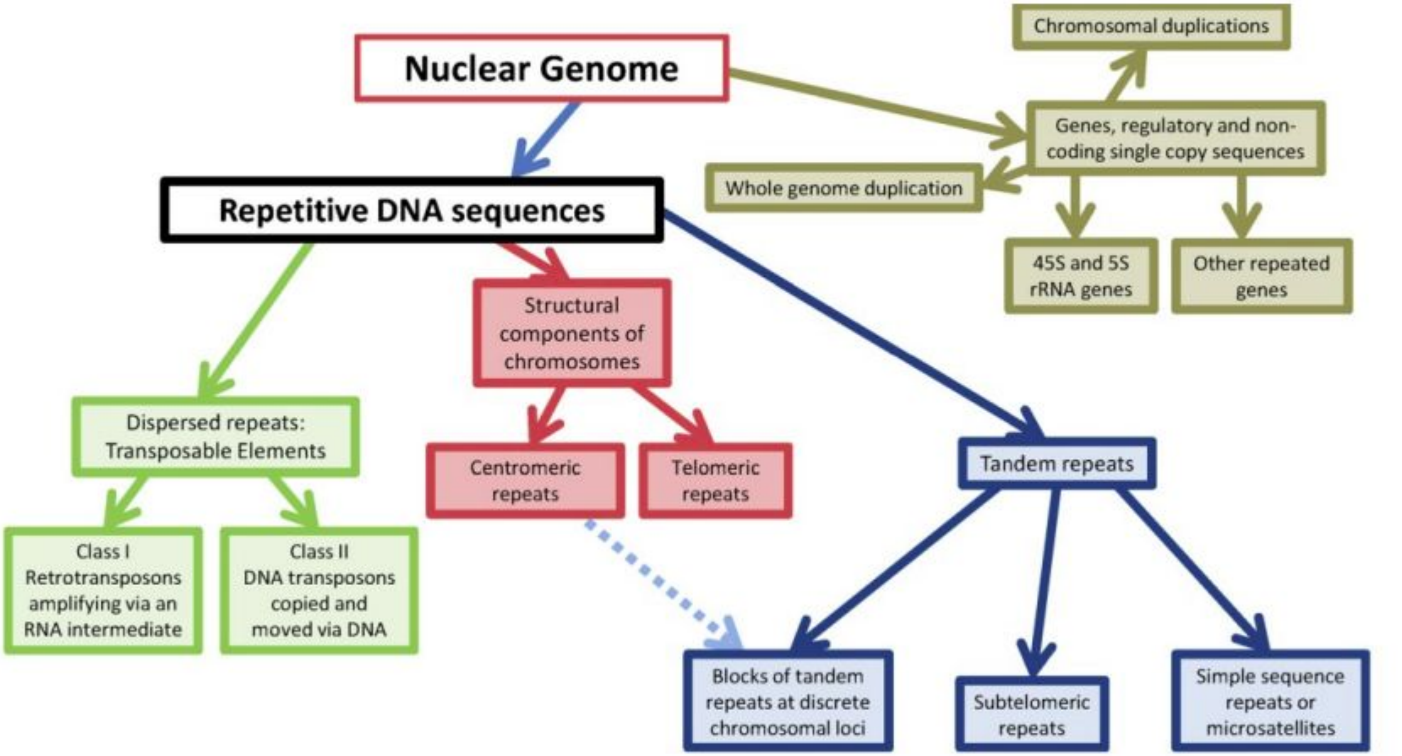
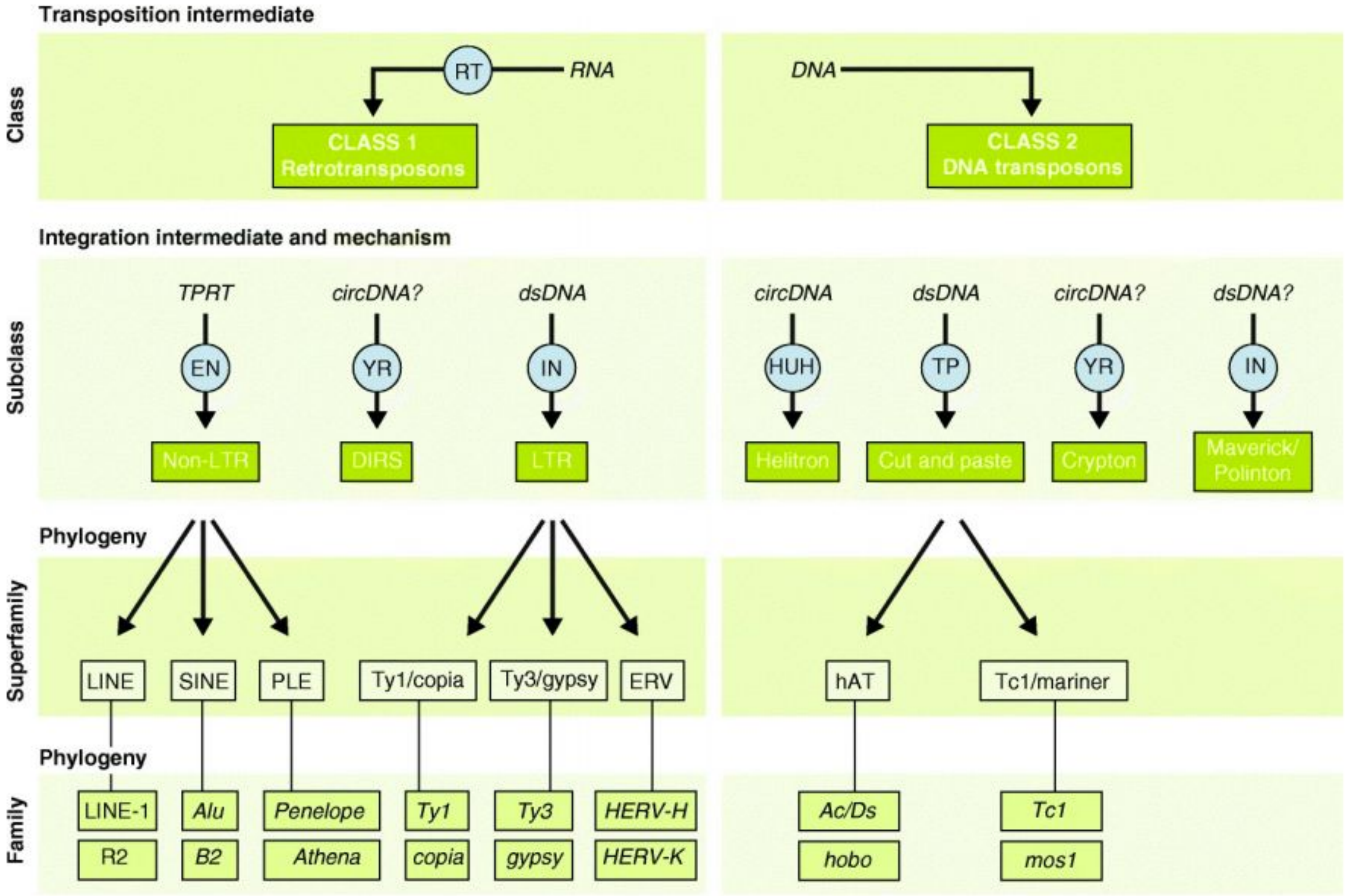
Elementos transponibles

Genómica Evolutiva 2021

Taxonomía de la repetición



Clasificación de ET



Transposition intermediate

Class

RT

RNA

CLASS 1
Retrotransposons

DNA

CLASS 2
DNA transposons

Integration intermediate and mechanism

Subclass

TPRT

EN

Non-LTR

circDNA?

YR

DIRS

dsDNA

IN

LTR

circDNA

HUH

Helitron

dsDNA

TP

Cut and paste

circDNA?

YR

Crypton

dsDNA?

