Molecular Cytogenetics 2019, **12**(Suppl 1):30
https://doi.org/10.1186/s13039-019-0439-z

Molecular Cytogenetics

MEETING ABSTRACTS Open Access

Abstracts of the 12th European Cytogenomics Conference 2019

Salzburg, Austria. 6-9 July 2019

Published: 5 July 2019

COMMITTEES

E.C.A. Committee
Mariano ROCCHI - President
Kamlesh MADAN - First Vice-President
Pat HESLOP - HARRISON - Second Vice-President
Konstantin MILLER - General Secretary
Jean-Michel DUPONT - Treasurer

Scientific Programme Committee
Mariano ROCCHI - Chair
Claudia HAFERLACH
Dieter KOTZOT
Damien SANLAVILLE
Joris VERMEESCH
Orsetta ZUFFARDI

Organizing Secretariat
DEKON Congress & Tourism
Sultan Selim Mah. Eski Büyükdere Cad. Hümeyra Sok. No.12
34415 Seyrantepe - Kağıthane - Istanbul - TURKEY
Telephone: +90 212 347 63 00
Fax: +90 212 347 63 63
E-mail: eca2019@eca2019.com or dekon@dekon.com.tr
Visit www.dekon.com.tr for more information

Scientific Programme

SATURDAY, 6 July 2019

14:00-17:00 Permanent Working Groups

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

18:00-19:00 **Opening lecture**. Chairs: Mariano Rocchi - Dieter Kotzot

Joris Vermeesch: Somatic chromosomal mosaicim

SUNDAY, 7 July 2019

08:30-10:15 **Plenary session 1 - Recent advances in cytogenomics**

Chairs: Mariano Rocchi – Thierry Lavabre-Bertrand

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

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

10:45-11:15 **Plenary session 2 - 50 Years of chromosome banding**
Chairs: Kamlesh Madan - José Garcia-Sagredo

10:45-11:15 **Felix Mitelman:** Chromosome banding: the end of the Dark Ages

© The Author(s). 2019 **Open Access** This article is distributed under the terms of the Creative Commons Attribution 4.0 International License (http://creativecommons.org/licenses/by/4.0/), which permits unrestricted use, distribution, and reproduction in any medium, provided you give appropriate credit to the original author(s) and the source, provide a link to the Creative Commons license, and indicate if changes were made. The Creative Commons Public Domain Dedication waiver (http://creativecommons.org/publicdomain/zero/1.0/) applies to the data made available in this article, unless otherwise stated.

(Continued)

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 and Satellite Symposia

14:30 - 15:45 Concurrent Sessions

Concurrent Session 1 - 3D chromatin organization and dynamics
Chairs: Jean-Michel Dupont - Darío G. Lupiáñez

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 *Mus musculus*

Concurrent Session 2 - Clinical cytogenomics

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

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

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

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

Concurrent Session 4 - Human infertility

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

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

17:20-18:30 Poster session

MONDAY, 8 July 2019

08:30-10:30 **Plenary session 3 - Tumor Cytogenomics I**

Chairs: Felix Mitelman – Roberta Vanni

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

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

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

Concurrent Session 6 - Animal and plant cytogenomics I

(Continued)

Chairs: Pat Heslop-Harrison - Valerie Fillon

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 (*Bubalus bubalis*, 2n=50) cows raised in urban and rural areas

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

14:30-15:45 **Plenary session 4 - Chromosomal Imbalances**

Chairs: Mariano Rocchi - Konstantin Miller

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

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

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

Concurrent Session 8 - Accreditation, quality control and education

Chairs: Konstantin Miller - Albert Schinzel

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?

17:30-18:30 Poster session

18:30 **E.C.A. General Assembly**

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

Tuesday, 9 July 2019

09:00-10:30 **Plenary session 5 - Prenatal diagnosis**

Chairs: Damien Sanlaville - Maria Rosario Pinto Leite

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

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

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

11:25-12:15 **Keynote lecture**

Chair: Mariano Rocchi

Stylianos Antonarakis: Chromatin and single cell genomics, to understand the gene dosage imbalance in aneuploidies

12:15 **Closing Ceremony**: Mariano Rocchi

cohort. Since hyperhaploid and analogous hyperdiploid forms cannot be distinguished based on array patterns alone, we consider it essential and mandatory to assess their individual contribution in an at least semiquantitative manner with cytogenetic, FISH and/or DNA index analyses.

