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Identification and Expression of Human Endogenous Retrovirus, HERV-W, in Human Tissues and Cancer Cells

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Human endogenous retroviral family, HERV-W, has been implicated in neuropsychiatric disease. Its transcriptional activation in the brain has been connected to schizophrenia. In this study, we examined the structural genes (*gag, pol, env*) sequences of HERV-W family from 12 normal tissues and 18 cancer cells of the human by RT-PCR analysis. For *gag* and *pol* genes, their expression pattern indicated tissue or cell specificity, whereas the *env* gene was expressed in all tissues and cancer cells examined. In order to identify active HERV-W elementsin tissues and cancer cells, the RT-PCR products were cloned and sequenced. Fifty and forty-four putative amino acids of the HERV-W *env* fragments from tissues and cancer cells, respectively, were isolated whereas only two putative amino acids of the HERV-W *pol* fragments from cancer cells were identified. Phylogenetic analysis indicated that several sequences identified from human monochromosome previously have sister relationship with those clones from different tissues or cancer cells. The data suggest that recently proliferated HERV-W elements are actively expressed in human tissues and cancer cells. These active HERV-W elements deserve further investigations as potential pathogenetic effects to various human diseases including cancers.

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Comparative Analysis of Large Genome in Cross-species

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With the availability of whole-genome sequence for an increasing number of species, we are now faced with challengeof annotating gene, regulatory elements and repeat elements within these genomes. Comparative analysis of large genomes from cross-species at varying evolutionary distances is a powerful approach for identifying coding and functional non-coding sequences.Here we describe a global alignment method that is designed to be fast and efficient sequence alignments of large genomic regions over mega bases pair. Using this alignment approach, we examined rearrangement loci in human chromosome 21 and chimpanzee chromosome 22. We also developed an approach for identifying orthologous regions by first HSP (Highest Segment Pair) regions using local alignments and then large orthologous genome based on extension of anchors at HSP regions in two species. We compared 30 Mb of human chromosome 21 with chimpanzee chromosome 22, and identified 41 genomic rearrangements (deletions and insertions ranging in size from 0.3 to 200 kb). We also discussed evolutionary features throughout comparative analyses of Ka/Ks(non-synonymous / synonymous substitutions) rate in orthologous 119 genes of chromosome 21 and 53 genes of MHC-I class in human and chimpanzee genome.

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HSAP: Human Endogenous Retroviruses Structure Analysis Program

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Human endogenous retroviruses (HERVs) comprise up to 8% of the human genome and are believed to have a significant impact on the human genome structure and function related to human evolution and disease. They have structural genes, *gag, pol, env*, and LTR elements in their flanking region. Bioinformatic analysis of HERVs was performed using the RepeatMasker program to identify repeat sequences. Following the analysis of the Repeat Masker, we used a mergence algorithm that makes to define more precisely HERV families and to ascribe individual members for their families. Using this information, we developed a program, named HSAP (HERVs Structure Analysis Program). The HSAP shows structural features of HERVs in the genome database by searching LTR-*gag*-*pol*-*env*-LTR sequences. In addition, it also shows graphical representation of HERV structures. Therefore, the HSAP could be of great use the exploring of HERV features and biological roles in human genome.

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New Finding of Hybrid Human Endogenous Retroviral HERV-9/IP10FH Elements and Reexamination of Hybrid HERV-H/E Elements

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Use the Repeatmasker program, a new hybrid human endogenous retroviral family (HERV-9/IP10FH) from human genome was identified and analysed. The HERV-IP10FH family encoded the *env* protein only is a non-autonomous endogenous retrovirus family. New hybrid family showed structure of LTR(9)-*gag*(9)-*pol*(9)/*env*(IP10FH)/LTR(9) with the size of 6.3-8.6kb. The hybrid HERV elements showed deletion event in LTR elements on human chromosomes 4, 5, and 6 with the size of 2.5-6.5kb, whereas complete structural forms were appeared on human chromosomes 1, 2, 4, 8, 10, 19, and Y. The HERV-9/IP10FH hybrid family seems to be generated by homologous recombination of some specific sequences 'AAGGGGAAGGAGA' near the boundary of their location. We also analysed HERV-H/E hybrid elements. The hybrid sequences contained structure of LTR(H)-*gag*(H)-*pol*(H)/ *gag*(E)-*pol*(E)-*env*(E)-LTR(E) /*pol*(H)-*env*(H)-LTR(H), indicating that HERV-E element truncated with 3' LTR element has been inserted into the structural *pol*gene of HERV-H element. Taken together, we suggest that bioinformatic analyses of the hybrid HERV could be contribute for understanding INDEL (insertion/deletion) mechanism.