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Current Opinion in Genetics & Development

Potential roles for transposable elements in creating imprinted expression

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Changes in gene expression can have profound effects on phenotype. Nature has provided many complex patterns of gene regulation such as imprinting. Imprinted genes exhibit differences in the expression of the maternal and paternal alleles, even though they reside in the same nucleus with access to the same trans-acting factors. Significant attention has been focused on the potential reasons that imprinted expression could be beneficial and stabilized by selection. However, less attention has focused on understanding how imprinted expression might arise or decay. We discuss the evidence for frequent turnover of imprinted expression based on evolutionary analyses in plants and the potential role for transposable elements (TEs) in creating imprinted expression patterns.

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Current Opinion in Genetics & Development 2018, **49**:8–14

This review comes from a themed issue on **Genome architecture and expression**

Edited by **Kathleen Burns** and **Damon Lisch**

https://doi.org/10.1016/j.gde.2018.01.008

Introduction

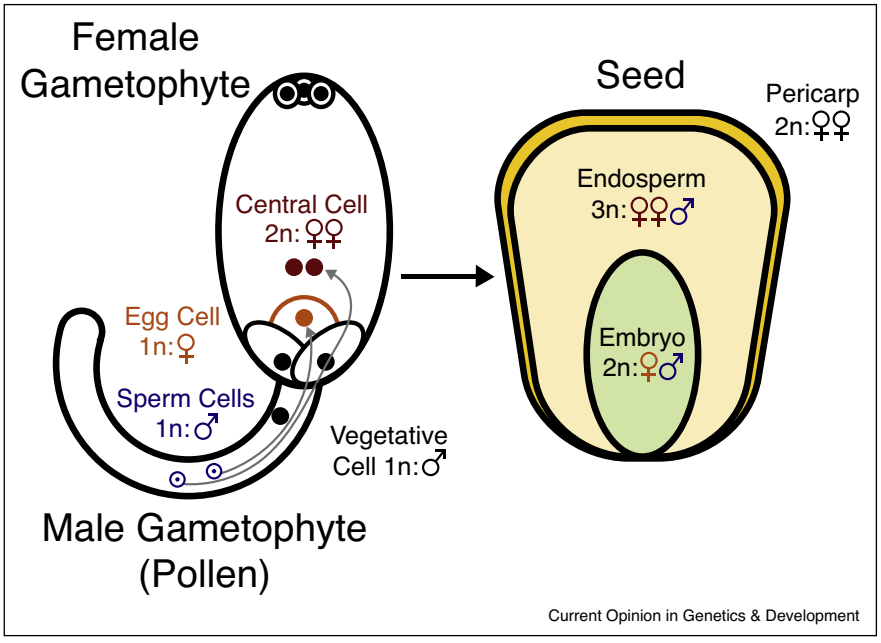
Genomic imprinting, the phenomenon where an allele is preferentially expressed from one parental genome relative to the other, has been extensively described in both angiosperm [1] and mammalian lineages [2]. While imprinted expression has the potential to create selective advantage [3–5], evolutionary theories proposed to describe the function of imprinting generally do not effectively explain how imprinting may arise and why imprinting status is poorly conserved. We will primarily focus on recent studies from plants that provide insights into the factors that create imprinted expression at a particular locus. In plants, which undergo double fertilization to produce a diploid embryo and triploid endosperm (Figure 1), imprinting is primarily restricted to the endosperm. Studies in several species have identified dozens to hundreds of imprinted genes in the endosperm [6–8,9••,10,11,12••,13–15], while parent-of-origin biased gene expression in the embryo is transient [16–18] and involves enough mechanistic differences to endosperm imprinting to make it difficult to consider them simultaneously [19]. Since endosperm is formed through the fusion of the maternal homodiploid central cell with a paternal haploid sperm cell, the expected ratio of maternal to paternal transcripts is 2:1, corresponding to the relative dosage of the two genomes (Figure 1). Imprinted genes have expression that significantly deviates from this ratio, with paternally expressed genes (PEGs) demonstrating higher than expected expression of the paternal allele and maternally expressed genes (MEGs) demonstrating higher than expected expression of the maternal allele. Imprinted expression can be established either through activation or suppression of expression of one of the parental alleles. Developmental analyses of imprinted genes reveals that many MEGs are expressed preferentially in the endosperm while PEGs include examples of expression in multiple tissues in addition to endosperm-specific expression [6,12••,20], which may suggest distinct regulatory mechanisms for MEGs and PEGs.

