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# LINE AND SINE

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ZOOLOGY: SEM- V, PAPER- C12T: GENETICS, UNIT 7: TRANSPOSABLE GENETIC ELEMENTS

**Retrotransposons** (also called Class I transposable elements or transposons via RNA intermediates) are a type of genetic component that copy and paste themselves into different genomic locations (transposon) by converting RNA back into DNA through the process reverse transcription using an RNA transposition intermediate.

Through reverse transcription, retrotransposons amplify themselves quickly to become abundant in eukaryotic genomes such as maize (49–78%) and humans (42%).

There are two main types of retrotransposon, long terminal repeats (LTRs) and non-long terminal repeats (non-LTRs). Retrotransposons are classified based on sequence and method of transposition. Most retrotransposons in the maize genome are LTR, whereas in humans they are mostly non-LTR.

## Long terminal repeats (LTRs):

Long strands of repetitive DNA can be found at each end of a LTR retrotransposon. These are termed long terminal repeats (LTRs)

that are each a few hundred base pairs long, hence retrotransposons with LTRs have the name long terminal repeat (LTR) retrotransposon. LTR retrotransposons are over 5 kilobases long.

## Non Long terminal repeats (Non-LTRs):

Like LTR retrotransposons, non-LTR retrotransposons contain genes for reverse transcriptase, RNA-binding protein, nuclease, and sometimes ribonuclease H domain but they lack the long terminal repeats. RNA-binding protein binds RNA-transposition

intermediate. Nucleases are enzymes that break phosphodiester bonds between nucleotides in nucleic acids. Instead they have short repeats that can have an inverted order of bases next to each other aside from direct repeats found in LTR retrotransposons that is just one sequence of bases repeating itself.

Although they are retrotransposons, they cannot carry out reverse transcription using an RNA transposition intermediate in the same way as LTR retrotransposons. Those two key components of the retrotransposon are still necessary but the way they are

incorporated into the chemical reactions is different. This is because unlike LTR retrotransposons, non-LTR retrotransposons do not contain sequences that bind tRNA.

They mostly fall into two types – LINEs and SINEs. SVA elements are the exception between the two as they share similarities with both LINEs and SINEs, containing Alu elements and different numbers of the same repeat. SVAs are shorter than LINEs but longer than SINEs.

# Long interspersed nuclear elements (LINEs):

- Long interspersed nuclear elements (LINEs) (also known as long interspersed nucleotide elements or long interspersed elements) are a group of non-LTR (long terminal repeat) retrotransposons that are widespread in the genome of many eukaryotes.
- They make up around 21.1% of the human genome. LINEs make up a family of transposons, where each LINE is about 7,000 base pairs long.

- LINEs are transcribed into mRNA and translated into protein that acts as a reverse transcriptase.
- The reverse transcriptase makes a DNA copy of the LINE RNA that can be integrated into the genome at a new site.
- LINEs are implicated in many genetic disease in humans and other eukaryotes.
- The only abundant LINE in humans is LINE1/L1, that are active in the human genome and found in all mammals except megabats.

- The first description of an approximately 6.4 kb long LINE-derived sequence was published by J. Adams *et al.* in 1980.
- LINEs are grouped into five main groups, called L1, RTE, R2, I and Jockey, which can be subdivided into at least 28 clades.
- A historic example of L1-conferred disease is Haemophilia A, which is caused by insertional mutagenesis.

## Short interspersed nuclear elements (SINEs):

- Short interspersed nuclear elements (SINEs) are non-autonomous, non-coding transposable elements (TEs) that are about 100 to 700 base pairs in length.
- They are a class of retrotransposons, DNA elements that amplify themselves throughout eukaryotic genomes, often through RNA intermediates.
- SINEs compose about 13% of the mammalian genome.

- The internal regions of SINEs originate from tRNA and remain highly conserved, suggesting positive pressure to preserve structure and function of SINEs.
- While SINEs are present in many species of vertebrates and invertebrates, SINEs are often lineage specific, making them useful markers of divergent evolution between species.
- Copy number variation and mutations in the SINE sequence make it possible to construct phylogenies based on differences in SINEs between species.

- Like LINEs, SINEs are also implicated in certain types of genetic disease in humans and other eukaryotes.
- SINEs are classified as non-LTR retrotransposons because they do not contain long terminal repeats (LTRs).
- There are three types of SINEs common to vertebrates and invertebrates: CORE-SINEs, V-SINEs, and AmnSINEs.
- Alu elements, short-interspersed nuclear element of about 300 nucleotides, are the most common SINE in humans, with >1,000,000 copies throughout the genome, which is over 10

percent of the total genome; this is not uncommon among other species.

- There are >50 human diseases associated with SINEs.
- When inserted near or within the exon, SINEs can cause improper splicing, become coding regions, or change the reading frame, often leading to disease phenotypes in humans and other animals.
- Insertion of Alu elements in the human genome is associated with breast cancer, colon cancer, leukemia, hemophilia, Dent's disease, cystic fibrosis, neurofibromatosis, and many others.

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# THANK YOU

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