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UNDERSTANDING ALLELIC VARIATION IN MAIZE: TOWARDS DISCOVERING OF THE MOLECULAR BASES OF HETEROSIS

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Heterosis, or hybrid vigor, is the increased performance of hybrid relative to the parents and is a result of the variation that is present within a species. Maize hybrids exhibit high levels of heterosis and as such provide an excellent system for the study and understanding of the phenomenon. Intraspecific comparison of sequences and expression levels in maize have documented a surprisingly high level of alleic variation, which includes variation in the content of genic fragments, variation in repetitive elements surrounding genes, and variation in gene expression levels. A major unresolved question is how the combined allelic variation and interactions in a hybrid give rise to heterotic phenotypes.

Our research is focused on the analysis of gene expression in the hybrid and inbred parents at the allele specific-level, which, as compared to solely measuring total transcript amount without allelic differentiation, may provide a different perspective and understanding of gene regulation and the molecular basis of heterosis. By quantifying allele-specific transcript levels we estimated that 70% of maize genes show differences in expression of at least 1.5 fold due to *cis*-regulatory variation. Allelic expression varied spatially in different tissue types and temporally as a consequence of environmental changes. Some of the allelic imbalances revealed expression overdominance or bidirectional expression, meaning that either allele was overexpressed depending on tissue, or growth phase or/and environmental conditions. In an attempt of identifying a source of *cis*-acting variation we assessed the effects of the maize intraspecific structural genome diversity on gene expression. Transposable elements located in or near genes could affect expression through the donation of transcriptional regulatory signals as well as through epigenetic silencing. To test the latter hypothesis we searched for the presence of both sense and antisense co-transcripts and examined the methylation state of the analysed genes and their promoters.

Our results show that *cis*-regulatory variation is very common in maize and that the repetitive sequences can influence gene transcription. It has been proposed that allele expression variability may explain phenotypic variability within species, and that the relatively modest changes in gene activity are a key feature of many speciation events. Our findings may thus uncover a possible mechanism that connects genomic plasticity to the evolution of whole organisms. In addition, *cis*-regulatory variation and observed expression overdominance may provide a possible molecular explanation of the heterosis phenomenon in maize. Expression overdominance could provide heterozygotes with a wider range of transcription modulation than the corresponding homozygotes, which in turn could determine both flexibility and stability.