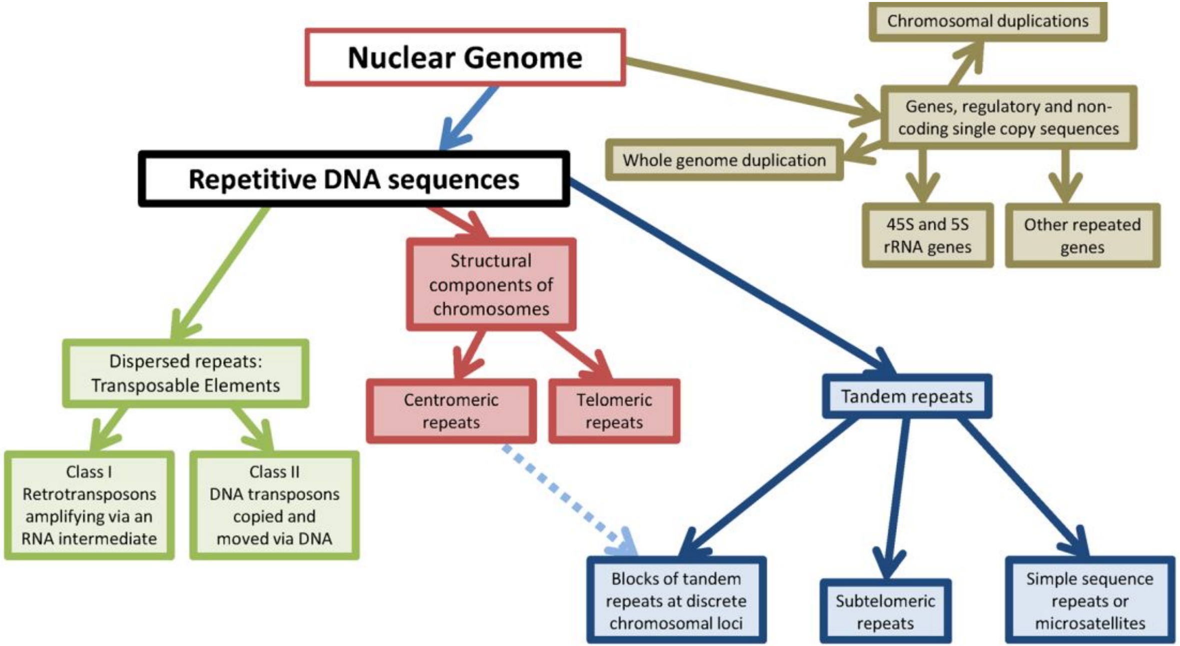
ELEMENTOS TRANSPONIBLES

Genómica Evolutiva 2021

Repetidos dispersos



Barbara McClintock

- Mostró que los genes estaban físicamente en los cromosomas
- Mostró que durante la meiosis ocurría crossing over
- Descubrió el silenciamiento génico

Pero la investigación que provocó que años después le otorgaran el premio nobel de fisiología y medicina fue la de los elementos móviles (1983)

- Demostró que los genes eran responsables de "*prender*" y "*apagar*" características físicas

- En 1953 dejó de publicar sus resultados

transposones = EM = ET

Barbara McClintock

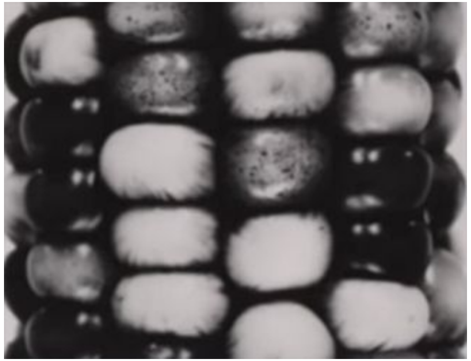
Ac -
Ds +

Dissociator (*Ds*)
Activator (*Ac*)

Ac +
Ds +

2Ac +
Ds +

3Ac +
Ds +



Barbara McClintock

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Dissociator (*Ds*)
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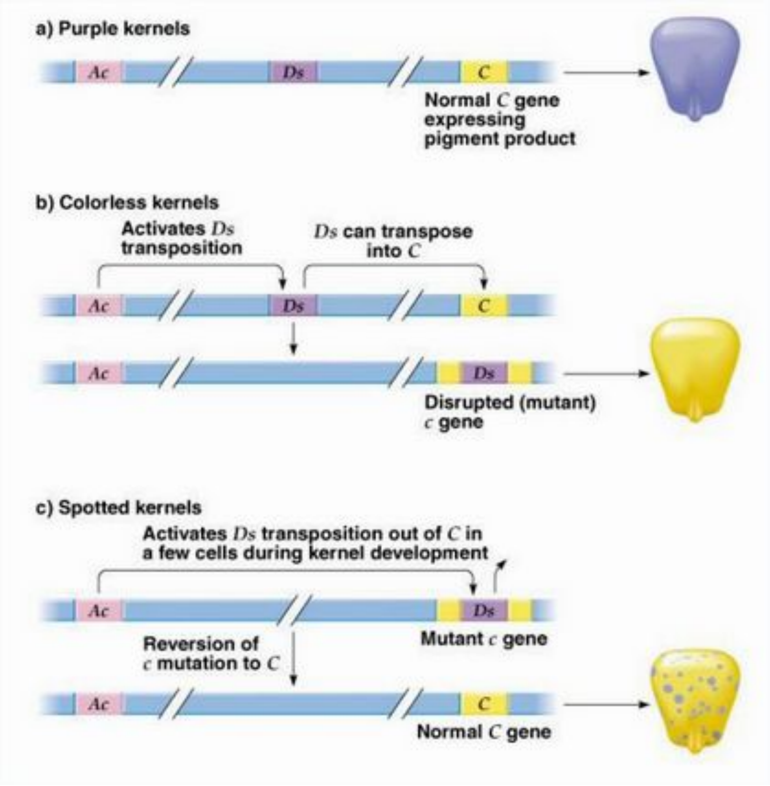

