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Interspersed telomeric sequences (TTAGGG)n distribution in Primates and Tupaia minor (Scandentia)

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Over recent years a growing number of studies suggest that the (TTAGGG)n repeats, characterizing telomeres of Vertebrates, can be detected not just at the traditional position of chromosomal ends, but at interspersed sites as well. Cytogenetic and molecular studies indicate these interspersed sequences, known as Interstitial Telomeric Sequences (ITSs), as important elements affecting genome plasticity and evolution. Even if ITSs role is still not fully understood they have been associated to (1) chromosomal rearrangements; (2) mechanism of genome reorganization, such as double DNA strand break repair, involving retrotransposons; (3) process of recombination and amplification of the terminal end of chromosomes; (4) mechanism of centromere/telomere interchanges. In this context, to elucidate ITSs role and distribution, we mapped through Fluorescence In Situ Hybridization (FISH) the telomeric PNA (TTAGGG)n probe on 16 species representative of each major primates groups and Tupaia minor (Scandentia). Our PNA mapping results, as expected, showed hybridization signals at telomeric ends of chromosomes in all samples and consistently at several centromerical and interstitial position in many analyzed species. The distribution of ITS in primates species have been analyzed in a phylogenetic perspective using Tupaia minor as outgroup. In particular in Tupaia the probe hybridization revealed many bright ITSs on at least eleven chromosome pairs, both bi-armed and acrocentric; our comparative analysis shows probe signals, both in correspondence to fusion points of human syntenic associations, and on other chromosomes uniformly composed of a single human synteny. These data compared with that of literature in Primates let us to support the hypothesis about a correlation between ITSs and rearrangements, thus indicating their possible role in genome organization and evolution.

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The Japanese quaıl genome a cytogenetıc revısıon

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Low quality of the existing quail genome assembly is an important limiting factor for using the Japanese quail as a developmental biology animal model and applying genomic selection to quail breeding. Japanese quail Coturnix coturnix japonica has a relatively small genome (≈1.41 Gb) packed into 39 chromosome pairs. Its karyotype is very similar to the chicken one. It is accepted that quail and chicken chromosomes are orthological, a few intrachromosomal rearrangements were described. The microchromosome morphology is different in these two species. Quail microchromosomes are mainly submetacentric having heterochromatic short arms while in chicken they are acrocentric. In 2016, the quail genome assembly was produced on the chicken genome assembly galGal3 as a reference. Currently, it includes 32 (of 38 + Z and W) linkage groups. However, the quail assembly contains a number of sequence gaps because of repetitive coding and non-coding DNA elements. In this work, we have identified new highly repeated tandem sequences within unassembled quail short raw reads and mapped them to Japanese quail genome. To verify the quail draft genome assembly we performed systematic ZOO-FISH experiments on Japanese quail chromosomes with chicken BAC clone probes from CHORI-261 chicken BAC library. BAC clones were selected according to their chromosomal positions and assigned for specific sequence markers and high cross-species hybridization efficiency. All BACs used were positioned in the quail genome assembly. In some cases we have found discrepancies between positions of the markers in the quail genome assembly and physical maps, caused by the newly identified interspecific rearrangements.

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Evaluation of chromosome microrearrangements of a horse applying array CGH

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A selection of research techniques and quality of obtained metaphase plates plays a crucial role in diagnostic cytogenetics. The dynamic development of high-resolution cytogenetic techniques such as array CGH allows increasing the accuracy of diagnosis of chromosomal disorders and the early elimination of breeding individuals carrying aberrations. Therefore, the aim of the study is a broad range analysis of chromosome abnormalities, including unbalanced microrearrangements, with the use of a highly precise technique of array comparative genomic hybridization (aCGH), in individuals affected by reproductive and/or development disorders.

One of the examined animals was 19 -year old mare, which has overall female body conformation and oestrus, however, has not had a foal. A karyotype analysis showed that of 333 metaphases,7 had the normal karyotype 64, XX while 326 had 65, XXX. In the next step, we carried out DNA labeling using SureTag DNA Labeling Kit (Agilent). The labelled DNA was then subjected to 40 h hybridization onto 2x400K custom equine CGH microarrays (Agilent). The washed microarrays were scanned with SureScan G2565CA Scanner (Agilent). The obtained raw data were analysed using Agilent Genomic Workbench (7.0).

The comprehensive analysis using aCGH technique revealed 426 aberrations, including 377 amplifications and 49 deletions, in the genome of the investigated horse. As many as 3 deletions and 374 amplifications were identified on chromosome X. The largest number of aberrations was detected on ECA1 and 12 and they mainly embraced genes connected with MHC II - DRB and DQA. The obtained results allowed characterising genes and pathways which are altered in the genome of the investigated mare.

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4.P28

Screening of the Polish young horse population for detection of sex chromosomes abnormalities preliminary results

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