IN

Maverick/
Polinton

Phylogeny

Superfamily

LINE

SINE

PLE

Ty1/copia

Ty3/gypsy

ERV

hAT

Tc1/mariner

Phylogeny

Family

LINE-1

R2

Alu

B2

Penelope

Athena

Ty1

copia

Ty3

gypsy

HERV-H

HERV-K

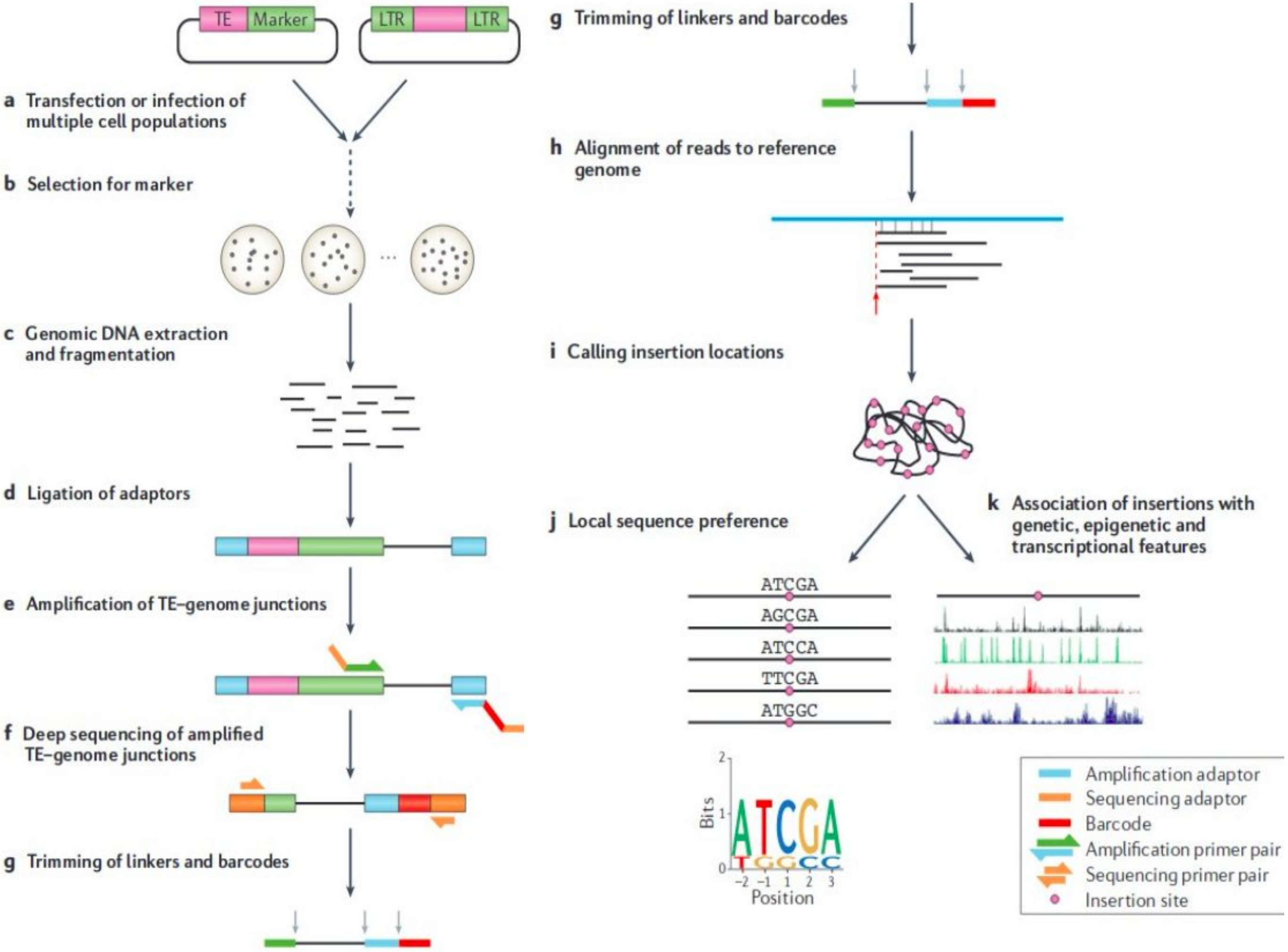
Ac/Ds

hobo

Tc1

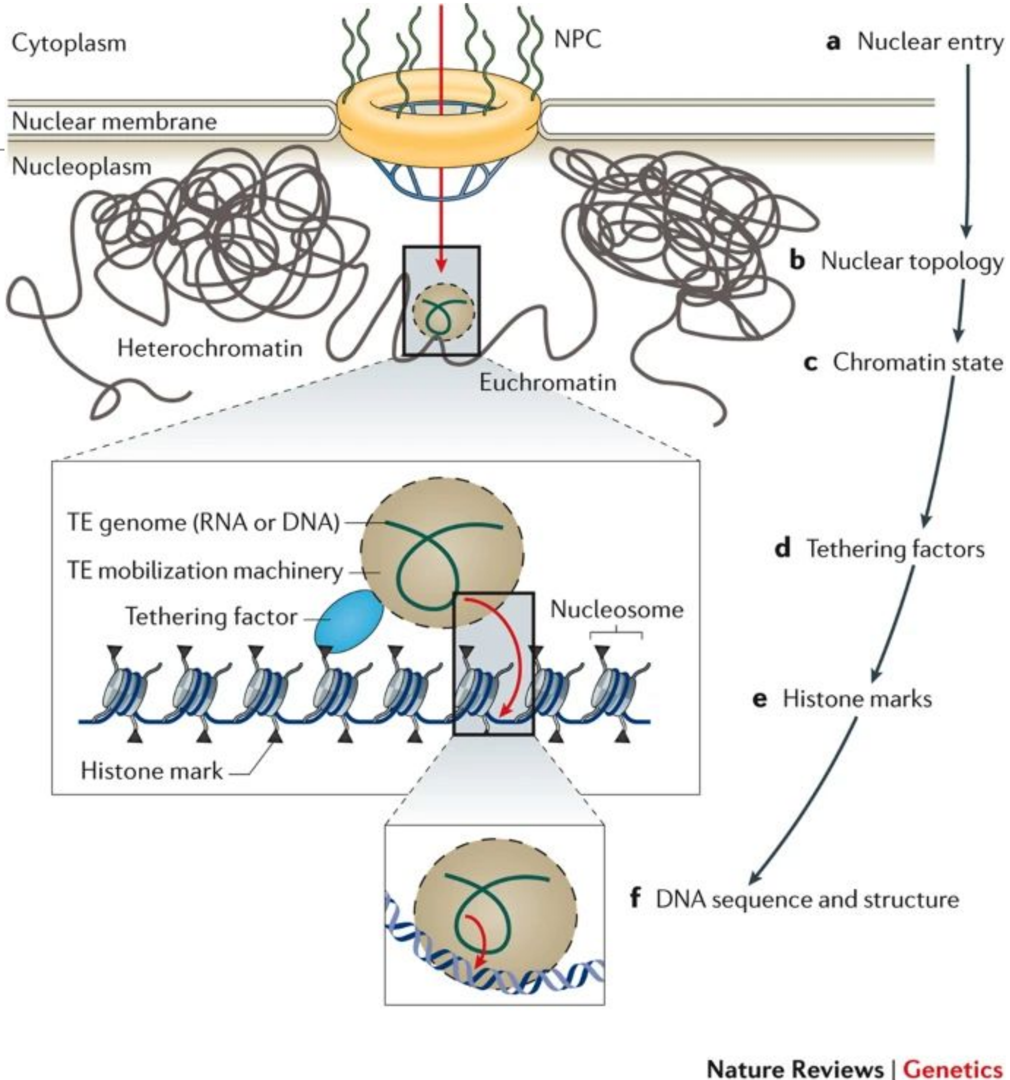
mos1

Sitios de inserción



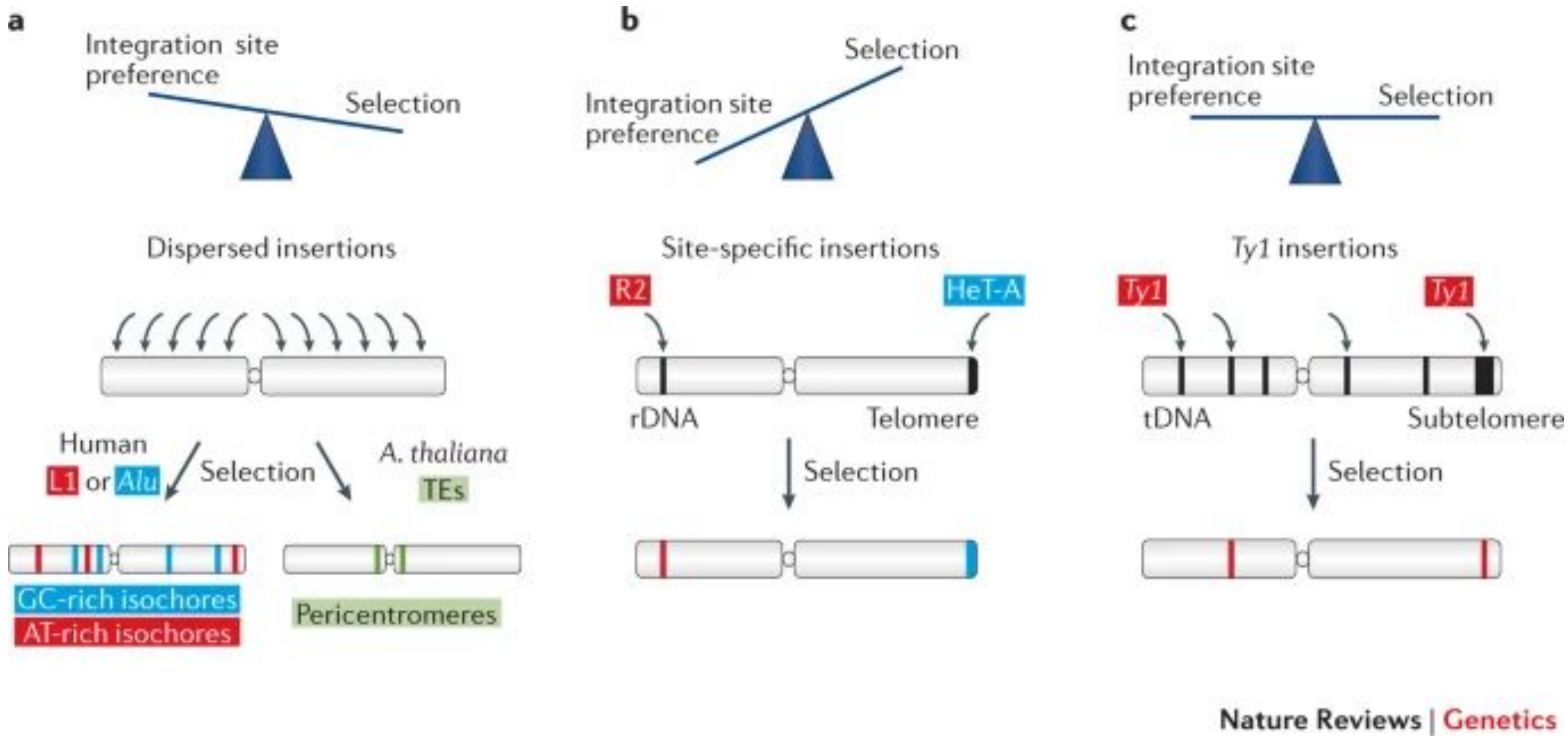
Sultana et al. 2017,https://doi.org/10.1038/nrg.2017.7