Barbara McClintock

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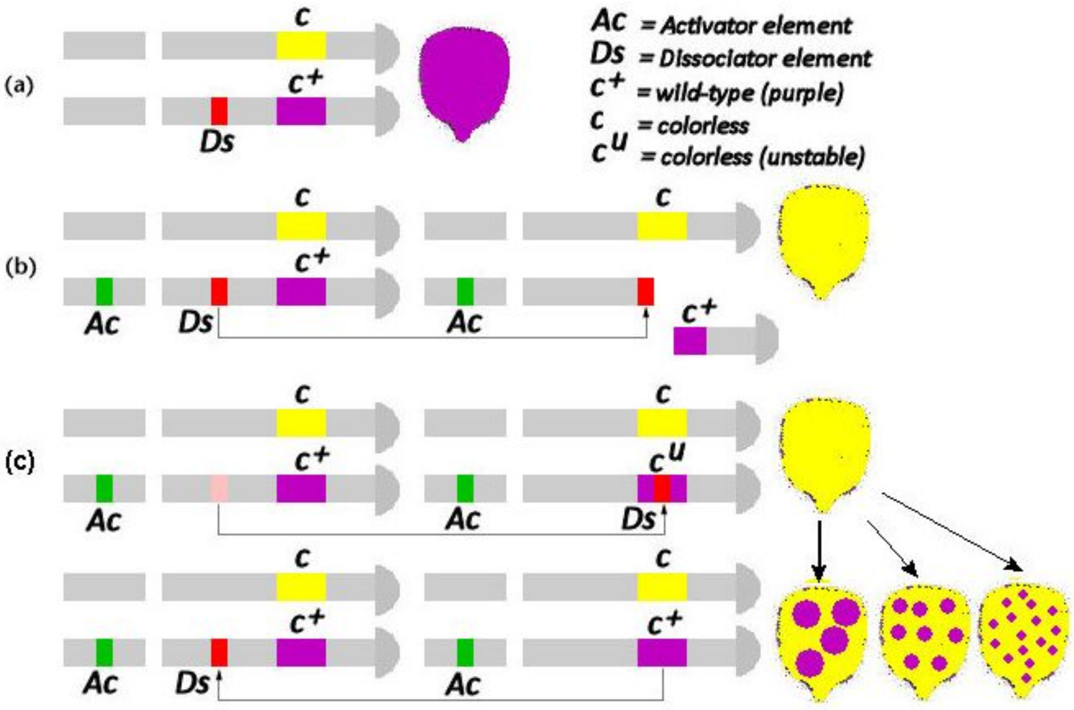
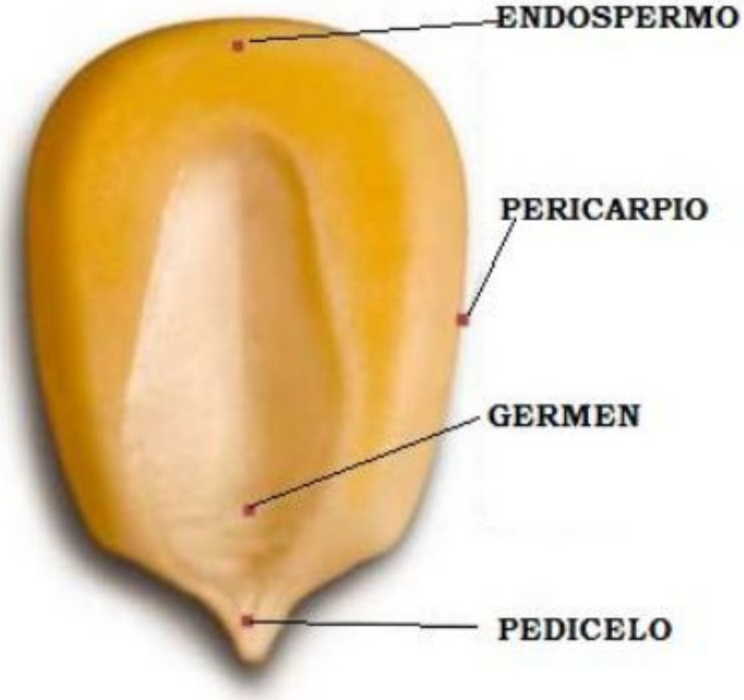
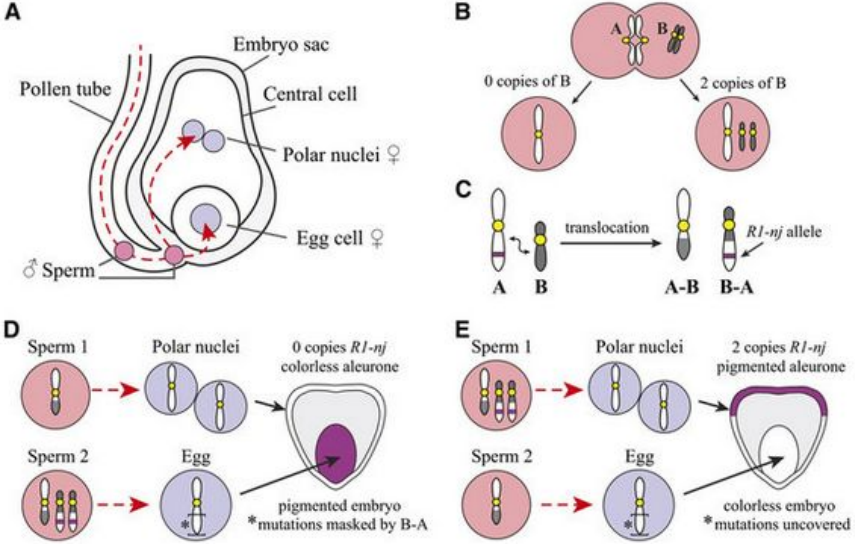
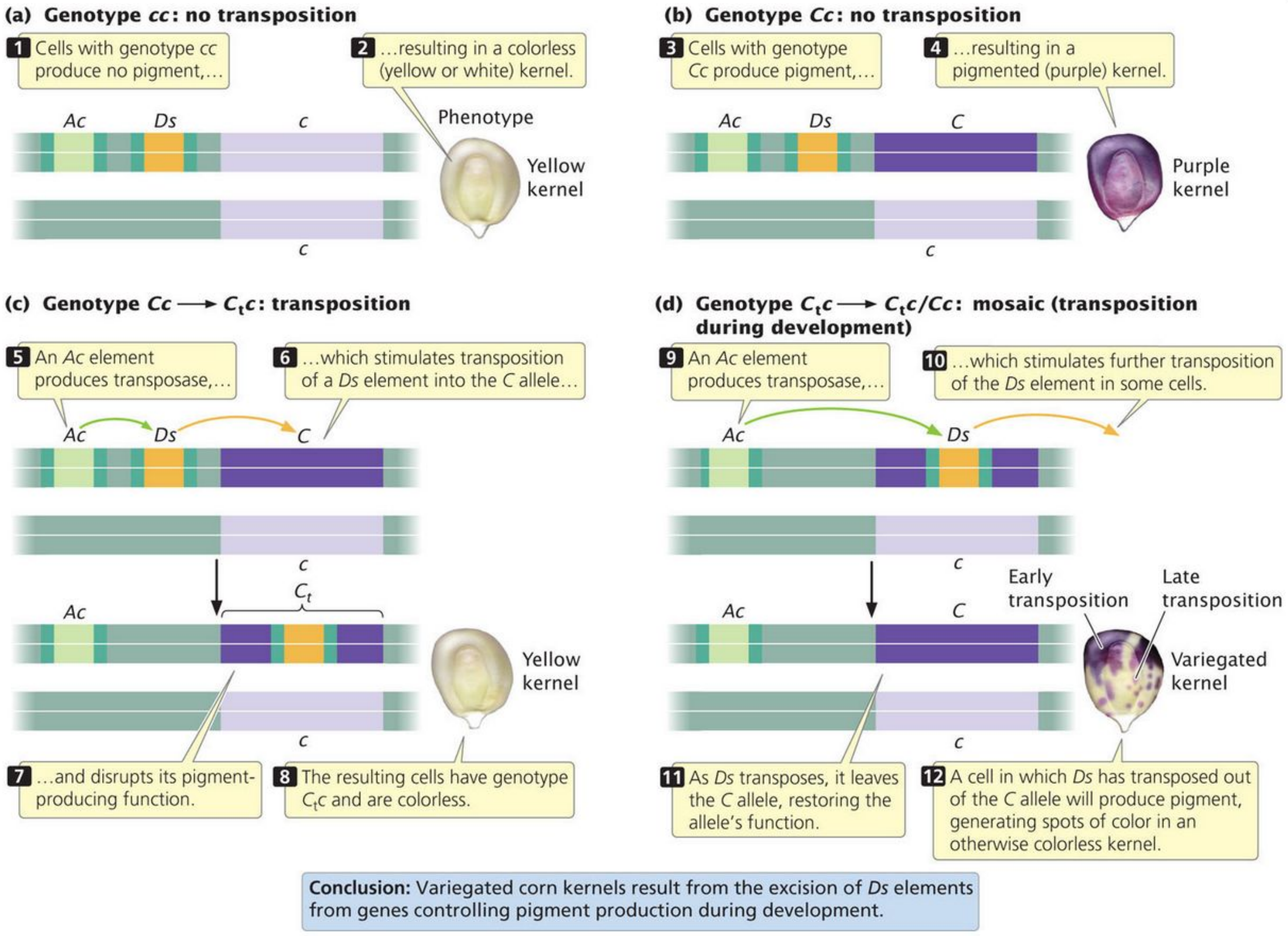
Ds +