Despite the recent advances elucidating mechanisms of chromatin and epigenetic regulation influencing the relative expression of the maternal and paternal alleles of imprinted genes, it is not clear how this regulation becomes targeted (established) to certain genes but not others. It has been suggested that the origin of imprinted expression may be due to regulation of transposable elements (TEs) rather than genes per se [14,21–24], with the dynamics that regulate these transposons during reproduction potentially resulting in parent-of-origin specific regulation of nearby genes [1]. We will begin by reviewing the evidence for parent-specific chromatin changes at TEs during plant reproduction. Then we will summarize comparative data on imprinting conservation between plants, where the variability in imprinting status parallels the high rate of variability for TE insertions between and within plant species. Finally, we will focus on the specific evidence that supports a role of TEs in imprinting and summarize the properties the TE would need to provide in order to condition imprinted expression.

Chromatin dynamics during reproduction

During reproduction, plant genomes undergo significant chromatin changes [25–28]. Numerous studies have

Figure 1



Double fertilization is required for angiosperm seed development. The male gametophyte, pollen, carries two sperm cells to the female gametophyte. One sperm fuses with the haploid egg cell to form the diploid embryo while the second fuses with the central cell to form the triploid endosperm. The mature seed is comprised of embryo (2N), endosperm (3N), and maternal pericarp tissues, with coordinated development of each tissue required to produce a viable seed.

documented the changes in DNA methylation that occur in specific cell types or tissues during plant reproduction [26,27,29]. DNA methylation is one source of epigenetic control of gene expression and transposon silencing, and can be stably inherited across generations. During the development of the *Arabidopsis* microspore to form the pollen grain, the vegetative cell (Figure 1) undergoes substantial DNA demethylation due to reduced expression of *DECREASE in DNA METHYLATION1* (*DDM1*) and activity of the *DEMETER* (DME) demethylase that frequently targets TEs [30,31]. The de-repression of TEs in the vegetative cell is followed by generation of siRNAs that can travel to the sperm cells and reinforce silencing of transposons in the germline [31,32]. During female gametophyte development, active demethylation by DME occurs throughout the genome of the central cell [33•,34]. This hypomethylation persists during endosperm development at the maternal alleles for many loci, providing the local chromatin differentiation that is required to establish imprinted expression. Many of the regions targeted for demethylation in the central cell (and inherited at the maternal allele in endosperm) correspond to TEs [13,21,34,35]. The endosperm is a terminal tissue but has close contact with the embryo and could provide signals for immunization against TEs, similar to the relationship between the pollen vegetative nucleus and sperm cells. There is increased RNA-directed DNA methylation (RdDM) activity in embryo at loci that correspond to hypermethylated loci in the endosperm [36•,37•]. Epigenetic differentiation of parental genomes is reinforced by the polycomb repressive complex 2 (PRC2) through deposition of H3K27me3 at some of the hypomethylated loci. In endosperm, PRC2 is targeted to DME targets, leading to maternal-specific H3K27me3 at many PEGs [7,28,38••,39]. In contrast to leaves where H3K27me3 is primarily localized to euchromatic regions, endosperm has an enrichment of H3K27me3 in pericentromeric regions, with significant overlap with certain transposons [38••]. There is also evidence that additional histone modifications may be associated with maternal or paternal alleles at imprinted loci [40•]. Together, these studies document dynamic changes to chromatin and DNA methylation at transposons during plant reproduction and provide an explanation for the parent-of-origin specific differences in chromatin, especially at TEs, observed in endosperm tissue.

High rates of turnover for imprinted expression patterns

There is substantial variation for the targets of imprinting status both between and within plant species. The application of next-generation sequencing in hybrid endosperm tissue has allowed for the analysis of allelic gene expression in a high-throughput manner and has provided catalogs of imprinted genes in numerous plant species [9••,10,11,12••,14,15,41,42]. There are some