Múltiples factores



Sultana et al. 2017,https://doi.org/10.1038/nrg.2017.7

Perspectiva evolutiva



Sultana et al. 2017,https://doi.org/10.1038/nrg.2017.7

Dinámica de un elemento

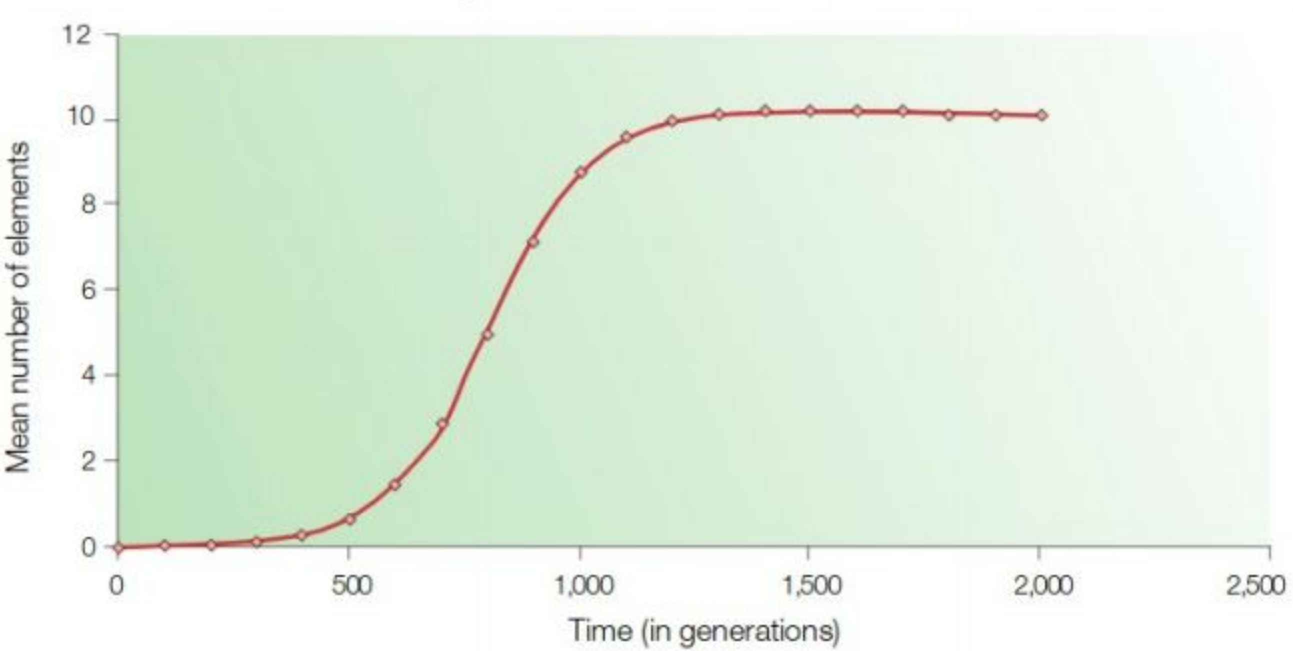
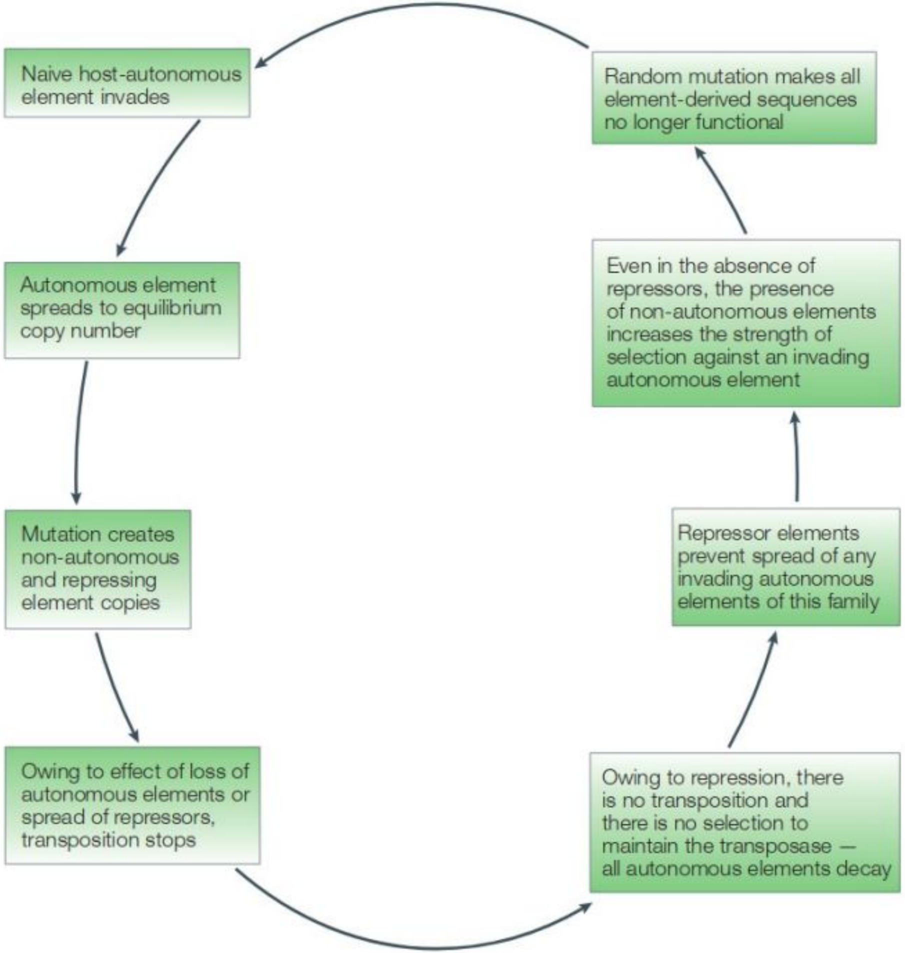


Figure 1 | **The spread of an invading transposable element to an equilibrium created by a balance between the forces of transposition, deletion and selection.** The graph shows the

Song y Schaak, 2018 http://orcid.org/0000-0001-5961-753X

Ciclo de un ET



Naive host-autonomous element invades

Autonomous element spreads to equilibrium copy number

Mutation creates non-autonomous and repressing element copies

Owing to effect of loss of autonomous elements or spread of repressors, transposition stops

Owing to repression, there is no transposition and there is no selection to maintain the transposase — all autonomous elements decay

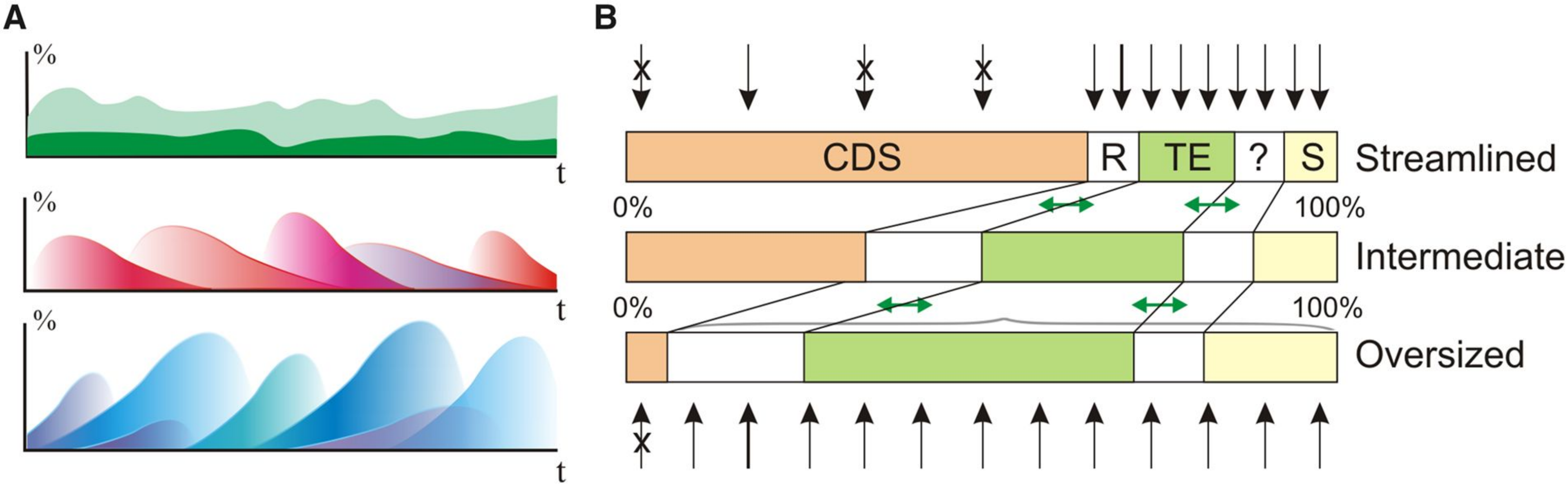
Repressor elements prevent spread of any invading autonomous elements of this family

Even in the absence of repressors, the presence of non-autonomous elements increases the strength of selection against an invading autonomous element

Random mutation makes all element-derived sequences no longer functional

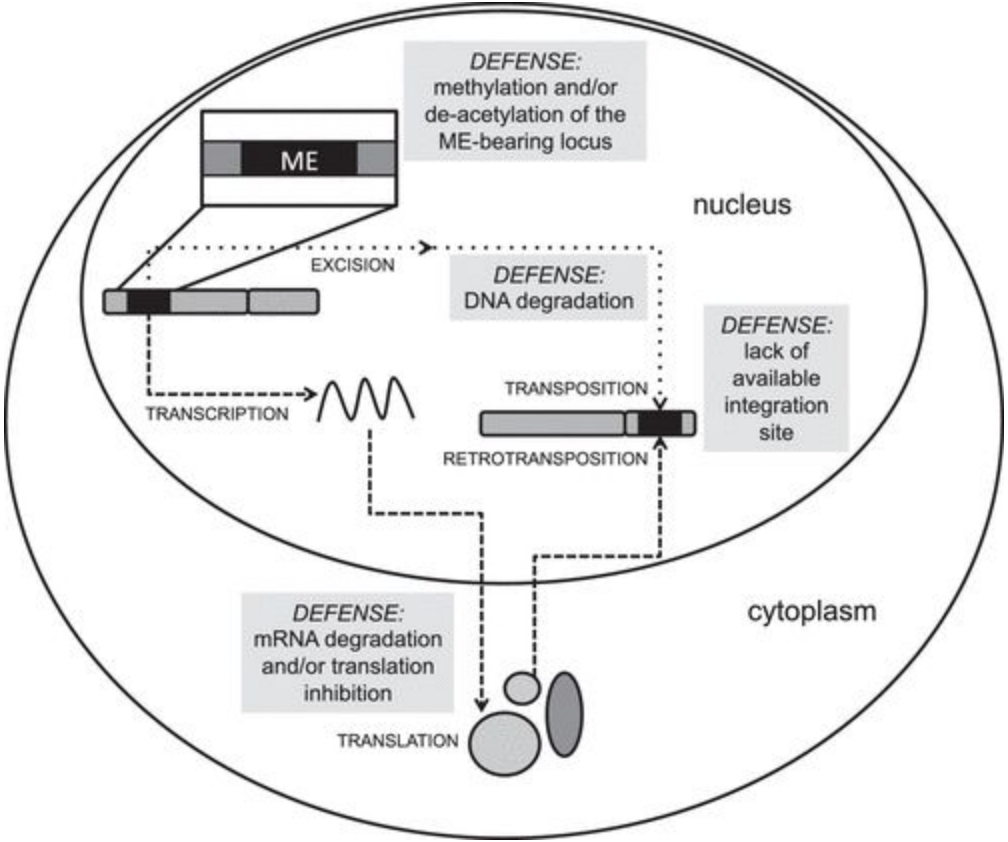
Brookfield, J. F. Y. , 2005. doi:10.1038/nrg1524