The *Ac* - *Ds* system in maize (*Zea*): genetic analysis of "*jumping genes*"


ENDOSPERMO
PERICARPIO
GERMEN
PEDICELO


A
Embryo sac
Pollen tube
Central cell
Polar nuclei ♀
Egg cell ♀
♂ Sperm
B
A
B
0 copies of B
2 copies of B
C
translocation
R1-nj allele
A
B
A-B
B-A
D
Sperm 1
Polar nuclei
0 copies R1-nj
colorless aleurone
Sperm 2
Egg
pigmented embryo
*mutations masked by B-A
E
Sperm 1
Polar nuclei
2 copies R1-nj
pigmented aleurone
Sperm 2
Egg
colorless embryo
*mutations uncovered



(a) Genotype cc: no transposition

1 Cells with genotype *cc* produce no pigment,…

2 …resulting in a colorless (yellow or white) kernel.

(b) Genotype Cc: no transposition

3 Cells with genotype *Cc* produce pigment,…

4 …resulting in a pigmented (purple) kernel.

(c) Genotype $Cc \longrightarrow C_t c$: transposition

5 An *Ac* element produces transposase,…

6 …which stimulates transposition of a *Ds* element into the *C* allele…

7 …and disrupts its pigment-producing function.

8 The resulting cells have genotype $C_t c$ and are colorless.

(d) Genotype $C_t c \longrightarrow C_t c/Cc$: mosaic (transposition during development)

9 An *Ac* element produces transposase,…

10 …which stimulates further transposition of the *Ds* element in some cells.

11 As *Ds* transposes, it leaves the *C* allele, restoring the allele's function.

12 A cell in which *Ds* has transposed out of the *C* allele will produce pigment, generating spots of color in an otherwise colorless kernel.

Conclusion: Variegated corn kernels result from the excision of *Ds* elements from genes controlling pigment production during development.

Figure 18.34: Transposition results in variegated maize kernels.

Barbara McClintock

Ac -
Ds +

Dissociator (*Ds*)
Activator (*Ac*)

12

13

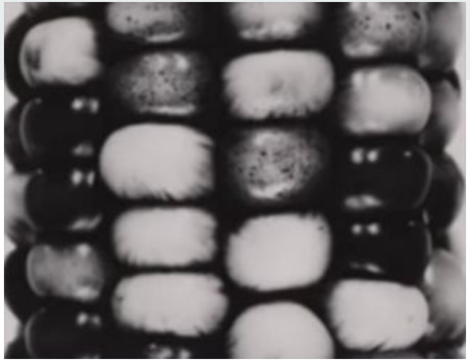
Ac +
Ds +

14

2Ac +
Ds +

15

3Ac +
Ds +



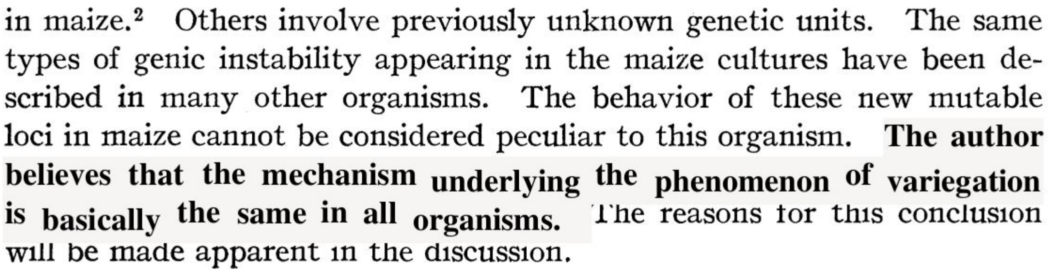
PNAS CLASSIC ARTICLE

The origin and behavior of mutable loci in maize

Barbara McClintock

PNAS June 1, 1950 36 (6) 344-355; https://doi.org/10.1073/pnas.36.6.344

Communicated April 8, 1950



in maize.[2] Others involve previously unknown genetic units. The same types of genic instability appearing in the maize cultures have been described in many other organisms. The behavior of these new mutable loci in maize cannot be considered peculiar to this organism. **The author believes that the mechanism underlying the phenomenon of variegation is basically the same in all organisms.** The reasons for this conclusion will be made apparent in the discussion.