2.P30

Is RB1 gene deletion a prognostic marker in chronic lymphocytic leukemia

Beyhan Durak Aras[1], Nur Sena Inci[1], Abdulvahab Aslan[2], Olga Meltem Akay[3], Eren Gunduz[4], Tuba Bulduk[4], Sevgi Isik[1], Oguz Cilingir[1], Gulcin Gunden[1], Filiz Yavasoglu[5], Mustafa Karagulle[6], Ahmet Seyhanli[7], Sevilhan Artan[1], Hulya Ozen[8], Zafer Gulbas[9]
[1]Eskisehir Osmangazi University Faculty of Medicine, Department of Medical Genetics, Eskisehir-Turkey; [2]Private Umit Hospital, Department of Hematology, Eskisehir-Turkey; [3]Koc University Faculty of Medicine, Department of Hematology, Istanbul-Turkey; [4]Eskisehir Osmangazi University Faculty of Medicine, Department of Hematology, Eskisehir-Turkey; [5]Afyon Health Sciences University Faculty of Medicine, Department of Hematology, Afyon-Turkey; [6]Yunus Emre State Hospital, Department of Hematology, Eskisehir-Turkey; [7]Dokuz Eylul University Faculty of Medicine, Department of Hematology, Izmir-Turkey; [8]Eskisehir Osmangazi University Faculty of Medicine, Department of Biostatistics, Eskisehir-Turkey; [9]Anadolu Medical Center, Department of Hematology, Kocaeli-Turkey
Correspondence: Beyhan Durak Aras (bdurak@ogu.edu.tr)
Molecular Cytogenetics 2019, **12(Suppl 1):**2.P30

Deletion of 13q14 is the most common cytogenetic change (50%) in chronic lymphocytic leukemia (CLL), and it is a good prognostic factor if detected as a sole aberration in FISH analysis. In recent studies two main types of 13q14 deletions are proposed: type I (short) and type II (larger) which includes RB1. Some investigators suggest type II deletion to be associated with a more aggressive course. In this study, we aimed to investigate the relationship between RB1 gene deletion and overall survival (OS), disease stage and time to first treatment (TTFT) in patients with isolated 13q deletion.
RB1 deletion was analyzed in peripheral blood of 53 patients with isolated 13q deletion by FISH. The median age of patients is 66.88 ± 8.83 (42-88), and the 64% of patients are male. RB1 deletion was detected in 41% (22/53) of patients, but there was no statistically significant difference between RB1 deletion and TTFT, stage and OS (p=0,26, p=0,73, p=0,92). At the same time, statistically significant difference was detected between high 13q14 deletion (>60%) and TTFT (p=0,0038).
The statistical analysis of our data regarding association between RB1 deletion and TTFT, stage, and OS has not confirmed that type II deletion cause poor prognosis. But the patiens with RB1 deletion have shorter OS and TTFT. Unfortunately, our statistical evaluation was done without clinical data for six patients (4/6 of them are RB1 deletion positive); thus, our results must be regarded as preliminary. In particular, more data on patients with isolated 13q deletions are needed. A continuation of our study is in progress.

