



Review

Paramutation in maize and related behaviors in metazoans



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ABSTRACT

Paramutation refers to both the process and results of *trans*-homolog interactions causing heritable changes in both gene regulation and silencing abilities. Originally described in plants, paramutation-like behaviors have now been reported in model metazoans. Here we detail our current understanding of the paramutation mechanism as defined in *Zea mays* and compare this paradigm to these metazoan examples. Experimental results implicate functional roles of small RNAs in all these model organisms that highlight a diversity of mechanisms by which these molecules specify meiotically heritable regulatory information in the eukarya.

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1. Introduction

Paramutation is a genetic term used to describe both the process and outcome of directed and meiotically heritable changes in both gene regulation and silencing abilities that are influenced by trans-homolog interactions (THI) [1]. Usage of the term is similar to that of classical “mutation” without regard to molecular hallmarks. Unlike mutations, however, paramutations occur in predictable, invariant, and sometimes reversible manners [2].

Deviations from expected Mendelian ratios of trait transmission – such as exclusive inheritance of a dominant trait – are one hallmark of paramutation events. However, pedigree analyses following independent genetic and/or cytogenetic markers distinguish examples of paramutation from other modes of transmission ratio distortion (TRD) [3] including cytoplasmic inheritance [4], preferential chromosome segregations [5], gametic competitions [6], and zygotic lethalties [7]. Dominant inheritance of abnormal leaf morphologies characteristic of the “rogue” phenotype in garden peas is commonly cited as the first published example of paramutation [8,9], without genetic evidence excluding other TRD models.

Several TRD examples occurring in metazoans have paramutation-like properties (see other contributions to this volume) fuelling the opinion that paramutation is also widespread in animals. This review defines the paramutation process as originally described in *Zea mays* (maize) and evaluates the similarities and differences among these metazoan examples. The involvement of small RNAs (sRNAs) in all these cases is specifically highlighted.

2. Definitions

Alleles typically conform to the Mendelian expectation of segregating unchanged from heterozygous condition (Fig. 1A). Deviations from this expectation can be due to various mechanisms including cytoplasmic inheritance (Fig. 1B). Inheritance patterns of seed pigment conferred by the *red1* (*r1*) locus in maize [10] established a definition of paramutation [1] as an invariant, locus-specific, yet parent-of-origin-independent, behavior (Fig. 1C). By following the inheritance of genetic markers from heterozygous individuals, a specific *r1* allele (*R:stippled*; *Rst*) was found to influence heritable properties of the alternate *r1* allele, *R-r:standard*; (*R-r*) [11,12].

2.1. Nomenclature

The terms “paramutable” and “paramutagenic” were applied to *r1* alleles either susceptible to, or capable of facilitating (or inducing) paramutations, respectively [1]. Certain *r1* alleles (*Rst* and *R:marbled*) are strictly paramutagenic [10,13] while others (*R-r*) are paramutable [10]. Another hallmark known as “secondary paramutation” occurs when paramutable alleles become paramutagenic (e.g. *R-r* is transmitted from *Rst/R-r* plants in a paramutagenic form denoted *R-r'*) [11]. This behavior distinguishes paramutations from

other examples of heritable trans-dominant silencing [14]. Alleles neither paramutable nor paramutagenic are termed “neutral”. In some cases, neutral alleles are genetically similar to “nulls” as reversions of *R-r'* to *R-r* occur in both *R-r'/-* hemizygotes and *R-r'/r-g* heterozygotes [15]. These reversion behaviors indicate that THIs are needed for both inducing and stabilizing paramutations.

2.2. Genetic definition of paramutation

Paramutation, as defined by the inheritance behaviors of endogenous *r1* alleles, occurs at three other maize loci (Table 1) [16–19] and at one in *Lycopersicon esculentum* (tomato) [20]. In all cases, results of reciprocal crosses show that there are no parent-of-origin effects. Additionally, all examples show locus-specific behavior: only meiotic products transmitting a paramutagenic allele confer paramutation to offspring (Fig. 1C). This behavior demonstrates a particularly important genetic proof that distinguishes paramutation from other TRD mechanisms. Lastly, all examples show secondary paramutation. Thus, paramutation has a classic genetic definition based on specific inheritance properties of alleles conferring a phenotypic trait.

2.3. Molecular definition of paramutation

There is currently no genomic context of a paramutation in any sense similar to that of a mutation. Recent examples of trans-dominant cytosine methylation and demethylation behaviors occurring in both *Arabidopsis thaliana* and maize hybrids have been cited as potential paramutation examples [21–24] without compelling evidence that such changes in 5-methylcytosine (5meC) patterns are causal to gene regulation. Locus-specificity and secondary paramutation tests also remain to be evaluated. Given these uncertainties, and the potential for confusion between genetic and molecular definitions, it remains prudent to reserve the term paramutation for strictly genetic behaviors related to gene regulation.

3. Paramutation in maize

3.1. Examples

Specific alleles of the *booster1* (*b1*), *purple plant1* (*pl1*) and *pericarp color1* (*p1*) loci all exhibit paramutation (Table 1) similar to that seen at *r1* [16–19,25]. Each locus encodes a transcription factor required for flavonoid biosynthesis, and these paramutable alleles are highly expressed in their respective tissues [26] conferring strong pigment production. High transcription rates are facilitated by specific enhancer sequences: seven tandem repeats (TRs) ~100 kb 5' of the *b1* coding region [27], a genetically defined region 3' of the *pl1* coding region [28], and a promoter-proximal region of a direct repeat flanking the *p1* transcription unit [18]. These high expression states are inherently unstable and can spontaneously change to transcriptionally repressed forms coincident

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