Barbara McCLintock

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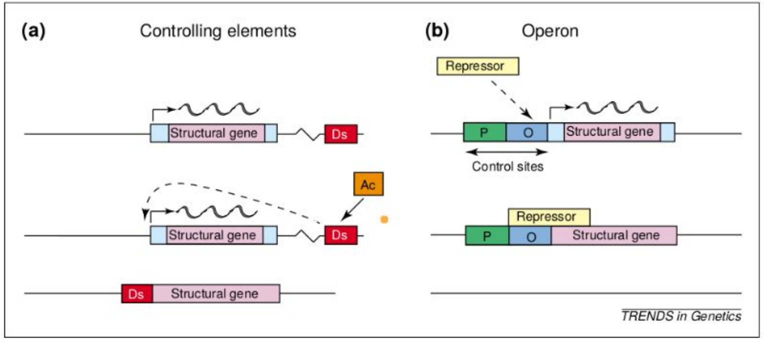
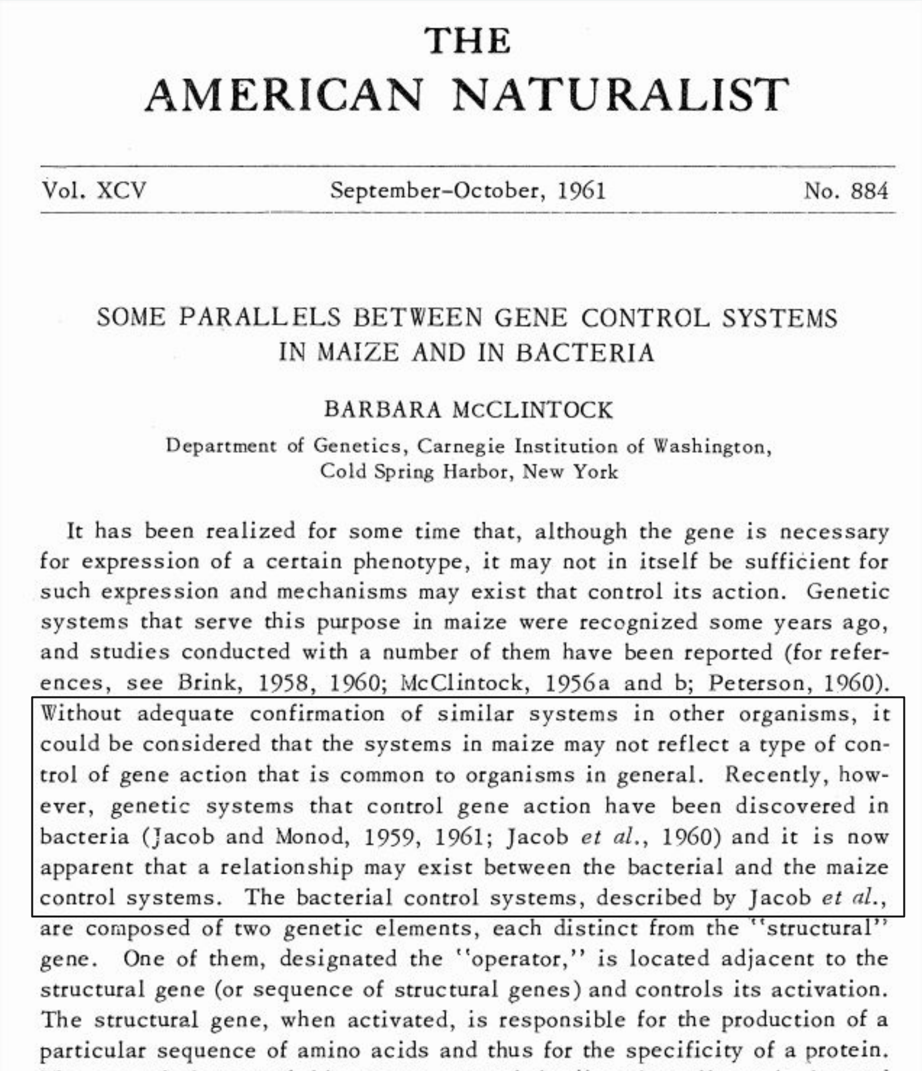
1944 tercer mujer en ser reconocida como académica en la Academia Nacional de Ciencias de Estados Unidos



TRENDS in Genetics



THE
AMERICAN NATURALIST

Vol. XCV September–October, 1961 No. 884

SOME PARALLELS BETWEEN GENE CONTROL SYSTEMS IN MAIZE AND IN BACTERIA

BARBARA MCCLINTOCK

Department of Genetics, Carnegie Institution of Washington,
Cold Spring Harbor, New York

It has been realized for some time that, although the gene is necessary for expression of a certain phenotype, it may not in itself be sufficient for such expression and mechanisms may exist that control its action. Genetic systems that serve this purpose in maize were recognized some years ago, and studies conducted with a number of them have been reported (for references, see Brink, 1958, 1960; McClintock, 1956a and b; Peterson, 1960). Without adequate confirmation of similar systems in other organisms, it could be considered that the systems in maize may not reflect a type of control of gene action that is common to organisms in general. Recently, however, genetic systems that control gene action have been discovered in bacteria (Jacob and Monod, 1959, 1961; Jacob *et al.*, 1960) and it is now apparent that a relationship may exist between the bacterial and the maize control systems. The bacterial control systems, described by Jacob *et al.*, are composed of two genetic elements, each distinct from the "structural" gene. One of them, designated the "operator," is located adjacent to the structural gene (or sequence of structural genes) and controls its activation. The structural gene, when activated, is responsible for the production of a particular sequence of amino acids and thus for the specificity of a protein.

Globalmente

The repeats of a parasite's genome, therefore -their presence and absence, their type, activity, and location – can be a window on the genomic organization that enables parasitism

Wickstead et al. 2003

Los elementos móviles son ubicuos, muy diversos
Han tenido y tienen un tremendo efecto en la arquitectura y plasticidad genómica

Lynch 2007

Grandes clasificaciones

EM **vivos**
Son elementos dinámicos que remodelan el genoma del hospedero generando reordenamientos, creando y destruyendo genes, barajando genes existentes y modulando patrones de expresión

EM **muertos**
(que ya no pueden proliferar) constituyen un registro paleontológico, se pueden estudiar en busca de pistas sobre eventos evolutivos que tuvieron lugar

Grandes clasificaciones

EM **Autónomos**
Son elementos que pueden moverse de manera autosuficiente a diferentes partes del genoma
Tiene los genes funcionales necesarios para moverse

EM **No Autónomos**
Carecen de alguno o todos los dominios de los ET autónomos. No pueden moverse por sí solos, pero pueden hacerlo gracias a la maquinaria (prestada o secuestrada) de los autónomos

