

HELITRON-LIKE ELEMENTS MEDIATE EXTENSIVE GENE DUPLICATIONS AND EXON SHUFFLING IN MAIZE

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Widespread occurrence of DNA sequence non-collinearity among maize inbreds has recently been reported. This has been attributed to recent LTR-retrotransposon insertions and to the presence/absence of genes or gene fragments. The molecular mechanism responsible for the gene content differences has not been elucidated, even though the evidence points to insertion rather than deletion events. Here we report a whole-genome comparison of gene content in allelic maize BAC contigs from two maize inbreds and show that gene presence/absence polymorphisms are very frequent in the maize genome. The allelic differences may involve as many as 10000 genes when genomes of two common maize inbreds are compared. The termini of eight out of nine of the genic insertions we examined in detail share the structural hallmarks of the rolling-circle transposon class named helitron. Closely related genic insertions equipped with helitron termini are found in multiple genomic locations, suggesting they are non-autonomous transposons. They usually contain multiple gene-derived fragments in the same orientation. These sequences can be transcribed across segments derived from different genes, suggesting their possible participation in exon shuffling and evolution of novel protein functions. We also identified putative autonomous helitron elements in maize genomic sequences. The expression of helitron ORF coding for a protein with helicase and replication initiator activities was confirmed by RT-PCR on mRNA from seedlings in 18 inbred lines. Analysis of expressed sequences indicates that more than one helitron subfamily is expressed and that different lines express different subfamilies.

Thanks to their ability to produce a very diverse set of non-autonomous elements helitrons are responsible for the frequent duplicative insertion of gene segments into new locations in maize and for a profound reshuffling of the maize genome, leading to an unprecedented gene content diversity within a species as well as between maize and related species.