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MEETING ABSTRACTS Open Access

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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

4.P3

Independent evolution in one homolog of the 20 21 syntenic association in Cercopithecini monkeys (possibly) involving evolutionary neocentromere seeding

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Although the Cercopithecini is the most karyotypically diverse tribe of Old World monkeys (2n= 48 to 72), all species share a syntenic association of chromosomes homologous to human 20 and 21. Various forms, particularly in centromere position, are known in both between and within species. We used molecular cytogenetic methods, including detailed BAC-FISH assays, to analyze this chromosome in four species: Chlorocebus aethiops (CAE), Erythrocebus patas (EPA), Cercopithecus mitis albogularis (CAL) and Cercopithecus petaurista (CPE). We defined the ancestral form (form A) and traced evolutionary pathways to four variant forms. CAE was homozygous for form B, EPA was homozygous for form C and, notably, both Cercopithecus species were heterozygous for the ancestral form and highly derived forms: CAL (A, D) and CPE (A, E). A series of common inversions show that forms D and E emerged before CAL/CPE divergence (five million years ago). Because all four derivative forms share an initial inversion, ancestral and derived forms of CAL/CPE may have coexisted for 8 million years (time of CPE-CAL/CAE-EPA divergence). The heterozygosity of ancestral and derived forms for such a long evolutionary time could be explained by a selective advantage and, even if theoretically heterozygous inversions produce unbalanced gametes, crossing over may be suppressed. Finally, FISH shows that derivative forms D and E harbor much smaller amount of centromeric alpha satellite DNA than either the ancestral form A or other chromosomes. This may be due to multiple rearrangements that occurred in the centromeric regions of these chromosomes or the presence of an evolutionary new centromere in the last common ancestor of CAL and CPE. Future research, including sequencing, may help determine between these hypotheses.

4.P4

New insights into chromomere organization provided by lampbrush chromosome microdissection and high throughput sequencing

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Giant lampbrush chromosomes (LBCs) typical for growing oocytes of various animal species are characterized by specific chromomere-loop appearance and massive transcription. Chromomere-loop complexes represent universal units of chromatin packaging at LBC stage. While quite a good progress has been done in investigation of LBCs structure and function, chromomere organization still remains poorly understood.
Our previous studies showed that lampbrush chromosome microdissection is a powerful tool for investigation of tiny chromosomal regions with high precision. To extend our knowledge on chromomere organization and genomic context, we applied microdissection on chicken LBCs. In particular, 30 individual chromomeres were dissected one after another along the macrochromosome 4. FISH on LBCs allowed to map the microdissected regions precisely and to evaluate their transcriptional activity in growing oocytes. The data on genomic context of individual chromomeres were obtained by high-throughput sequencing. Alignment of adjacent chromomeres to chicken genome assembly (build Galgal5) provided information on chromomeres' size and genomic boarders indicating that prominent marker chromomeres are about 4-5 Mb in size, while common chromomeres - 1.5-3.5 Mb. Analysis of genomic features showed that the majority of chromomeres combine gene-dense and gene-poor regions, while massive loopless DAPI-positive chromomeres lack genes and are remarkably enriched with different repetitive elements. Finally, LBC chromomeres were compared with chromatin domains (topologically associated domains (TADs) and A/B-compartments) earlier identified by Hi-C technology in interphase nucleus of chicken embryonic fibroblasts. Generally, the results obtained suggest that chromomeres of lampbrush chromosomes do not correspond unambiguously to any type of well-established spatial domains of interphase nucleus of somatic cells.
The research was supported by Interuniversity Partnership Programme, grant of the President of the Russia (MK-1630.2017.4) and RSF (19-74-20075) and performed using experimental equipment of the Research Resource Center MCT of SPbU.

4.P5

Cytogenomics of a sex specific marker on the W chromosome of the invasive mosquitofish Gambusia affinis

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The Western mosquitofish, Gambusia affinis is an interesting model for sex chromosome organization and evolution, because it shows female sex chromosome heterogamety and a ZW/ZZ sex chromosome system. Using genomic and transcriptomic data, we previously identified a G. affinis female sex specific marker highly homologous to the aminomethyl transferase (amt) gene of the closely related platyfish (Xiphophorus maculatus).
In the study presented here, we dissected the genomic region using exonic PCR probes from the G. affinis amt gene, which we localized on the long arm of the W chromosome (Wq) by fluorescent in-situ hybridization. We then obtained a deeper insight into the large-scale genomic structure of the G. affinis W chromosome in comparison to its sister species G. holbrooki with male heterogamety and an XY sex chromosome system. To this end, we applied intra- and interspecific comparative genomic hybridization (CGH) and comparative expressed sequence hybridization (CESH), as well as FISH with rDNA and oligonucleotide repeat probes, and immuno-fluorescence.
Our CGH analyses showed that the long arm of the G. affinis W chromosome is enriched for repetitive sequences. Despite this, by conventional C-banding and by anti-5-methylcytosine immunofluorescence staining we could demonstrate that the G. affinis Wq is neither heterochromatic nor hypermethylated and therefore not epigenetically silenced. In contrast, according to our CESH data the entire Wq appears to be highly transcribed. In addition, the terminal region of Wq comprises one of the three major active NORs, which we detected by silver staining and rDNA FISH.
Taken together, these findings led us to speculate that certain expressed noncoding elements from the amt genomic region with architectural localization along Wq may play a role in sex specific gene dosis compensation in this ZZ/ZW system.