Grandes clasificaciones

EM **Clase I**
Retrotransposones

EM **Clase II**
Transposones de ADN

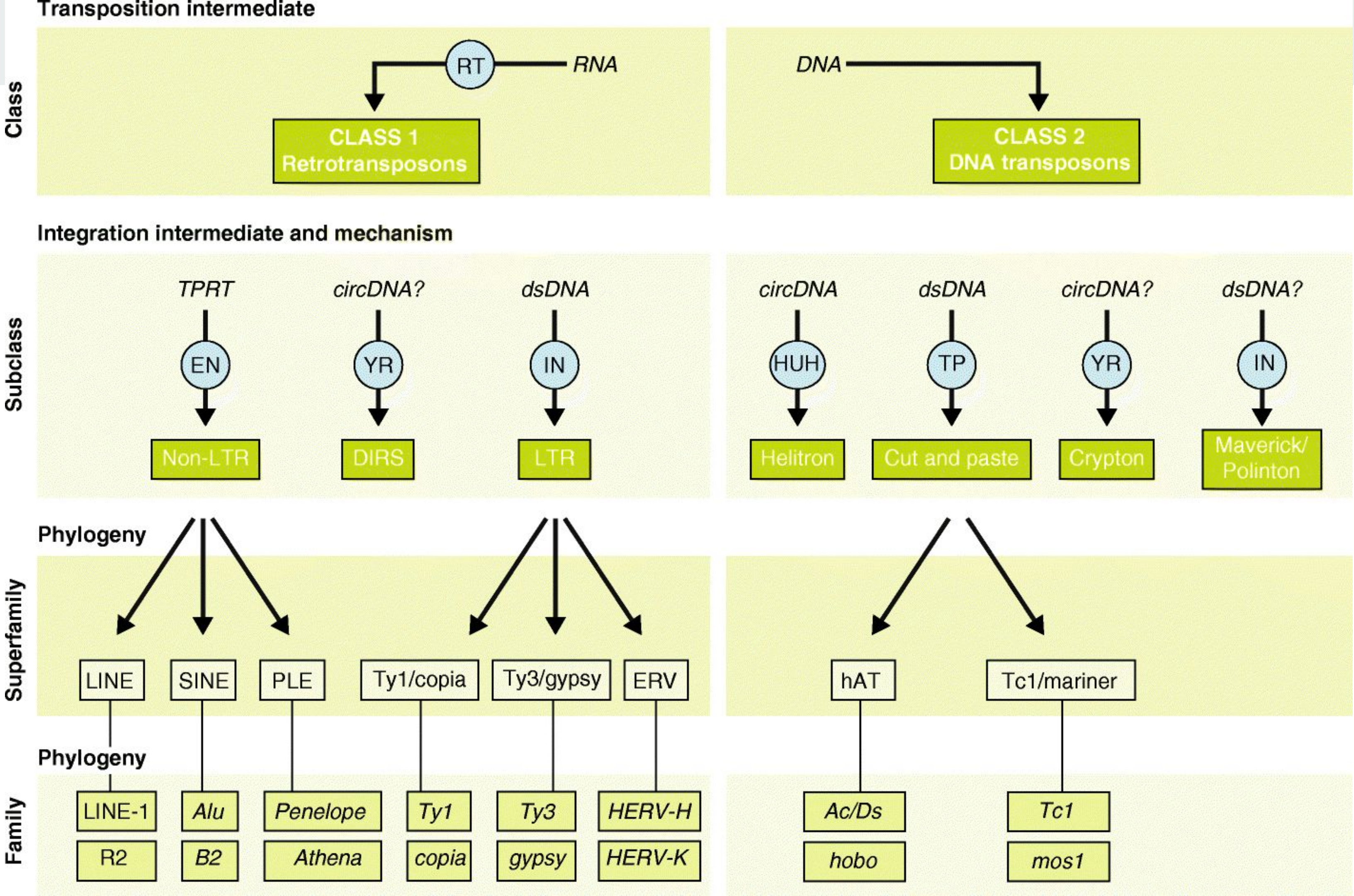
Sub-clasificaciones

Sistema jerárquico

- Mecanismos de transposición
- Similitud entre los elementos
- Relación de las estructuras

Sub-clasificaciones

- **Clase** I (retrotransposones) y Clase II (transposones)
- **Subclase** - Distingue entre los ET que se copian de los que se mueven. Los retrotransposones siempre se copian. Los transposones se dividen en dos, aquellos que que sólo se mueven y aquellos que se copian, se replican (sin retrotranscribirse) y se insertan
- **Orden -** Marca las diferencias en el mecanismo de inserción y por lo tanto en su organización interna y enzimática
- **Superfamilia** - Comparten la estrategia de replicación, pero son diferentes en la estructura proteica, los dominios que codifican y la presencia y el tamaño de los repetidos flanqueantes.
- **Familias** - Definidas por conservación de secuencia. Dentro de la superfamilia, hay similaridad proteica pero no nucleotídica.

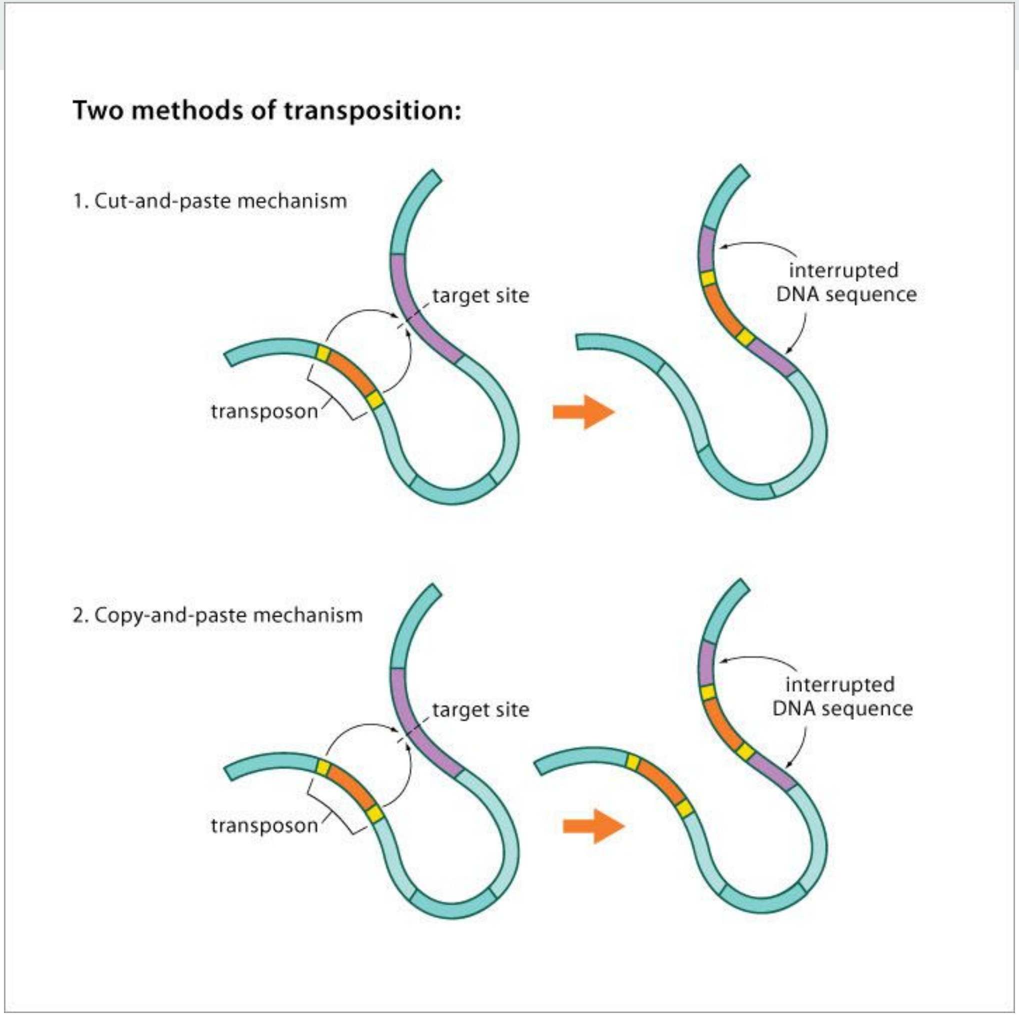

Transposition intermediate
Class
RT
RNA
CLASS 1
Retrotransposons
DNA
CLASS 2
DNA transposons
Integration intermediate and mechanism
Subclass
TPRT
EN
Non-LTR
circDNA?
YR
DIRS
dsDNA
IN
LTR
circDNA
HUH
Helitron
dsDNA
TP
Cut and paste
circDNA?
YR
Crypton
dsDNA?
IN
Maverick/
Polinton
Phylogeny
Superfamily
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

Repetidos dispersos

- Interspersed repeats
  - DNA transposons → Clase II
  - Retroelements → Clase I