Dinámica de un elemento

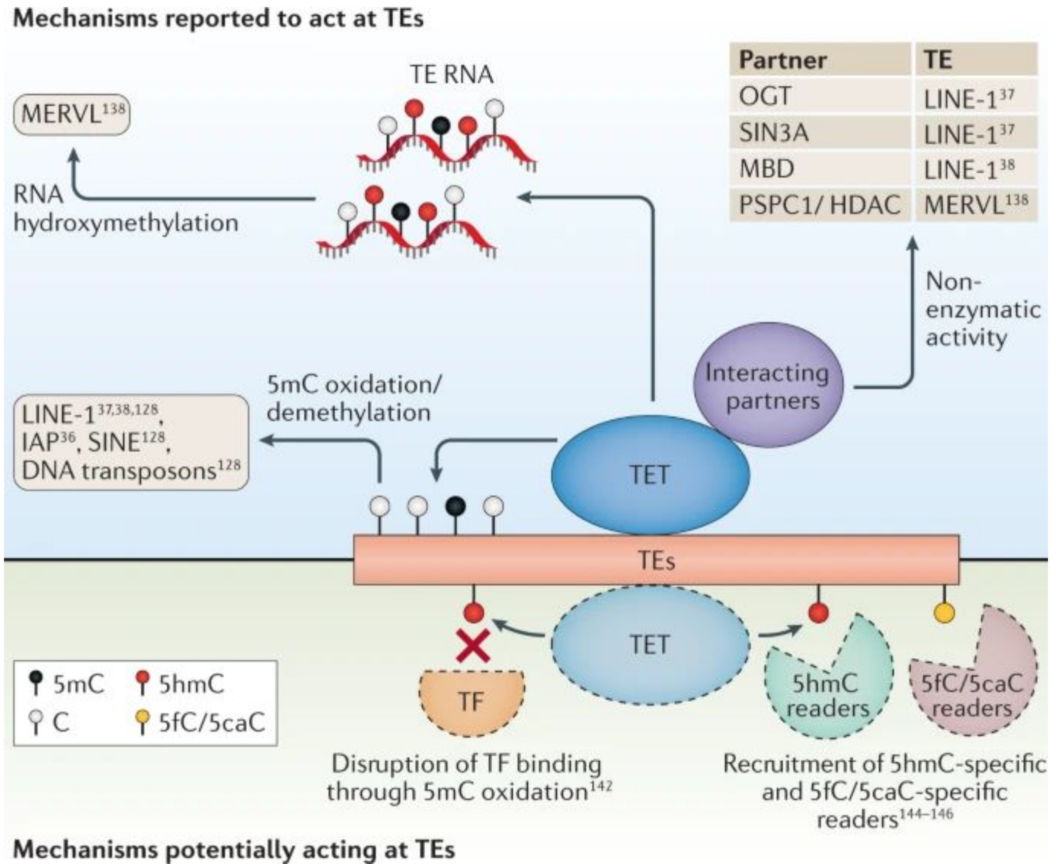


Song y Schaak, 2018 http://orcid.org/0000-0001-5961-753X

El genoma responde



El genoma responde



Partner	TE
OGT	LINE-1[37]
SIN3A	LINE-1[37]
MBD	LINE-1[38]
PSPC1/ HDAC	MERVL[138]

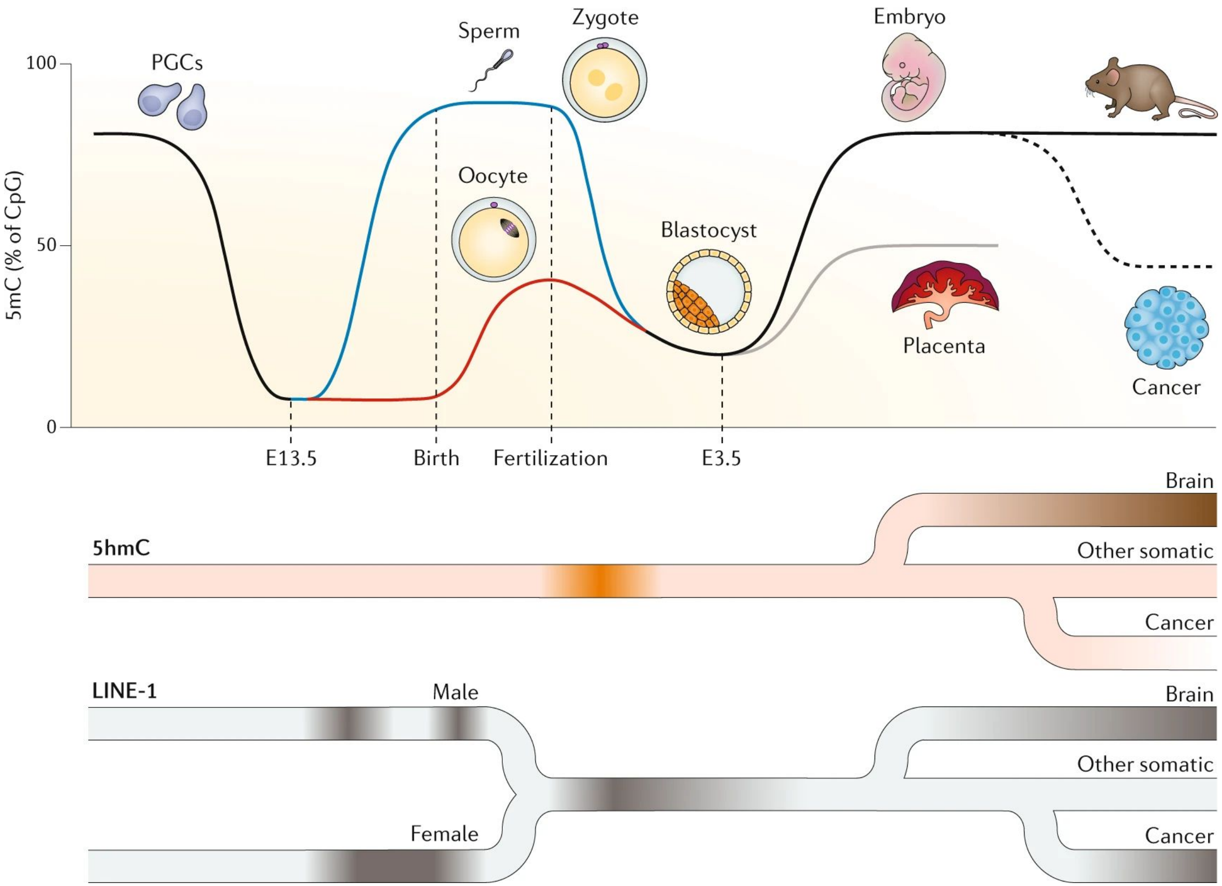
Equilibrios en represión

· The intrinsic expression/repression balance is that the effects of TEs on a host can vary considerably among tissue types and stages of an organism’s life cycle.

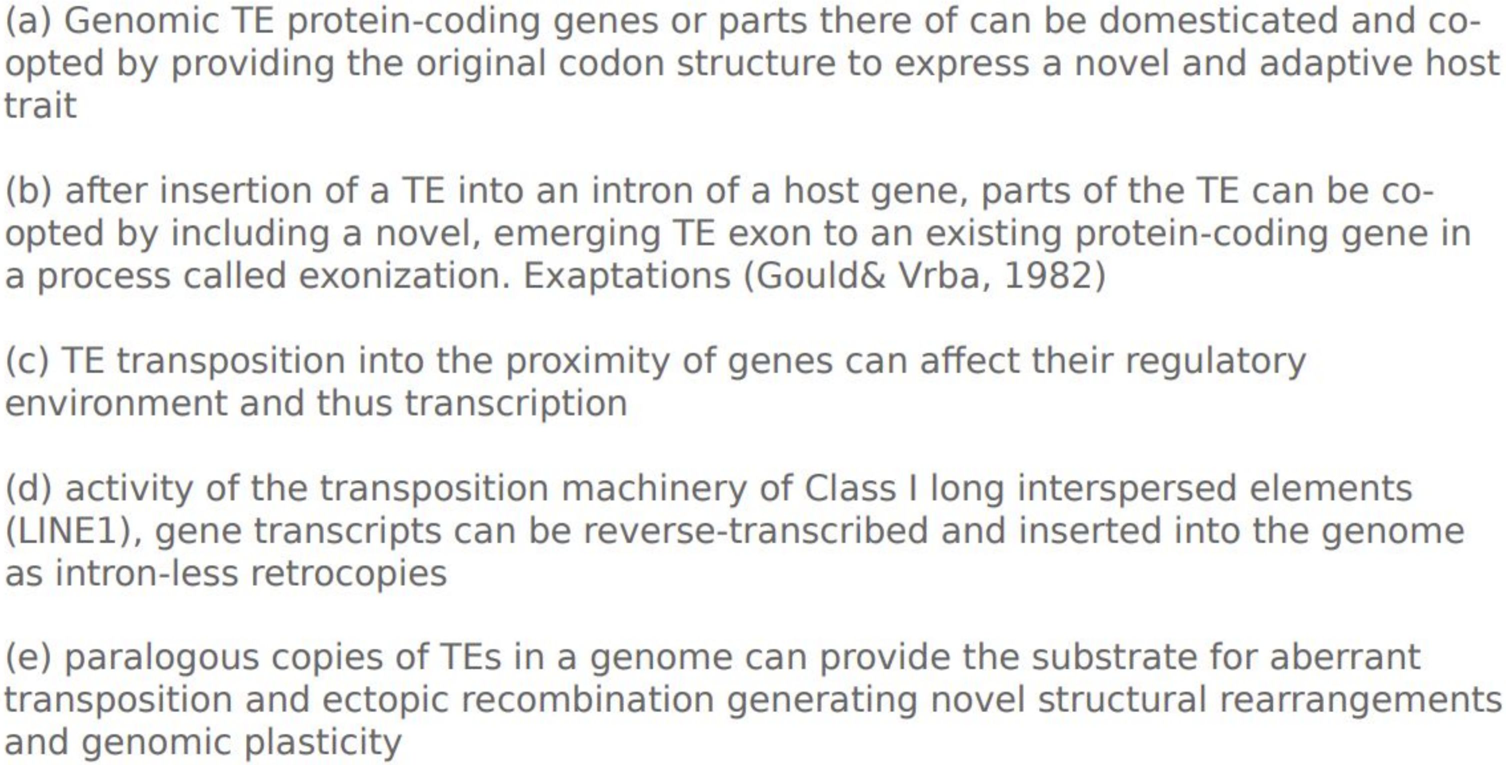
· Ideal scenario is to be expressed and active in the germline, but not in the soma.

· This is indeed observed among many species, with ciliates representing an extreme example of this division—TEs are actively deleted from the somatic macronucleus but retained in the micronucleus, or germline.

