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Second Announcement

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We describe an eight year old male and his first degree double cousin with a distal 10q trisomy resulting from a maternal balanced reciprocal translocation involving chromosome 9 and 10. Their karyotype using GTG-banding was 46,XY,der(9)(9qter→9p24::10q25→10qter)mat. The translocation was also confirmed by FISH studies. We found a balanced translocation involving chromosomes 9 and 10 [46,XX,t(9;10)(p24;q25)] with cytogenetic and FISH studies in the mothers of these children who had a normal phenotype. The clinical features of our cases are as follows: mental retardation, small nose with depressed nasal bridge, hypertelorism, blepharophimosis, micrognathia, dental anomaly and high-arched palate.

1.249-P

Cytogenetic and clinical study of a male infant with ambiguous genitalia

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A one-year-old male infant was clinically diagnosed as an intersex case with ambiguous genitalia and hypospadias. Clinical, hormonal and genetic findings are presented. In the examination of patient, bilateral testicular volume and phallus was found undersized. Serum concentration of testosterone was found low level. G-banding of his chromosomes show that patient has balanced translocation involving chromosome 3 and 4 [46,XY,t(3;4)(p25;q31.3)]. This finding was confirmed by fluorescent in situ hybridization (FISH). The proband inherited this translocation from his father. His sister has this translocation as well. But clinically father and sister of proband were normal.

2.1-O

Progression dynamics of evolutionary-new centromeres

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Extensive FISH experiments with BAC probes, as an independent and complementary approach to the official sequence assembly of the macaque genome, were utilized to compare macaque (MMU)/human synteny organization. Surprisingly, we found that 9 macaque and 5 human Evolutionary New Centromeres (ENC) originated after Old World monkey/Hominoidea divergence. Clearly, ENCs have a significant impact on shaping genomes. Construction of a BAC contig of the pericentromeric region of the ENC of macaque chromosome 4 (human 6) allowed us to disclose the dynamics of ENC formation and progression by comparison to human region at 6q24.3, which conserves the ancestral genomic organization. The analysis revealed that a segment of 250 kb in the seeding region was extensively duplicated around the macaque ENC. These duplications were strictly intrachromosomal. Our results support hypotheses that novel centromeres triggers only local duplication activity, and that the absence of genes in the seeding region played an important role in ENC tolerance and progression.

2.1-P

New polymorphism in heterochromatin regions of Equus asinus chromsomes detected by horse DNA satellite probes

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Non centromeric heterochromatin bands have been described in Equus asinus species (EAS, 2n=62 F. Equidae) in the juxtacentromeric and telomeric positions in different chromosomes of the karyotype. Chromosome EAS1 presents heterochromatic bands in a juxtacentromeric position in the q-arm and in a terminal position in the p-arm. Satellite DNA polymorphism in the constitutive heterochromatin of Equus asinus chromosome 1 (EAS1) is presented. In our cytogenetic study, the chromosomes of five animals with normal karyotype were analysed by sequential G and C banding. Fluorescence in situ hybridization was then performed on the chromosomes of these animals using two probes isolated from a horse genomic library and containing sequences from the two major horse satellite families (C37 and 2P1) Metaphases spreads were prepared from fibroblast cultures and peripheral blood cultures of five different non related and phenotypically normal Equus asinus individuals. The metaphases were then in situ hybridised with two horse satellite DNA probes: the C37 and 2P1 clones were isolated from a horse genomic library in the phage vector lambda-GEM11. C37 contains a tandem repeat of 221 bp and 2P1 clone contains a satellite sequence composed of tandem reiterations of a 23 bp unit. FISH results obtained have shown different hybridization patterns in different donkey specimens; four different polymorphic forms of EAS1 have been identified. These patterns can be interpreted as the result of different rearrangements that occurred in the heterochromatic regions of chromosome EAS1.

2.2-P

Chromosomal mapping of rRNA and histone gene clusters in mussels and clams

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The class Bivalvia includes some of the best-known marine invertebrate species, many of which are commercially harvested around the world. Most chromosome studies on species of the families Mytilidae and Veneridae have been performed using classical cytogenetic techniques and there are only a few works using fluorescent in situ hybridisation (FISH). In order to physically map the rRNA and histone gene clusters to mitotic and meiotic chromosomes of species of Mytilidae (Mytilus, Brachidontes, Perumytilus,) and Veneridae (Dosinia, Venerupis, Ruditapes, Venus), chromosome preparations were obtained from gill and gonadic tissues of juvenile individuals after hypotonic treatment and fixation with methanol/acetic acid. Surface spread synaptonemal complexes from mature males were also obtained. Species specific probes for histone genes, 18+28S rDNA internal transcribed spacers (ITS) and 5S rDNA were generated by PCR. While a variable number of both histone gene clusters and major and minor ribosomal gene clusters were detected in the species belonging to the Mytilidae, a single gene cluster for each gene family was noted in all Veneridae species. On the contrary, the distribution patterns of the family clusters on the chromosomes of both Mytilidae and Veneridae showed large differences among species.