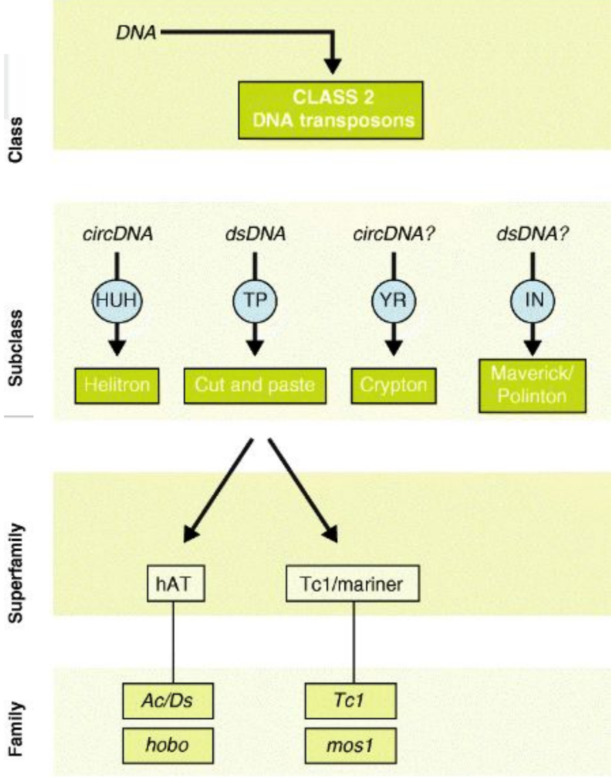
~45% del genoma humano
~55% o más, del genoma del maíz.

Clase II


Two methods of transposition:
1. Cut-and-paste mechanism
target site
transposon
interrupted DNA sequence
2. Copy-and-paste mechanism
target site
transposon
interrupted DNA sequence

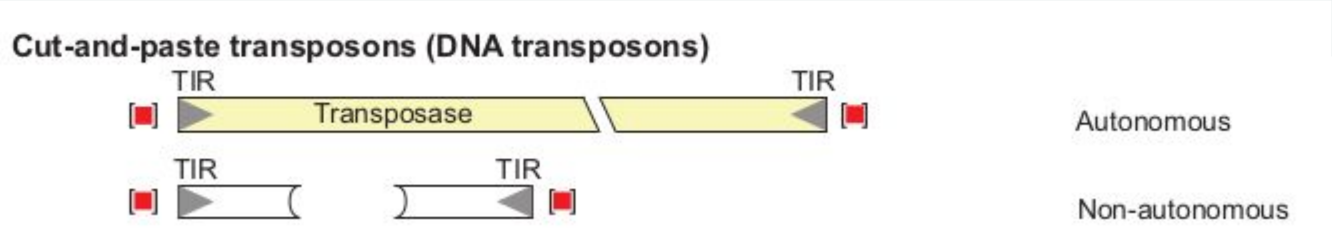
Clase II

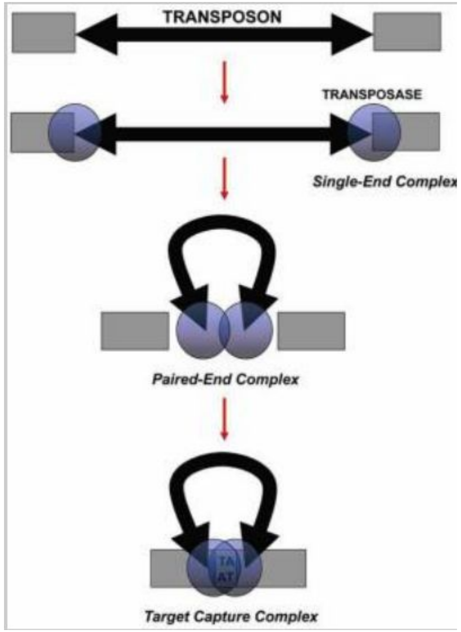

TIR
TRANSPOSASE GENE
TIR

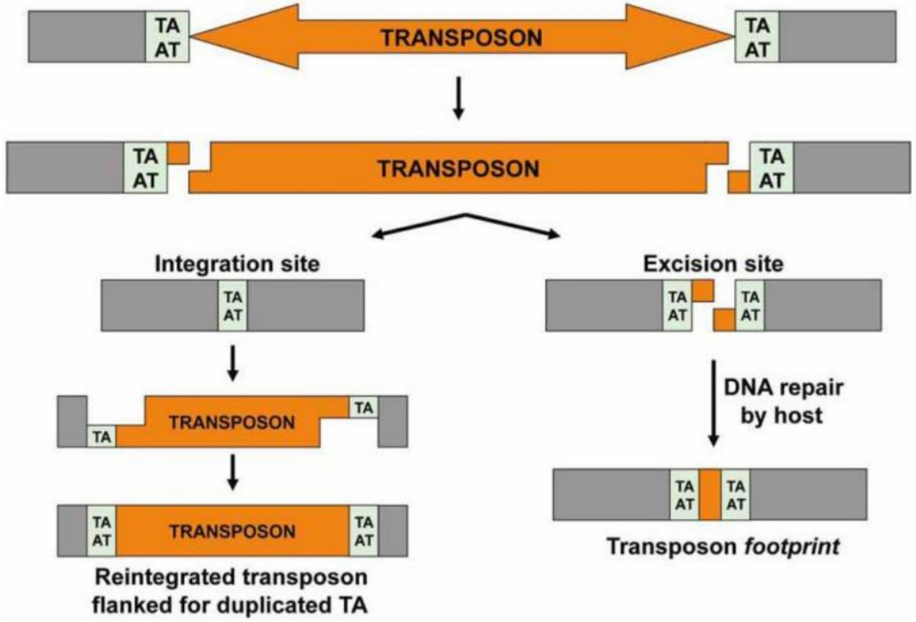

Class
Subclass
Superfamily
Family
DNA
CLASS 2
DNA transposons
circDNA
dsDNA
circDNA?
dsDNA?
HUH
TP
YR
IN
Helitron
Cut and paste
Crypton
Maverick/
Polinton
hAT
Tc1/mariner
Ac/Ds
hobo
Tc1
mos1