Equilibrios en represión



Trayectorias adaptativas



(a) Genomic TE protein-coding genes or parts there of can be domesticated and co-opted by providing the original codon structure to express a novel and adaptive host trait

(b) after insertion of a TE into an intron of a host gene, parts of the TE can be co-opted by including a novel, emerging TE exon to an existing protein-coding gene in a process called exonization. Exaptations (Gould& Vrba, 1982)

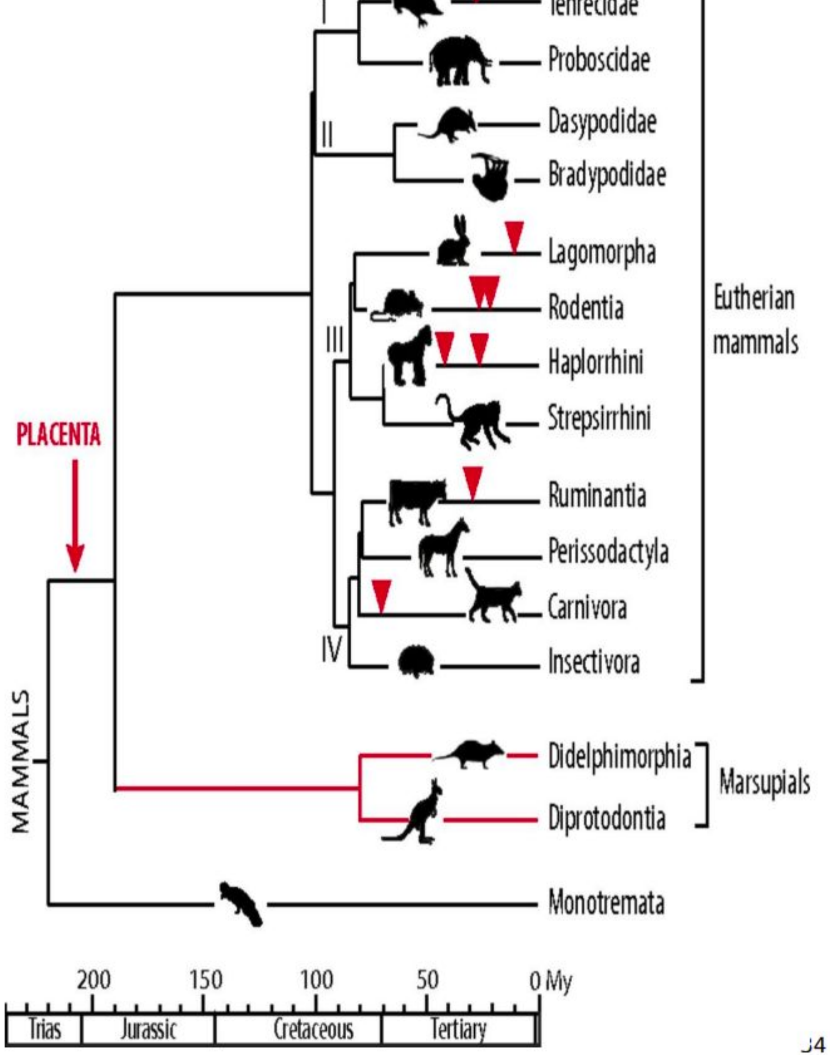
(c) TE transposition into the proximity of genes can affect their regulatory environment and thus transcription

(d) activity of the transposition machinery of Class I long interspersed elements (LINE1), gene transcripts can be reverse-transcribed and inserted into the genome as intron-less retrocopies

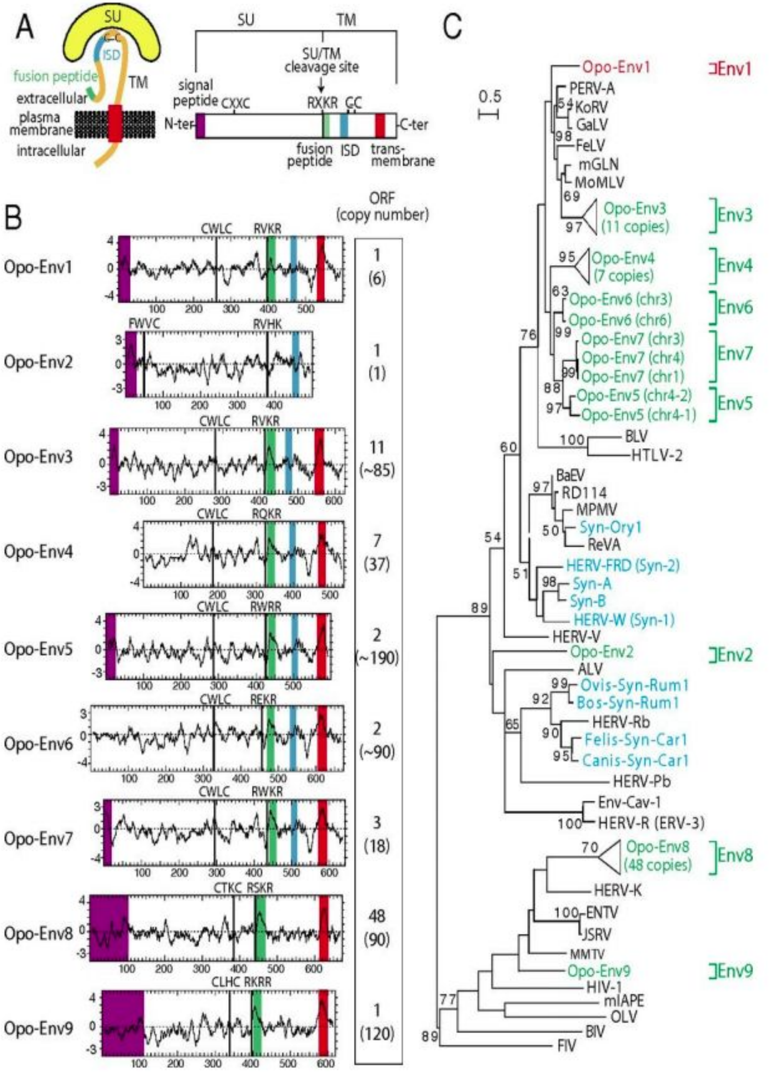
(e) paralogous copies of TEs in a genome can provide the substrate for aberrant transposition and ectopic recombination generating novel structural rearrangements and genomic plasticity

Domesticación

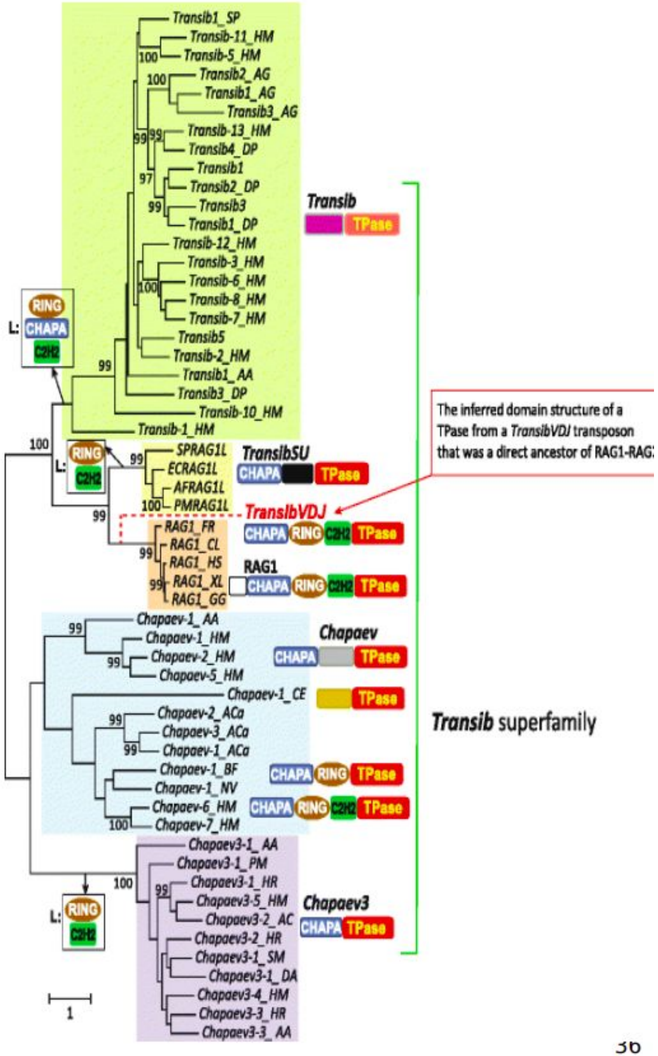
n



Domesticación



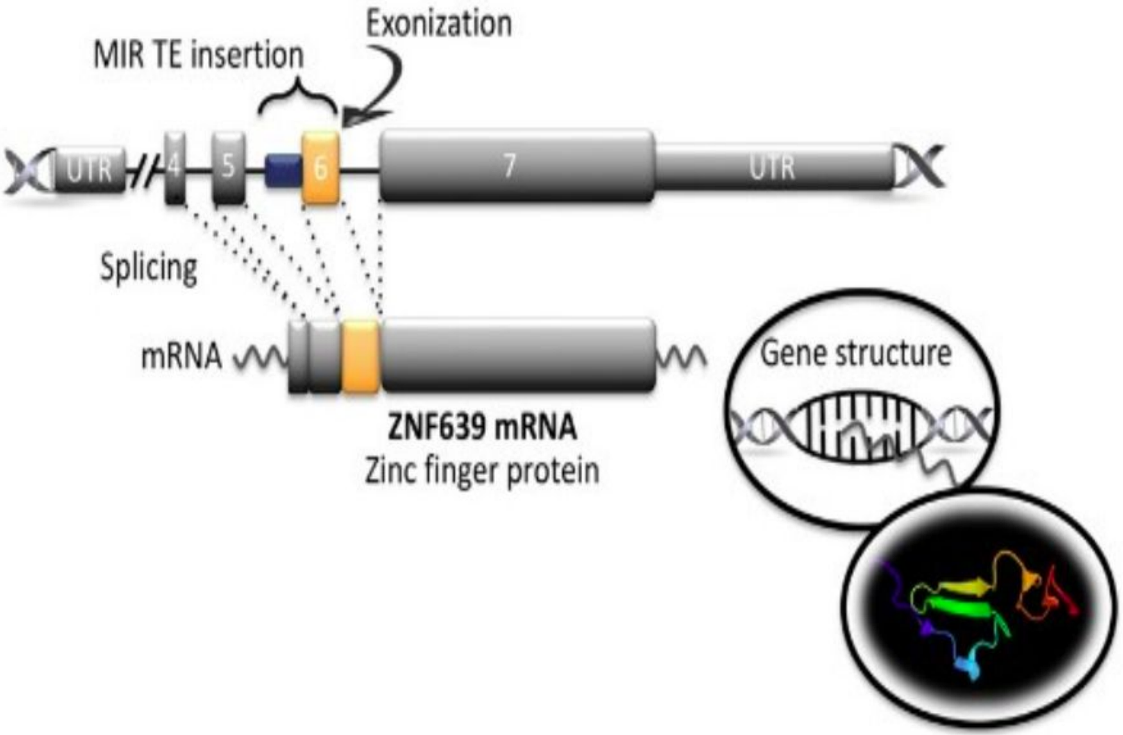
Domesticación



The inferred domain structure of a TPase from a *TransibVDJ* transposon that was a direct ancestor of RAG1-RAG

