN



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Laboratory Imaging	3
Yourgene Health	4
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BioView	15
Thermo Fisher Scientific	16-17

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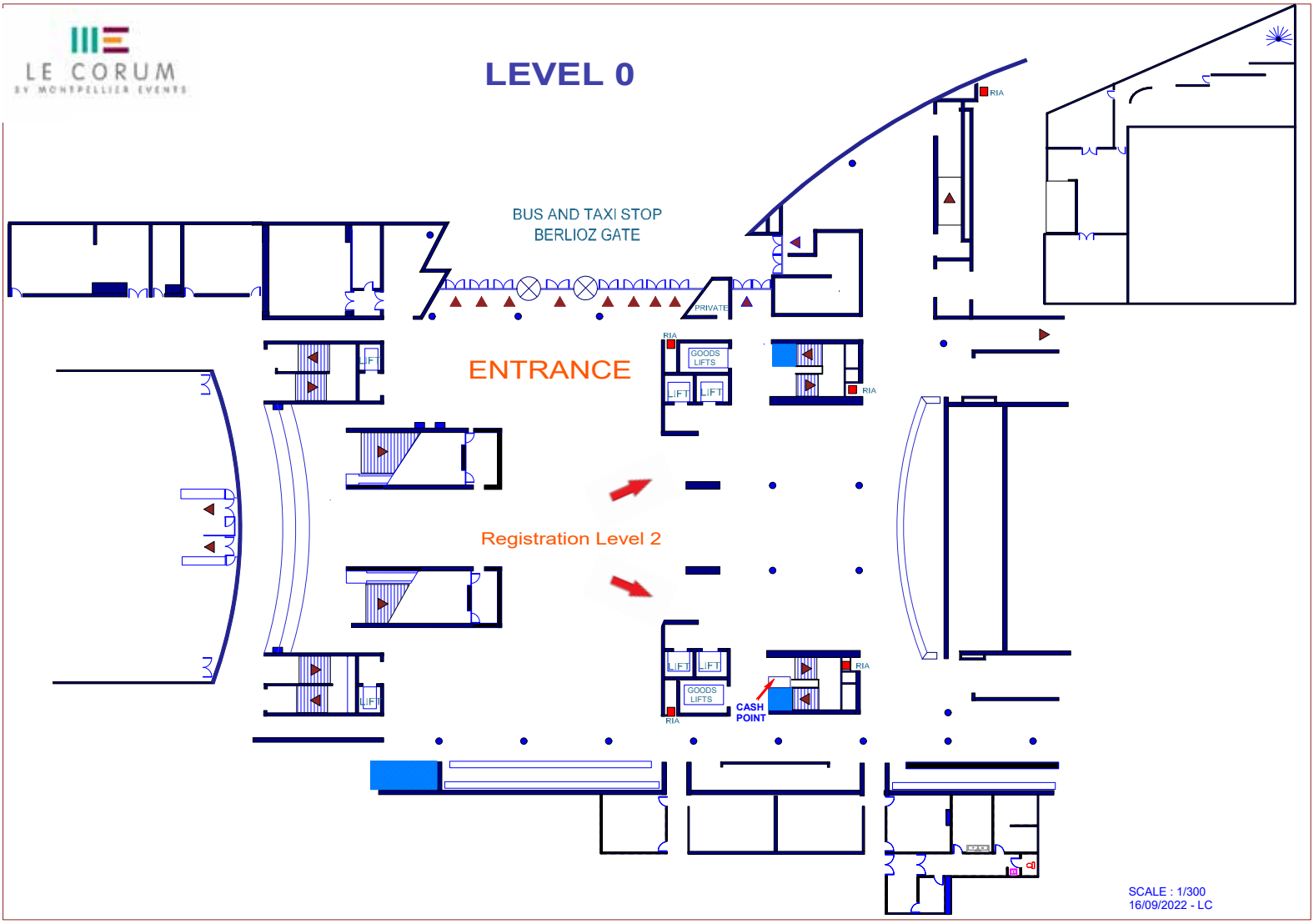
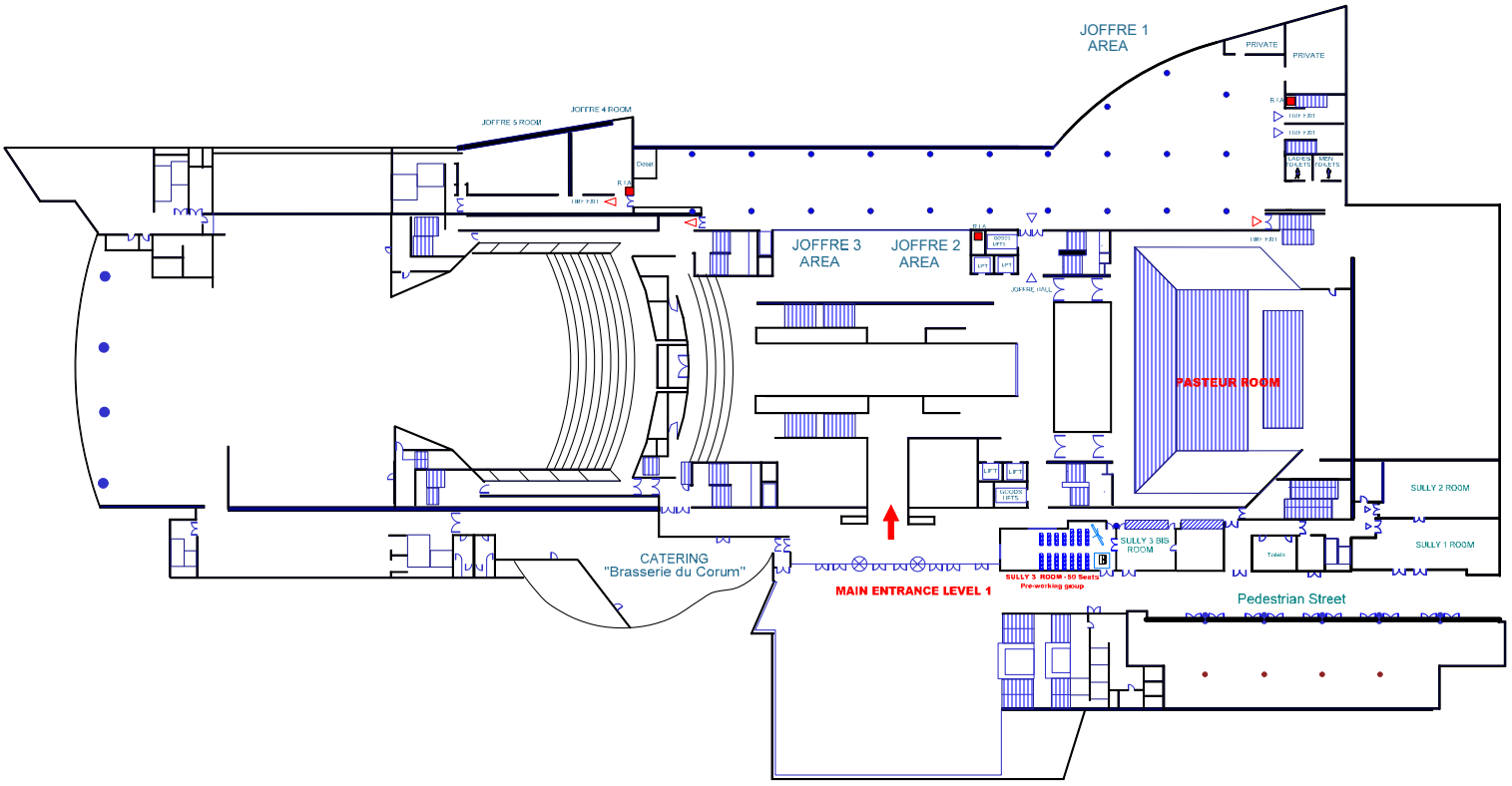