Clase II

Tc1/*mariner*

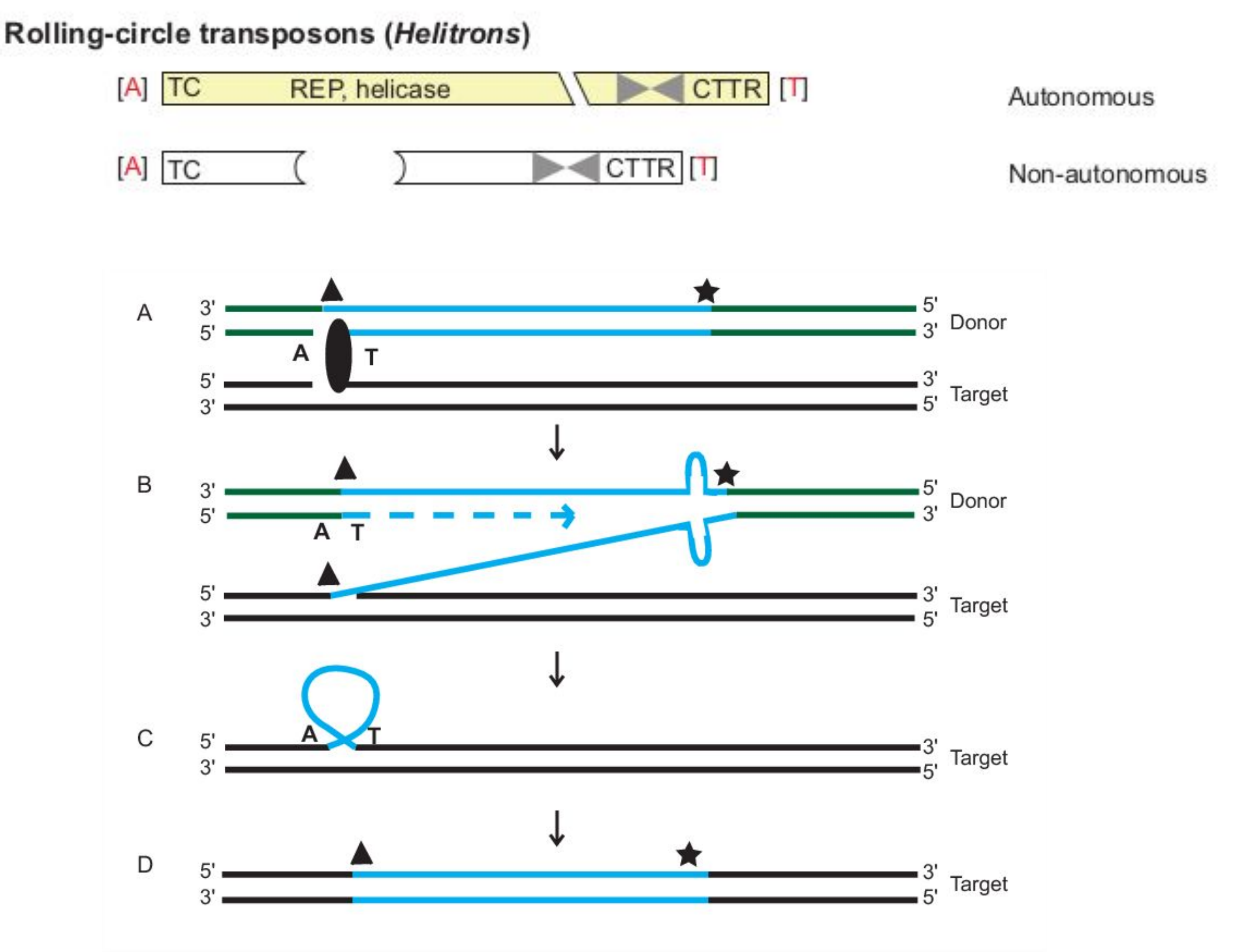

Cut-and-paste transposons (DNA transposons)





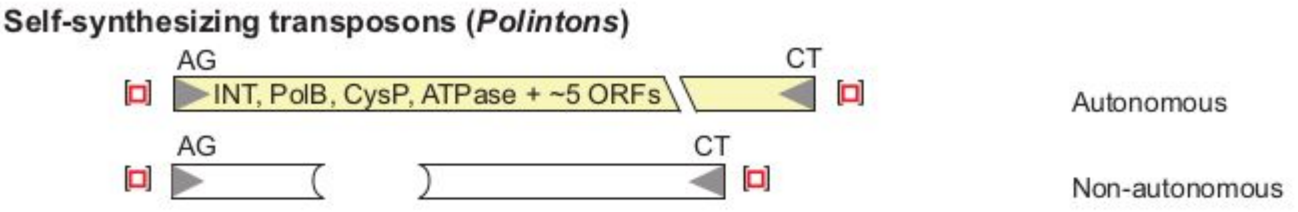
Clase II

Helitron



Clase II

Polintons (Mavericks)



- Descubiertos y caracterizados por análisis computacionales (Jurka et al 2006)
- Tienen entre 15-20Kb // TSD de 6pb // 100-1000 pb TIR
- Codifican para más de 10 proteínas

en resumen

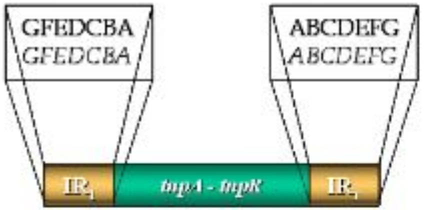
Clase II (DNA Transposons)

- Algunos pueden transponerse en todas las células, otros son altamente específicos
- Los sitios receptores son variables
- Algunos elementos tienen preferencia por algunos sitios receptores

- Son fuente de cambios genéticos y han jugado un rol importante en la evolución de genomas
- Son usados como herramientas, para introducir ADN en un genoma (mutagénesis, transgénesis)

Transposones de ADN en bacterias

- Son segmentos de ADN con capacidad de moverse entre una locación a otra
- Poseen una transposasa y no requiere zonas de homología entre los sitios
- Los ET más simples son Secuencias de Inserción o elementos
  - **Posee inverted repeat y una transposasa (750-1600pb)**



Secuencia de inserción
(transposón simple)

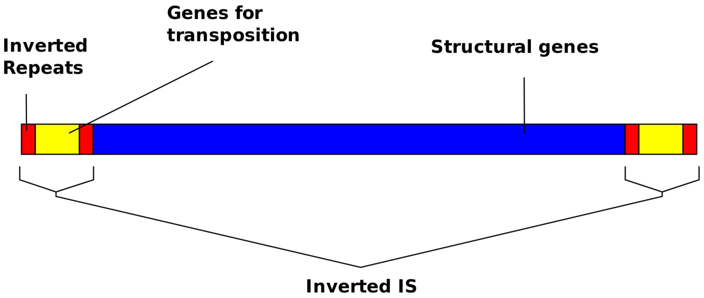
Transposón compuesto

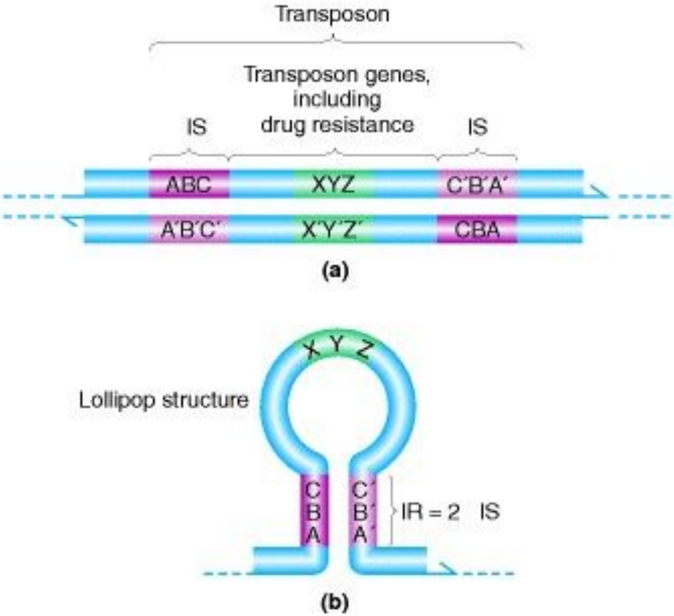
Direcciones probables de transcripción de los IS

