

HUMAN AND NON-HUMAN PRIMATE PSEUDOGENES - INTRODUCTION-

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A pseudogene is defined to be a gene that has lost its function, especially it has lost the ability of coding protein. In general, a pseudogene is generated by gain of premature stop codons due to point mutations or frame-shift mutations. The acceptance of premature stop codons in a gene depends strongly on a functional constraint or functional importance of the gene product. In most cases, premature stop codons are accepted only when a gene is functionally compensated by duplicates. However, pseudogenization of single copy genes (a single-copy pseudogene) has been sometimes found in humans and non-human primate genomes. Furthermore, in some cases, deterioration of a gene has taken place independently in different primate lineages. This might be “convergent evolution in pseudogenization.” For example, urate oxidase gene, of which product converts purine to allantoin in a purine catabolic pathway, was deteriorated independently in great-apes/humans and gibbons. Further, a search of human specific pseudogenes reveals 14 cases of independent loss of function in single-copy genes in human and non-human primate lineages. These single-copy pseudogenes might reflect changes in functional significance in biosystems by some particular reasons. These reasons could be related to species-specific traits in morphology and physiology. In this introduction, I review these single-copy pseudogenes and the convergent evolution of pseudogenization in humans and non-human primates and argue the biological significance of pseudogenizations.

Keywords: functional compensation, pseudogenization, environmental changes, gene duplication