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Transcriptional Variation of NOS3 LTR Element and Its implication in Dementia

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Alzheimer's disease (AD) has been reported to have an association with vascular risk genetic factor (angiotensin converting enzyme, methyltetrahydropholate-reductase, and NOS3 gene polymorphisms). Previous studies demonstrated that β-amyloid peptides (AβP), the paramount component of senile plaques of AD, inhibit the enzymatic activity of eNOS. We studied NOS3 gene which encode eNOS (NOS3, nitric oxide synthase 3) protein in transcriptional level and relationship with HERV (human endogenous retroviruses). The NOS3 gene contains a solitary long terminal repeat (LTR) between promoter region and first exon. In silico analysis, we compared the NOS3 gene sequences between human and mouse, indicating that human NOS3 gene acquired the LTR element during primate evolution. The LTR element has been integrated into the primate genome after divergence of Old World Monkey. We also demonstrate that the LTR of the NOS3 gene has a promoter activity in humans.

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HYBRIDdb: a database of transcriptional and genomic hybrid genes in human genome

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Hybrid genes have been reported to be candidate risk factors of human tumors through the gene mutation, translocation, inversion, and rearrangement. The occurrence of hybrid gene could be also advantageous factors of new gene and different transcripts during the hominid evolution. HYBRIDdb is a database of transcriptional and genomic hybrid gene in humans. Our system encompasses the bioinformatics analysis of mRNA, EST, cDNA, and genomic DNA sequences in the INDC databases (http://insdc.org). The system could identify hybrid genes containing overlapping sequences of transcripts or sense/antisense genes such as tail-to-tail or head-to-head gene pairs. We searched the 28,171 genes listed in NCBI database for hybrid genes and analyzed the structural patterns in the human genome, indicating that 3,404 gene pairs were detected as hybrid form at the genomic (1,060) and transcriptional products (2,344).The hybrid genes are classified into 4 groups as chromosome translocation-derived fusion transcript, splicing-derived fusion transcript, tail-to-tail genome-level hybrid and head-to-head genome-level hybrid. We believe that our work will be of significant interest to genome scientists and may help them to gain insight into implication of hybrid genes in human evolution and diseases.