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ACTIVITY OF LINE TRANSPOSONS AND SELECTED GENETIC DISEASES

Summary

LINE transposons (Long Interspersed Nuclear Elements) are mobile, endogenous genetic elements widespread in eukaryotic genomes. Their ability to spread out with help of reverse transcriptase by using RNA intermediates indicates that they belong to autonomous retrotransposons. Original element is transcribed, then RNA undergoes reverse transcription and as a DNA fragment it is inserted into another part of the genome. Despite of the presence of over 500.000 of their copies in the human genome, majority of LINEs became inactive due to structural changes during the process of evolution. Sequences that retained their original function, play an important role in organization and functioning of genomes. Their activity results in destabilization of a genome structure, as a result of *de novo* insertions of LINEs and changes caused by homologous recombination between them. They can cause changes in the level of gene expression by interfering with alternative splicing (resulting in exon skipping or selecting cryptic splice sites), generating polyadenylation signals or providing alternative promoters. LINE retrotransposition is active mainly during the early stages of embryogenesis. In normal somatic cells this process is silenced by epigenetic mechanisms. Changes in DNA methylation levels of these elements is one of the main indicators of their activity associated with multiple genetic diseases. Correlations between LINE activity and multiple forms of neoplasms are mostly described in this paper.

Key words: cancer genetics, human genetic diseases, L1 sequences, LINE, transposon activity