2.P31

Cytogenetic and molecular techniques (karyotype FISH mFISH and array CGH) for precise characteristization of structural chromosome abnormalities in chronic lymphocytic leukemia a case report

Maria Kislitsyna[1], Tatiana Obukhova[1], Tatiana Abramova[1], Lyuba Grebenyuk[1], Darima Badmazhapova[2], Milyausha Tagirova[3]
[1]National Research Center for Hematology, Cytogenetic Laboratory, Moscow-Russia; [2]National Research Center for Hematology, Gematology, Moscow-Russia; [3]National Medical Research Center for Obstetrics, Gynecology and Perinatology Named After Academician V.i.kulakov, Pathology, Moscow-Russia
Correspondence: Maria Kislitsyna (makislitsyna@gmail.com)
Molecular Cytogenetics 2019, **12(Suppl 1):**2.P31

Chronic lymphocytic leukemia (CLL) is a clinically and genetically heterogeneous disease. Interstitial deletion of 13q14 is the most frequent chromosomal aberration detected by FISH and can be represented by mono- or biallelic deletion. In 50% of patients del13q14 is submicroscopic and is not visible by chromosome banding analysis (CBA). aCGH allows not only to detect submicroscopic copy number changes, but also to determine their size. Using mFISH identification of all balanced and unbalanced chromosomal rearrangements is possible.
We report on a case of 81-year-old male with CLL analyzed by CBA, FISH, aCGH and mFISH for precise characteristization of the tumor clone.
CBA was performed by using oligonucleotide DSP30 и IL2. We used DNA probes for detection trisomy 12, deletions 13q14, 11q22, 17p13, rearrangements IGH for FISH; a CytoSure Constitutional v3 array (80x60k) and a 24XCyte mFISH probe.
By CBA we identified del(13)(q12q22) in one allele of chr13. FISH-analysis detected interstitial del13q14 in one allele and large deletion of chr13 in another allele (lack of signals of 13q14 and 13q34) and dim signal of IGHV (14q32). aCGH confirmed loss 14q32.33 and 13q14, but deletion region was q13q34 (116,74 Mb) and revealed additional change - gain of 3q26.1q29 locus, that was not consistent with karyotype data. mFISH showed additional chromosomal material 3q26q29 on chr13. Thus, on the basis of the results of all used methods we identified interstitial del13q14 in one allele, unbalanced t(3;13)(q26-q29;q13) with large del13(q13-q34) in another allele and partial deletion of locus IGHV/14q32.
We conclude that the combination of cytogenetic and molecular methods CBA, FISH, mFISH and aCGH is effective for detailed characterization of structural chromosomal abnormalities in patients with hematologic tumors.
"Written, informed consent for publication was obtained from the patient [or parent/guardian for patients under 16]"

2.P32

The assessment of chromosomal instability using telomere and centromere staining followed by M FISH technique Requirements and perspectives

Radhia M'kacher[1], Bruno Colicchio[2], Leonhard Heidingsfelder[3], Kevin Soehnlen[1], William M Hempel[1], Alain Dieterlen[2], Patrice Carde[4], Eric Jeandidier[5]
[1]Cell Environment, Dna Damage R&d, Paris-France; [2]Université De Haute-Alsace, Laboratoire Mips-Groupe Tiim3d, Mulhouse-France; [3]Metasystems Gmbh, Microscopy, Altlussheim-Germany; [4]Gustave Roussy Cancer Campus, Department Of Medicine, Villejuif-France; [5]Groupe Hospitalier De La Région De Mulhouse Et Sud-alsace, Service De Génétique, Mulhouse-France
Correspondence: Eric Jeandidier (jeandidiere@ghrmsa.fr)
Molecular Cytogenetics 2019, **12(Suppl 1):**2.P32

Purpose:
Chromosomal instability is the most important predictive and prognostic biomarker in the management of cancer patients. Telomere dysfunction is associated with dicentric chromosome formation leading to malignant cancer progression and poor clinical outcome. However, the detection of dicentric chromosomes using conventional techniques can easily be mistaken for translocations. In this study, we improved the detection of dicentric chromosomes and telomere dysfunction, both being chromosomal instability driving forces.
Material and methods:
A cohort of fifty patients with hematopoietic malignancies and 100 healthy donors were analyzed. In addition, twenty human cell lines were examined. Sequential analysis using telomere and centromere staining followed by M-FISH technique was performed to characterize telomere dysfunction and chromosomal aberrations.
Results:
Significant telomere shortening was found in cancer patients compared to similar age matched healthy donors (p<10-8). Telomere dysfunction (losses and deletions) was also detected in cancer