2.3-P

Effects of chromium picolinate on micronucleus frequency and morphology of lymphocytes in calves

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Polymorphism of *Equus asinus* chromosome 1 detected by horse satellite probes

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Introduction

Non centromeric heterochromatin bands have been described in *Equus asinus* species (EAS, 2n=62 F. Equidae) in the juxtacentromeric and telomeric positions in different chromosomes of the karyotype. Chromosome EAS1 presents heterochromatic bands in a juxtacentromeric position in the q-arm and in a terminal position in the p-arm. It was shown that the size of these bands is polymorphic (Alaoui et al. 2004)
In this work we analyze the polymorphism in the constitutive heterochromatin of EAS1 by FISH using probes for the two major horse satellite families.

Material and Methods

Cell culture and chromosome preparations

In this work we analyze the polymorphism of EAS1 by FISH using probes for the two major horse satellite families. Metaphase spreads were prepared from fibroblast cultures or peripheral blood cultures of five different non related and phenotypically normal *Equus asinus* male specimens. The chromosomes were sequentially G-C banded (adapted from Seabright 1971 & Sumner 1972).

Fluorescent *in situ* hybridization (FISH)

The metaphases were then hybridized *in situ* with two horse satellite DNA probes: C37 and 2P1. The C37 and 2P1 clones were isolated from a horse genomic library in the phage vector lambda-GEM11 (for a description of the library see Anglana et al. 1996). Fragments from C37 and 2P1 phage clones were subcloned in pUC18 plasmid and sequenced. The sequences were deposited in the GenBank under accession numbers AY029358-AY029360.
In situ hybridization with biotinilated probes was performed as previously described (Carbone et al. 2006)

Results and Discussion

FISH results obtained have shown different hybridization patterns in different donkey specimens (Fig.1); four different polymorphic forms of EAS1 have been identified (Fig.2). These patterns can be interpreted as the result of different rearrangements that occurred in the heterochromatic regions of chromosome EAS1.

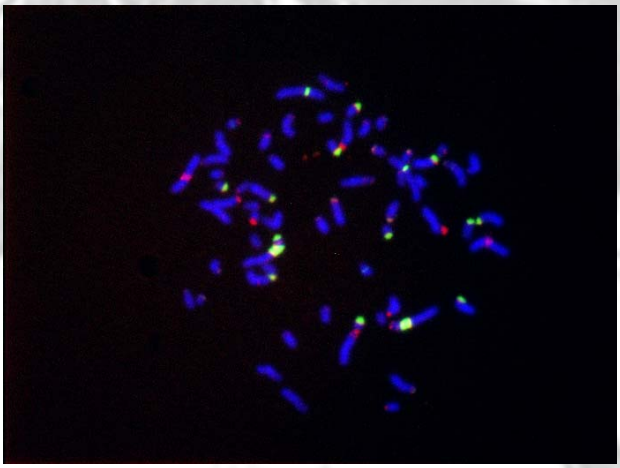

Figure1: Results of FISH using horse DNA satellite probes C37 and 2P1 to a metaphase of *Equus asinus*

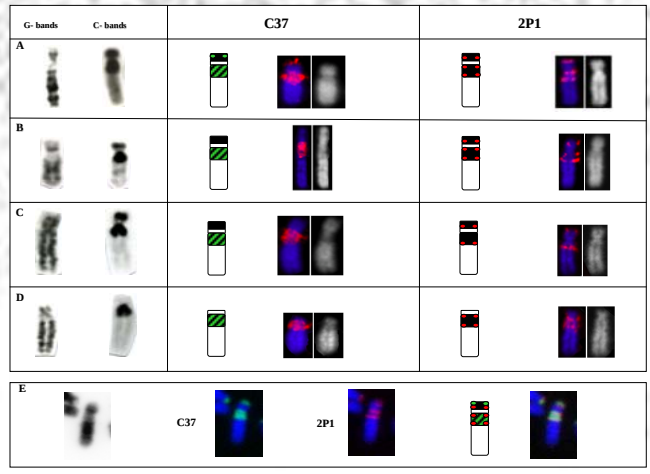

Figure 2: Polymorphism of *Equus asinus* chromosome 1.
From A to D sequential G/C banding patterns (left panels), hybridization with the C37 satellite probe (middle panels) and hybridization with the 2P1 satellite probe (right panels) of the four variants are shown.
E: two-color hybridization of EAS1. C37 hybridization (green), 2P1 hybridization (red)

Bibliography

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Agreements

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