LE CORUM FLOOR PLAN

LE CORUM
BY MONTPELLIER EVENTS

LEVEL 1



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LE CORUM
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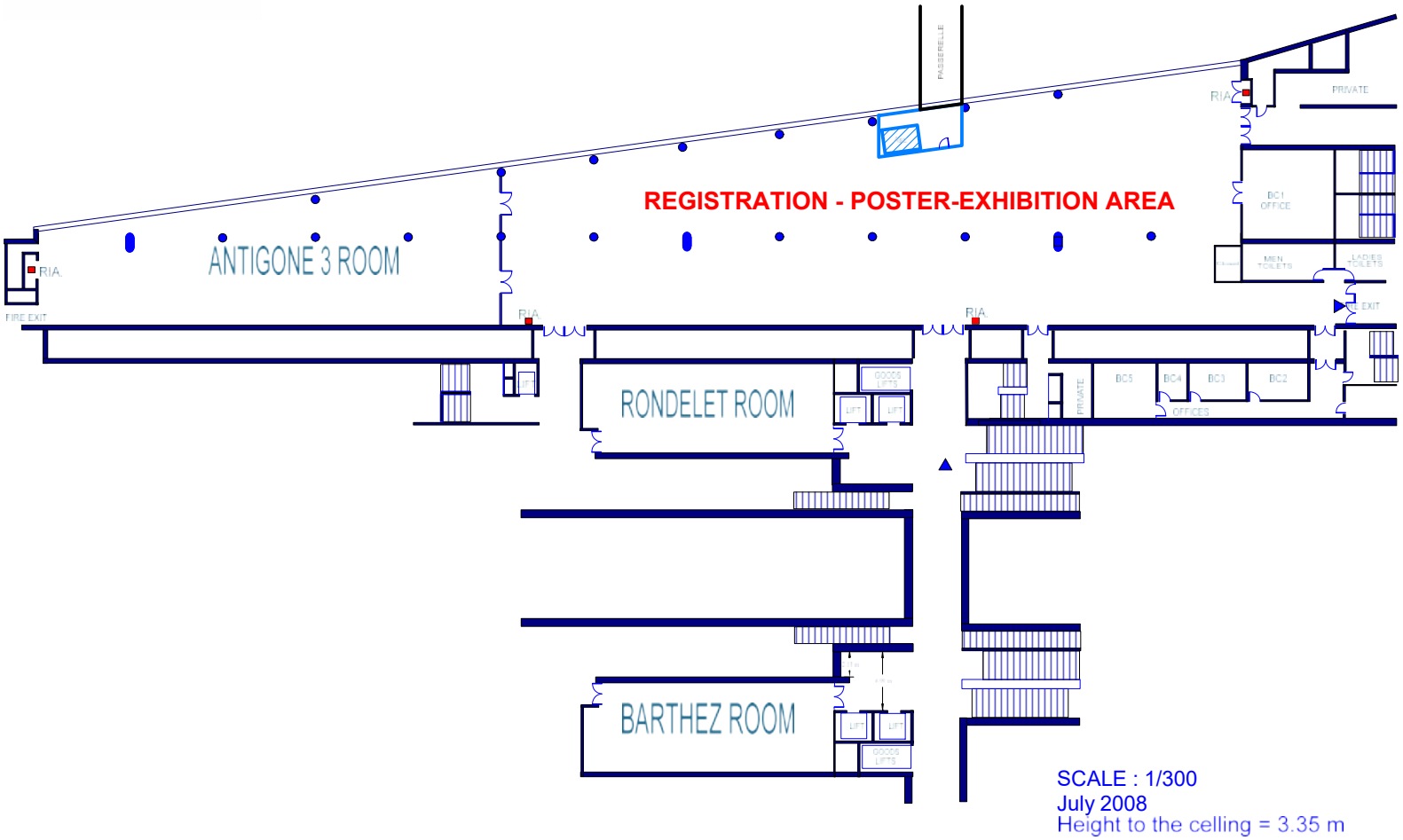
ANTIGONE HALL - LEVEL 2
Gross Surface = 1.440 sq m



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July 2008
Height to the celling = 3.35 m

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