

CIS-ACTING REGULATORY VARIATION AFFECTING MAIZE GENE EXPRESSION

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Mutations in *cis*-regulatory DNA sequences, which affect gene expression levels, have been proposed to influence quantitative variation, disease susceptibility, and to be a primary substrate for the evolution of the species.

Our recent studies of allele-specific expression in a random set of non-imprinted maize (*Zea mays*) genes in F1 hybrids demonstrated that more than 70% of the genes show allelic differences in expression of at least 1.5 fold due to *cis*-regulatory variation and that these differences are tissue specific. We further examined allele-specific expression under different types of abiotic stress. Stress induced significant imbalance in expression ratios for the alleles that initially showed no disparity in expression, and it enhanced alteration in the relative expression ratios for those that originally showed differential expression of the two alleles.

Maize genome is known to harbor especially high nucleotide diversity within single copy regions, including genes, and lack of colinearity in intergenic regions due to the presence of different LTR-transposons which is making it a unique example of within species diversity. To investigate the potential for *cis*-acting variability in genes analyzed we sequenced the 5' flanking regions and ORFs of 5 of them in a sample of 17 inbred lines and assessed nucleotide diversity and linkage disequilibrium. In attempt at identifying *cis*-acting elements responsible for the differences in allelic expression, upstream regions were screened for putative binding sites and phylogenetic footprinting was performed. We also estimated allelic expression imbalance in genes within fully sequenced genomic regions from both alleles to assess the effects of the lack of colinearity in intergenic regions.

Our findings suggest that the *cis*-regulatory variation is a highly common phenomenon in maize and may provide a possible molecular explanation of the heterosis phenomenon. The heterozygous state found in hybrids for many genes may represent a buffering mechanism to improve stress tolerance and more in general to ensure optimal gene expression in a wide variety of environmental conditions.