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Structural variation has played an important role in the evolutionary restructuring of human and great ape genomes. Recent analyses have suggested that the genomes of chimpanzee and human have been particularly enriched for this form of genetic variation. We investigated the extent of structural variation in the gorilla lineage by generating 10-fold genomic sequence coverage from a western lowland gorilla and integrating these data into a cytogenetic framework of structural variation. We discovered and validated over 7665 structural changes within the gorilla lineage, including sequence resolution of inversions, deletions, duplications, and mobile element insertions. Thus we demonstrated that the gorilla genome has been subjected to the highest rate of segmental duplication.

Besides, gorilla and chimpanzee chromosomes differ from human chromosomes by the presence of large blocks of subterminal heterochromatin thought to be composed primarily of arrays of tandem satellite sequence. We explored their composition and organization and showed a complex organization made of specific sets of segmental duplications, which have hyperexpanded in concert with the formation of subterminal satellites. These regions are highly copy number polymorphic between and within species and copy number differences can be accurately estimated by assaying read-depth of next-generation sequencing datasets. Phylogenetic analyses suggest that the structures have arisen independently in the two lineages with the exception of a few seed sequences present in the common ancestor of humans and African apes. We proposed a model where an ancestral human-chimpanzee pericentric inversion and the ancestral chromosome 2 fusion both predisposed and protected the chimpanzee and human genomes respectively to the formation of subtelomeric heterochromatin. Our findings highlight the complex interplay between duplicated sequences and chromosomal rearrangements that alter the cytogenetic landscape in a short period of evolutionary time.

9.P7

Distribution of rDNA in diploid and poliploid Lolium multiflorum Lam. reveals fragile sites in 45S rDNA region

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Distribution, number and location of ribosomal DNA (rDNA) sites on chromosomes were evaluated and region transcriptional activity was described in seven plants of two genotypes of Italian ryegrass (Lolium multiflorum Lam.) (diploids and polyploids), as well as in an offspring resulting from interbreeding. Nucleolus and nucleolus organizing regions (NOR) were marked with silver nitrate and fluorescence in situ hybridization (FISH) was performed with 5S and 45S rDNA as probes. Hybridization procedures were performed on slides previously stained with AgNOR method. The 5S rDNA site was highly conserved, while the 45S rDNA ones had wide variability, even showing more than one site in the same chromosome (synteny). One of the genotypes had number of 45S signals higher than expected. Approximately 93 % of the metaphases analyzed had at least one chromosome breakage/gap in the 45S rDNA site, resulting in fragments formation. Thus, the 45S rDNA site corresponded a fragile chromosomal region in L. multiflorum. These events can affect genome organization and cause new chromosomal rearrangements which, along with some other events, are responsible for the microevolutionary changes involved in differentiation and speciation processes. Not all 45S rDNA regions was transcriptionally active, and epigenetic mechanisms may be related to the silencing of ribosomal genes. Also, both variation in number and size of nucleoli and mechanisms of nucleolar fusion were observed in L. multiflorum.

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9.P8

Robertsonian translocation polymorphism in impalas (Acpyceros melampus): impact on meiosis and reproduction

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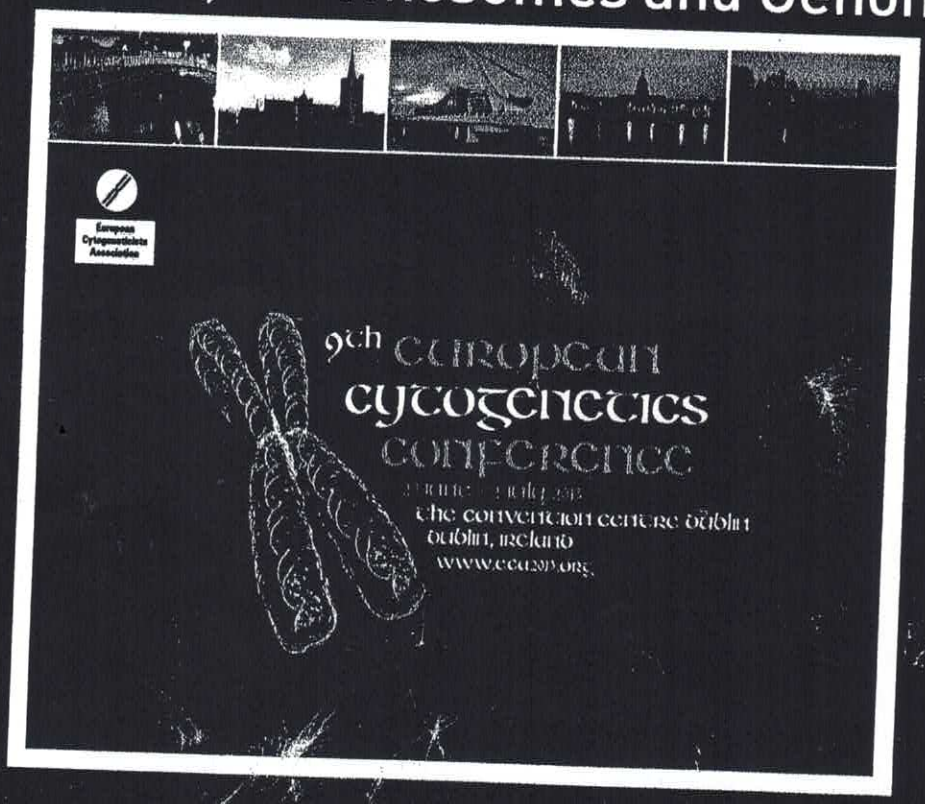

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