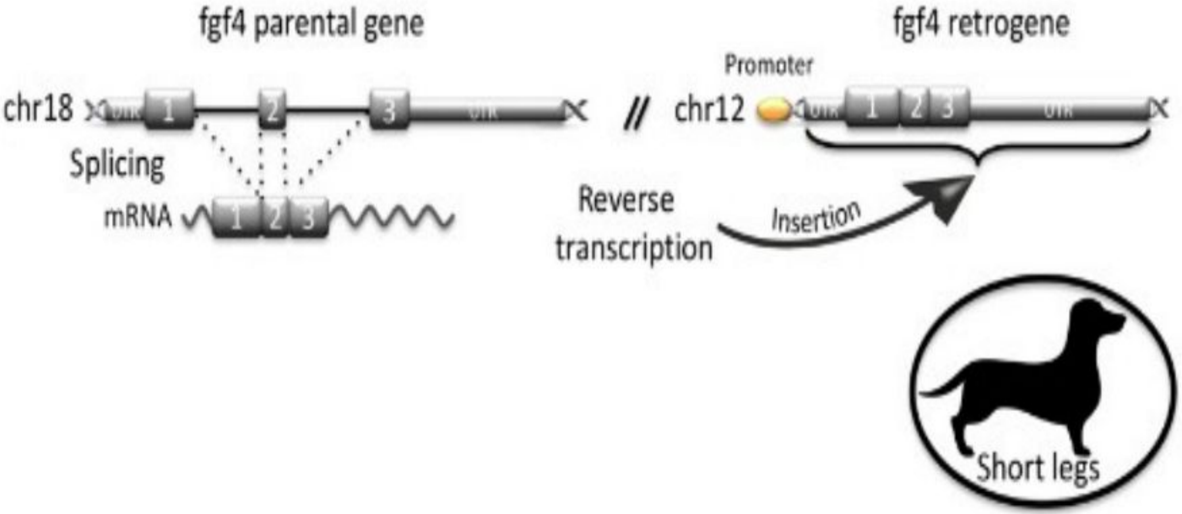
RAG1-RAG2 VDJ Junctions for antibody diversity generation

Exaptación



"features that now enhance fitness but were not built bynatural selection for their current role" (Gould & Vrba, 1982)

Exaptación/Retrogenes



“retropseudogenes” that are separated from their original flanking regulatory units depends on how subsequently arising mutations, selection and drift change the structure and regulation of the retrocopy. Potentially, these events can lead to the evolution of an adaptive, novel trait conferred by a young and functional retrogene (e.g., neofunctionalization or subfunctionalization).

Genome plasticity

· “two-speed genomes.” In these genomes, certain fast-evolving regions are enriched in TEs, have increased frequencies of single-nucleotide and large-scale mutations and often harbour genes related to pathogenicity and host–pathogen interaction

· Probably, the strongest argument in favour of an adaptionist view on the structure and composition of these genomes is that they have evolved convergently in distinct lineages of eu-and prokaryotes

· TE-conferred genome plasticity in different pathogens has been argued to essentially be an adaptation foradaptability

Genome plasticity

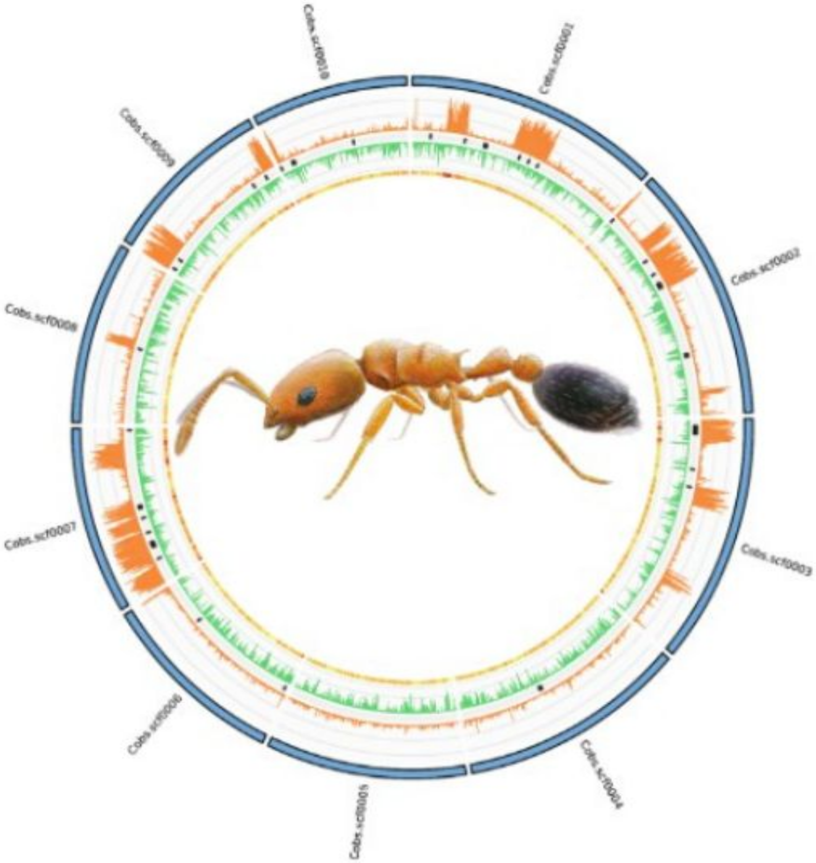
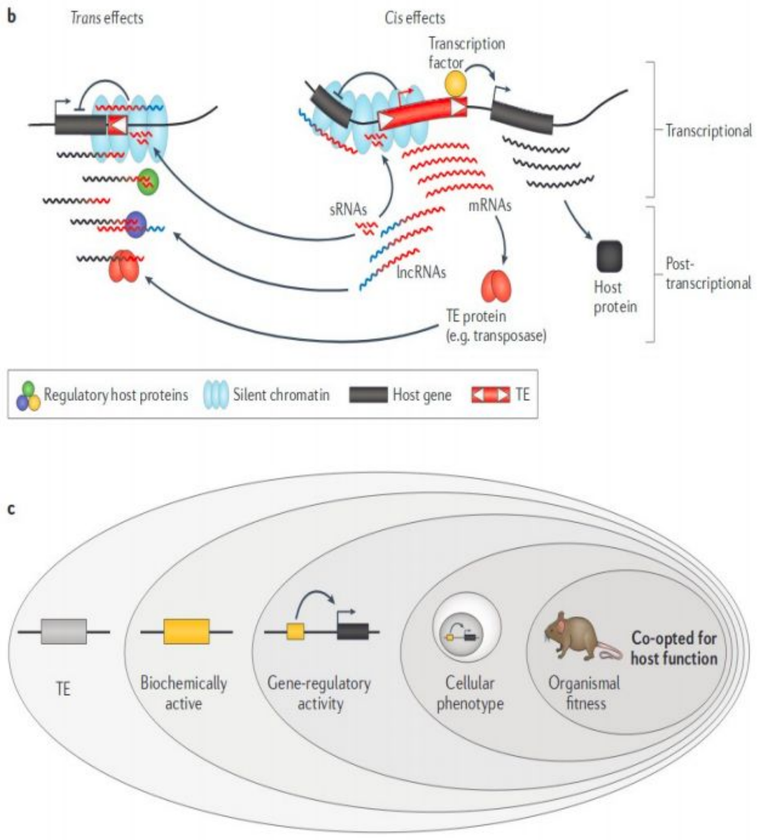
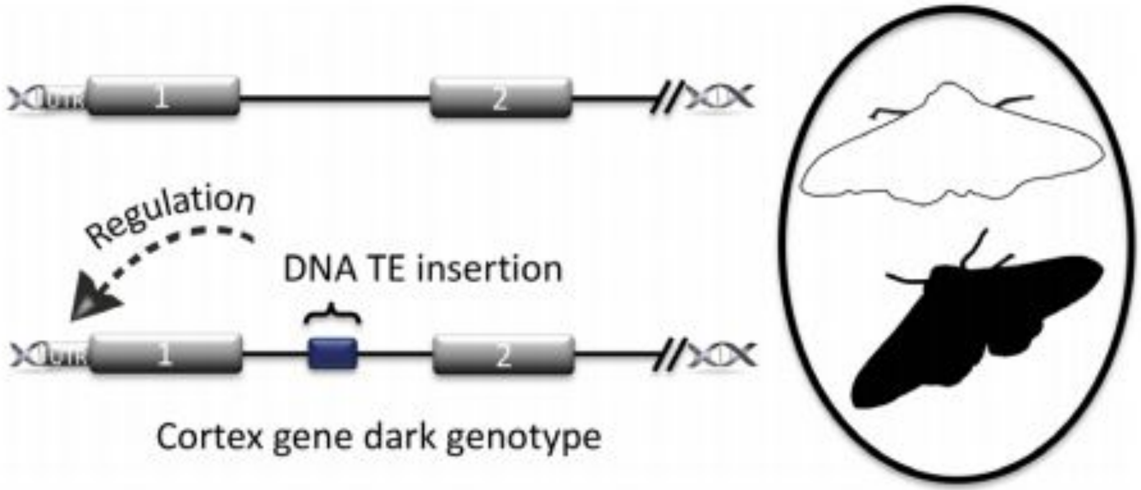
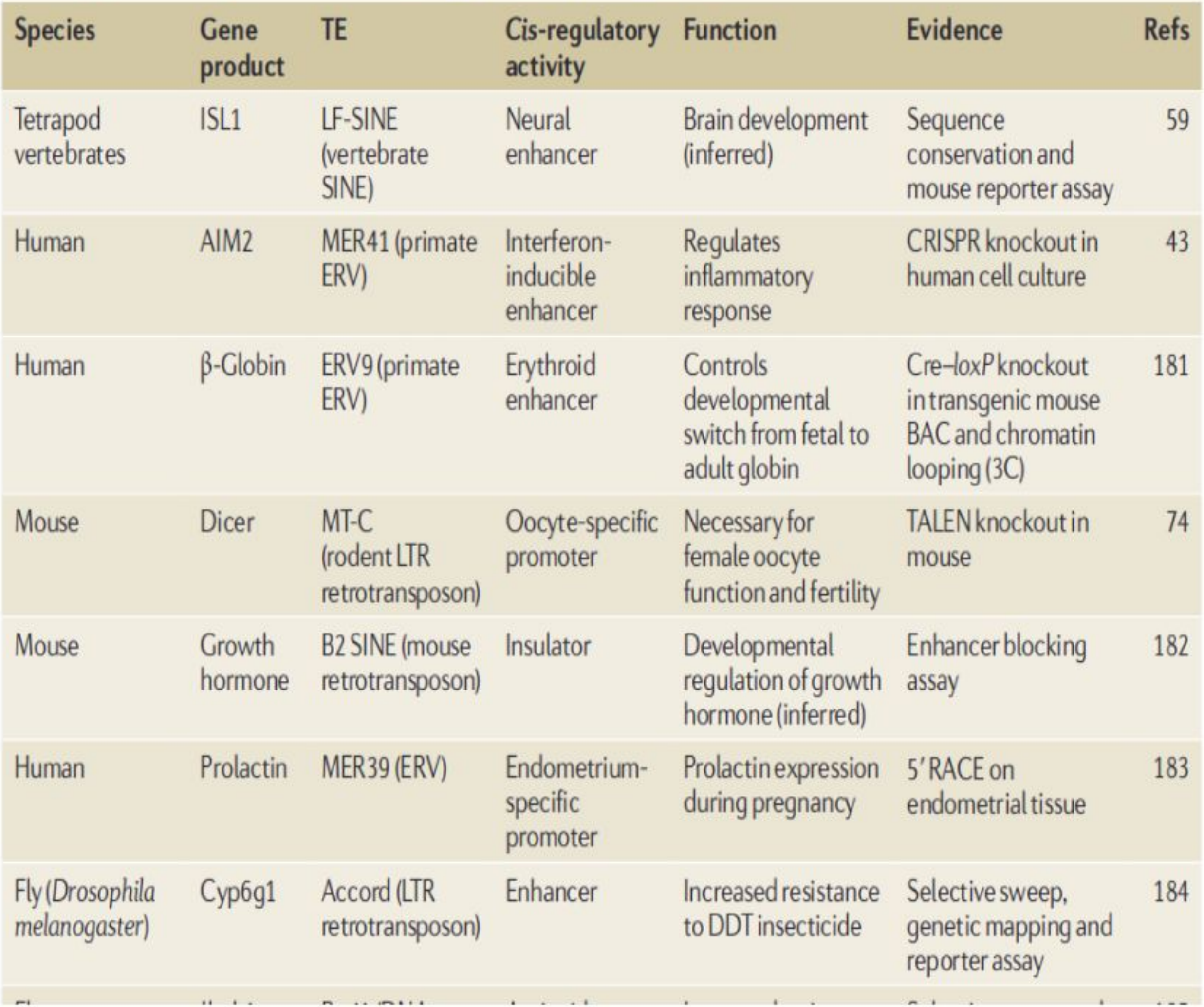


