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TRANSPOSONS IN GENOME SIZE VARIATION

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ABSTRACT
Genome size is constant to a species, differences in species is brought out by genome size variation. Changes in the genome size is mainly by the changes in the copy number driven by the various repetitive sequences. Transposons are the kind of repetitive sequences put forth the genome size variation by its mobility characteristics within the genome causes a great variation even closely related species. Studying the structure and functions of transposons is necessary to understand the mechanism of genome size variation and thereby it is possible to study the species relatedness in the evolutionary history.

Introduction

Genome size is one of the intrinsic characteristics, being considered as a constant species specific character that can help to explain relationships between species. There is a tremendous variation in DNA content exists, even within closely related species. Although flowering plants vary tremendously in nuclear DNA content, most of this variation is not associated with differences in gene number or gene size. It is mainly attributed to changes in repetitive sequence of altering copy number. In plants, genome size ranges from 63 Mbp in *Genlisea margaretae* to 124,852 Mbp in *Fritillaria assyriaca* (Greilhuber *et al*., 2006), a 2000 fold difference. The diversity of plant genomes is manifested through a wide range of interacting forces not only by internal genomic factor but also the external environmental forces which drives variation in genome size by changing chromosome number and genome size by expansion and contraction mechanisms (Leitch and Leitch, 2013; Poggio *et al*., 2014). One among the important genome expansion mechanism is transposable element proliferation. Transposable elements (TE_s) simply called as "transposons" was first reported experimentally by Barbara McClintock in maize in 1938. These elements are associated with kernel colour change, due to their ability to change from one position of the genome to another that is why these elements are known as "jumping gene". While the course of their movement it will alter the genome size and it is considered as the foremost driving force in the genome size variation in flowering plants.

Transposable elements (TEs) proliferation

TEs are stretches of DNA that are competent to integrate into new positions in the genome, that are competent to increase their copy number over time and that rely on one or more enzymatic function provided by an autonomous element. TEs are the single largest component of the genetic material of most eukaryotes. They account for at least 45% of the human genome and 50–80% of some grass genomes. TEs were discovered in maize by Barbara McClintock more than a half century ago as the genetic agents that are responsible for

the sectors of altered pigmentation on mutant kernels in maize. This discovery and the ensuing characterization of the genetic properties of TEs led to her being awarded a Nobel Prize in 1983, after TEs had been documented in the genomes of *Drosophila melanogaster*, yeast (*Saccharomyces cerevisiae*), *Escherichia coli*, *Caenorhabditis elegans*, *Drosophila melanogaster*, yeast (*Saccharomyces cerevisiae*), *Escherichia coli*, *Caenorhabditis elegans* and humans. Waves of expansion and contraction in numbers of TEs can result in dramatic differences in the overall architecture of the genomes of even closely related plant species. TEs make up the majority, probably the vast majority, of all plant DNA.

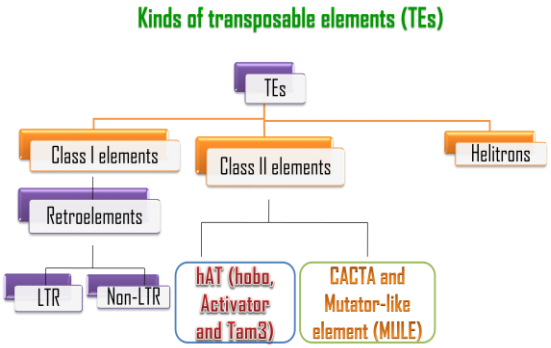

Fig. 1. Classification of transposons

Characterizing TEs

The first element that was recognized to be transposable was a site of chromosome breakage in maize and, as such, was named *Dissociation* (*Ds*). *Ds* could transpose or break chromosomes only in the presence of another genetic locus, called *Activator* (*Ac*), which could also promote its own transposition. Together, *Ac* and *Ds* constitute a TE family that includes autonomous (*Ac*) and non-autonomous (*Ds*) elements. Similar TE families were later found to underlie unstable mutant phenotypes in other plants (for example, snapdragon, petunia, soybean and sorghum) and in animals (such as *Drosophila* and *C. elegans*). Analyses of the growing database of plant genomic sequence soon revealed that plant genomes harbor a huge diversity of TEs, and those two types — MITEs and LTR retrotransposons have made important and recent contributions to plant genome organization and evolution. Analysis of repetitive DNA in humans, in which two TE families, *L1* and *Alu*, were found to be present at very high copy number. *L1* and *Alu* are members of large groups called long interspersed nuclear elements (LINEs) and short interspersed nuclear elements (SINEs), respectively, and are classified as non-LTR retrotransposons. Together, these two families make up more than one quarter of the human genome. LINEs and SINEs are also present in plant genomes, where they have attained very high copy numbers in some species. Although there are many kinds of transposable elements (TEs), they fall into a small number of general classes. The three classes of TEs that are present in plant genomes are discussed below.

Class I elements: These elements are retrotransposons. In plants, they are the most common class of element and can make up the bulk of many genomes. Retroelements transpose via a 'copy-and-paste' mechanism in which mRNA transcribed from the element by RNA polymerase II (RNA Pol II) is converted into a cDNA by reverse transcription and then integrated by an integrase enzyme at a new position in the genome. Class I elements are further divided into long terminal repeat (LTR) retroelements and non-LTR retroelements, which differ in the mechanism of integration.

Class II elements: These elements transpose via a 'cut-and-paste' mechanism in which the element is physically excised from the chromosome and reintegrated at a new location, a process that involves the transposase enzyme encoded by the TE. In plants, the most common class II elements include members of the hAT (*hobo*, *Activator* and *Tam3*), *CACTA* and *Mutator*-like element (MULE) super families.

Helitrons

Helitrons are a class of elements that are thought to transpose via a 'rolling circle' mechanism. This process involves nicking at the *Helitron* terminus, followed by strand invasion, DNA synthesis, strand displacement and resolution of a heteroduplex by DNA replication. If the initial DNA synthesis and strand displacement proceeds farther than the end of the *Helitron*, flanking sequences can be co-replicated.

Structural and functional changes caused by transposable elements

- Knockout of function
- Introduction of new functions
- Changes in the structure of genes
- Mobilization and rearrangement of gene fragments and
- Epigenetic silencing of genes.

Differential proliferation of TEs explains the majority of genome size differences among species. In the wild rice species *Oryza australiensis*, for example, amplification of three TEs accounts for a 2-fold increase in genome size. Similarly, evidence from *Gossypium* (cotton) indicates that the majority of the threefold range in diploid genome sizes may be accounted for by amplification of the *Gorge3 gypsy* element in the larger genomes. These and other studies also have revealed that different types of TEs (e.g., *copia* or *gypsy* LTR retrotransposons) or different subfamilies of a single type (e.g., *copia* LTR retrotransposons) may episodically proliferate at different times. The result is that lineages experience periodic quantum gains in genome size that are likely controlled by myriad factors (e.g., epigenetics, recombination rate, etc.), which vary by element type/family and, presumably, in response to genomic and environmental factors (e.g., hybridization or environmental stress).

Conclusion

Transposable element proliferation has the importance driven mechanism in the genome size variation. Hence studying this helps to understand species relatedness in the evolution and there introgressing gene of interest into a particular species.

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