FIGURE 5 Genomic plasticity: Increased frequency of genomic rearrangements and mutations, directly or indirectly caused by TEs. To establish stable populations from genetically homogeneous founding populations, ants of the species *Cardiocondyla obscurior* are required to adapt to novel environmental challenges. Well-defined regions in the genome are enriched in TEs and genes likely involved in environmental adaptation (e.g., olfactory receptors). These "TE islands" are suspected to generate genetic novelty under environmental stress, thus facilitating the genetic diversification of incipient populations and ultimately enabling adaptation to novel environments. The figure shows the genomic architecture for the ten largest scaffolds of the *C. obscurior* draft assembly. The bar plots show TE content (orange) and gene content (green) in 10 kb windows. Black bars indicate the location of OR gene arrays, and GC content is shown in the heatmap

Transcriptional regulatory networks




UTR
1
2
Regulation
DNA TE insertion
UTR
1
2
Cortex gene dark genotype



Species	Gene product	TE	Cis-regulatory activity	Function	Evidence	Refs
Tetrapod vertebrates	ISL1	LF-SINE (vertebrate SINE)	Neural enhancer	Brain development (inferred)	Sequence conservation and mouse reporter assay	59
Human	AIM2	MER41 (primate ERV)	Interferon-inducible enhancer	Regulates inflammatory response	CRISPR knockout in human cell culture	43
Human	β-Globin	ERV9 (primate ERV)	Erythroid enhancer	Controls developmental switch from fetal to adult globin	Cre–*loxP* knockout in transgenic mouse BAC and chromatin looping (3C)	181
Mouse	Dicer	MT-C (rodent LTR retrotransposon)	Oocyte-specific promoter	Necessary for female oocyte function and fertility	TALEN knockout in mouse	74
Mouse	Growth hormone	B2 SINE (mouse retrotransposon)	Insulator	Developmental regulation of growth hormone (inferred)	Enhancer blocking assay	182
Human	Prolactin	MER39 (ERV)	Endometrium-specific promoter	Prolactin expression during pregnancy	5′ RACE on endometrial tissue	183
Fly (*Drosophila melanogaster*)	Cyp6g1	Accord (LTR retrotransposon)	Enhancer	Increased resistance to DDT insecticide	Selective sweep, genetic mapping and reporter assay	184

TE y Especiación

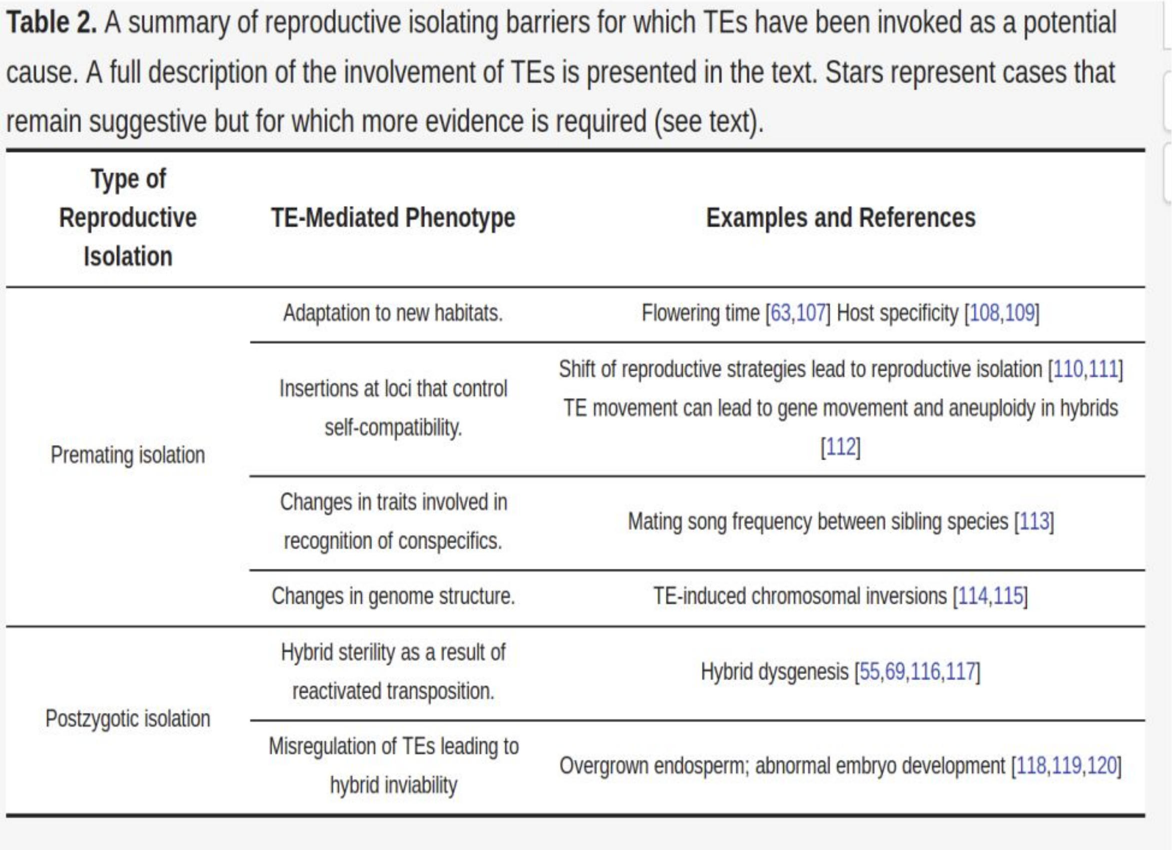
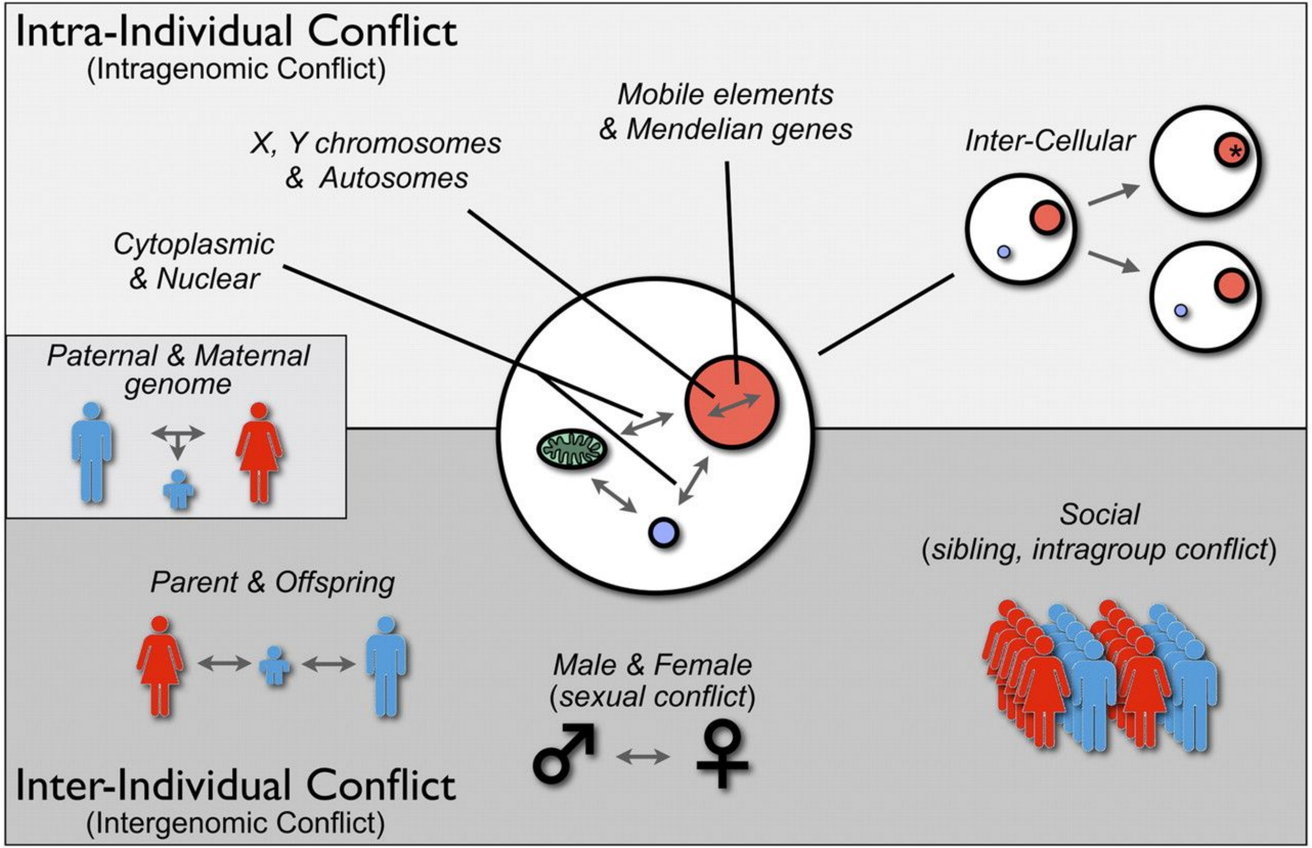


Table 2. A summary of reproductive isolating barriers for which TEs have been invoked as a potential cause. A full description of the involvement of TEs is presented in the text. Stars represent cases that remain suggestive but for which more evidence is required (see text).

Type of Reproductive Isolation	TE-Mediated Phenotype	Examples and References
Premating isolation	Adaptation to new habitats.	Flowering time [63,107] Host specificity [108,109]
	Insertions at loci that control self-compatibility.	Shift of reproductive strategies lead to reproductive isolation [110,111] TE movement can lead to gene movement and aneuploidy in hybrids [112]
	Changes in traits involved in recognition of conspecifics.	Mating song frequency between sibling species [113]
	Changes in genome structure.	TE-induced chromosomal inversions [114,115]
Postzygotic isolation	Hybrid sterility as a result of reactivated transposition.	Hybrid dysgenesis [55,69,116,117]
	Misregulation of TEs leading to hybrid inviability	Overgrown endosperm; abnormal embryo development [118,119,120]

Conflicto Genético



Werren, 2011. https://doi.org/10.1073/pnas.1102343108

Resultados del conflicto evolutivo

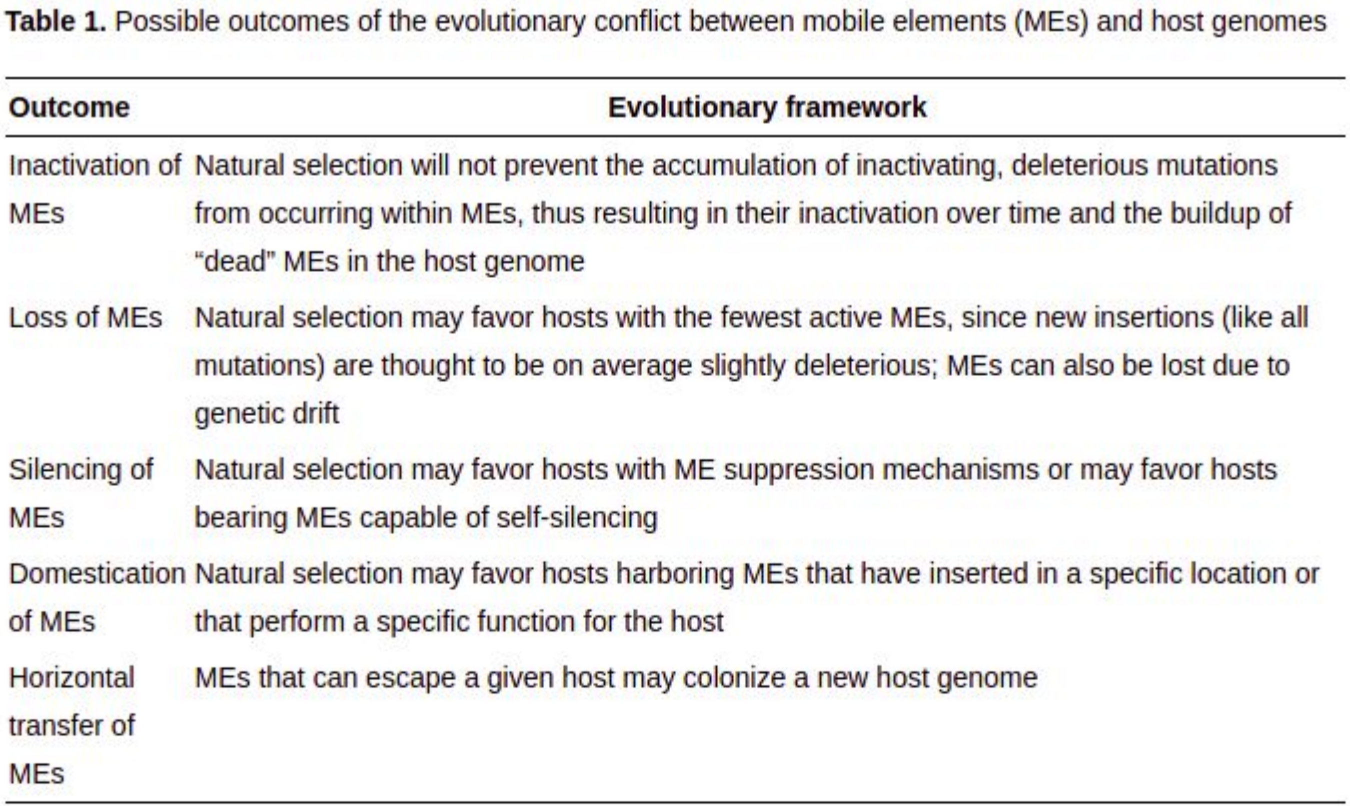


Table 1. Possible outcomes of the evolutionary conflict between mobile elements (MEs) and host genomes

Outcome	Evolutionary framework
Inactivation of MEs	Natural selection will not prevent the accumulation of inactivating, deleterious mutations from occurring within MEs, thus resulting in their inactivation over time and the buildup of "dead" MEs in the host genome
Loss of MEs	Natural selection may favor hosts with the fewest active MEs, since new insertions (like all mutations) are thought to be on average slightly deleterious; MEs can also be lost due to genetic drift
Silencing of MEs	Natural selection may favor hosts with ME suppression mechanisms or may favor hosts bearing MEs capable of self-silencing
Domestication of MEs	Natural selection may favor hosts harboring MEs that have inserted in a specific location or that perform a specific function for the host
Horizontal transfer of MEs	MEs that can escape a given host may colonize a new host genome

Ecología y Evolución al rescate

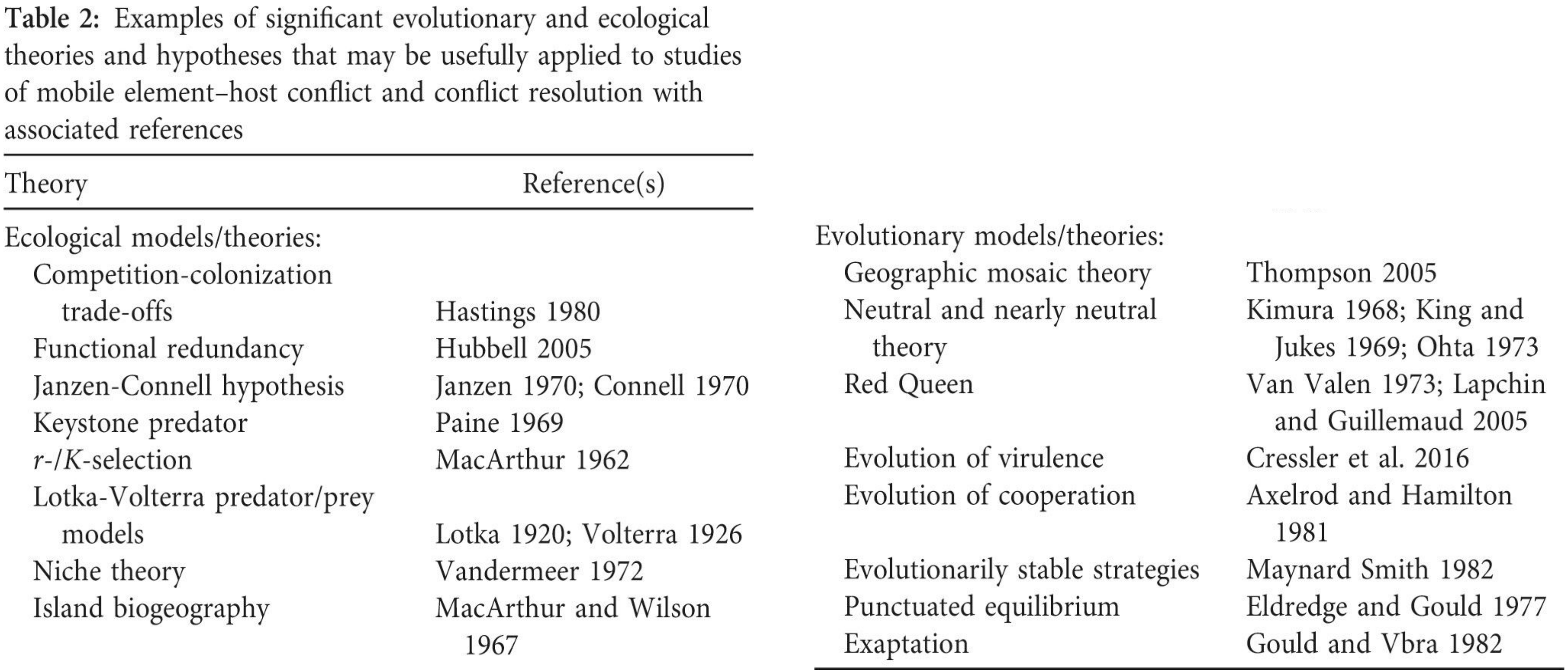


Table 2: Examples of significant evolutionary and ecological theories and hypotheses that may be usefully applied to studies of mobile element–host conflict and conflict resolution with associated references

Theory	Reference(s)
Ecological models/theories:	
Competition-colonization trade-offs	Hastings 1980
Functional redundancy	Hubbell 2005
Janzen-Connell hypothesis	Janzen 1970; Connell 1970
Keystone predator	Paine 1969
r-/*K*-selection	MacArthur 1962
Lotka-Volterra predator/prey models	Lotka 1920; Volterra 1926
Niche theory	Vandermeer 1972
Island biogeography	MacArthur and Wilson 1967
Evolutionary models/theories:	
Geographic mosaic theory	Thompson 2005
Neutral and nearly neutral theory	Kimura 1968; King and Jukes 1969; Ohta 1973
Red Queen	Van Valen 1973; Lapchin and Guillemaud 2005
Evolution of virulence	Cressler et al. 2016
Evolution of cooperation	Axelrod and Hamilton 1981
Evolutionarily stable strategies	Maynard Smith 1982
Punctuated equilibrium	Eldredge and Gould 1977
Exaptation	Gould and Vbra 1982