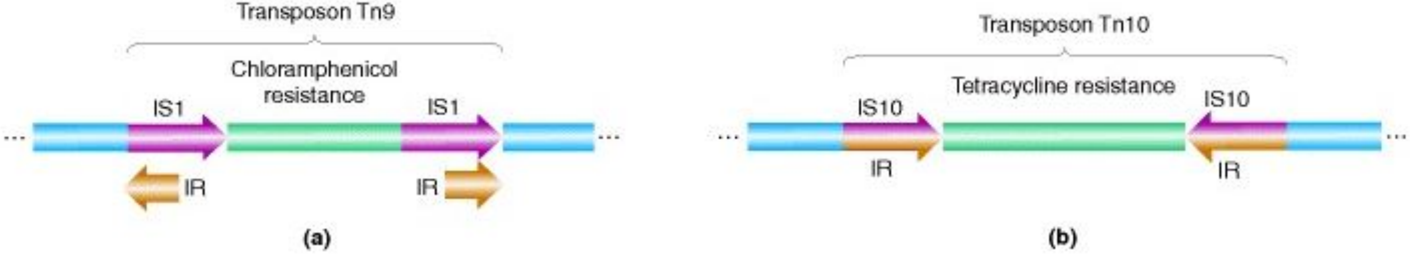
- Fundamentales para la dispersión de resistencia
- Contribuyen a la movilización de integrones

Transposones en bacterias

Bacterial composite transposon



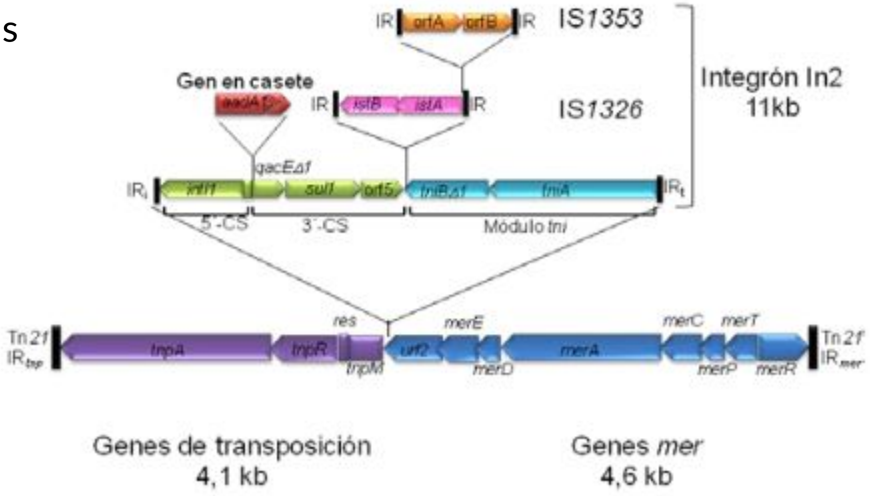




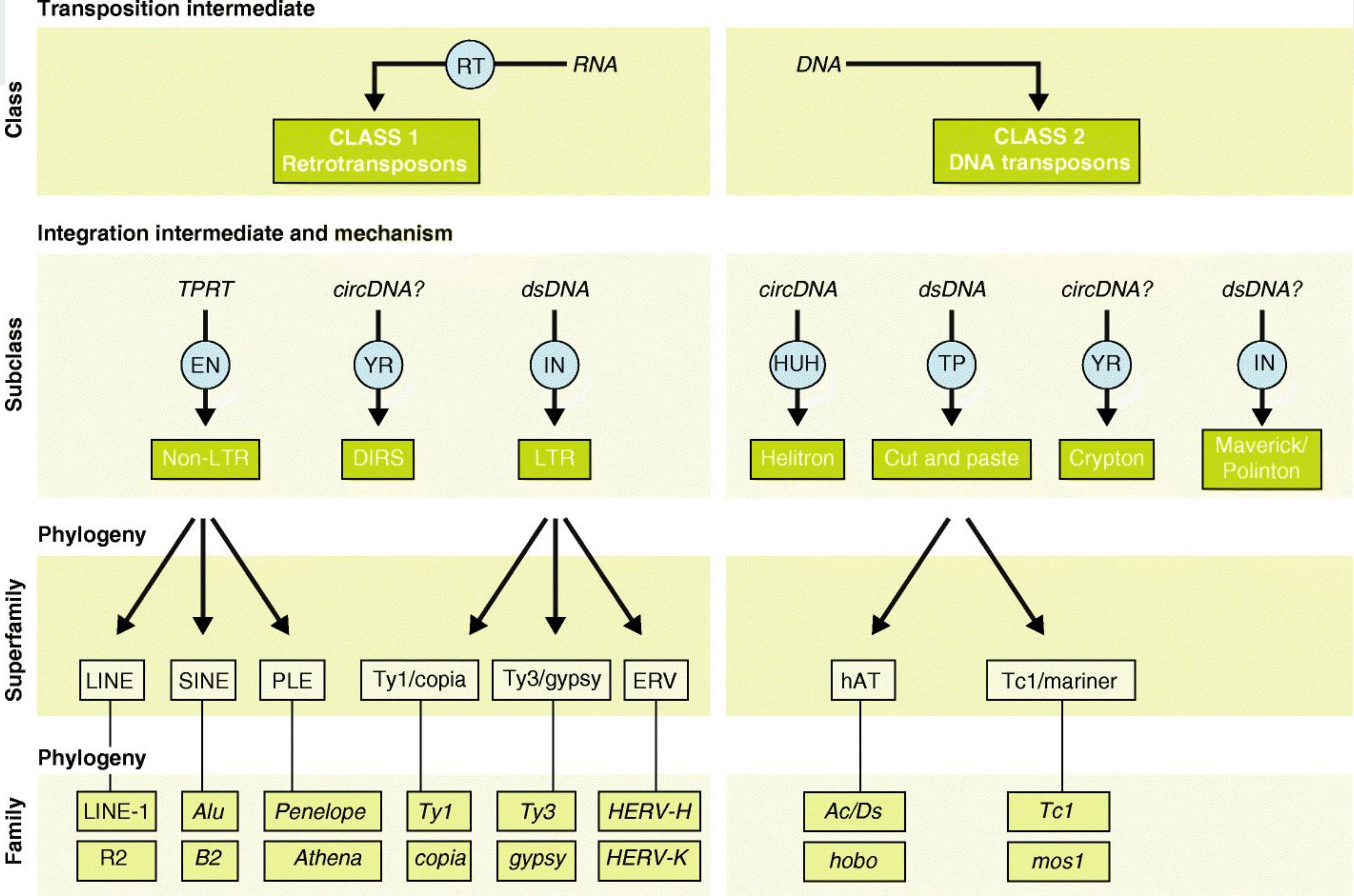
Transposones en bacterias

transposones complejos

- Presentan repeticiones terminales IR de 35-40pb
- Codifican para una transposasa y resolvasa que median la transposición
- El mecanismo es replicativo en dos etapas



Transposón Tn21



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*

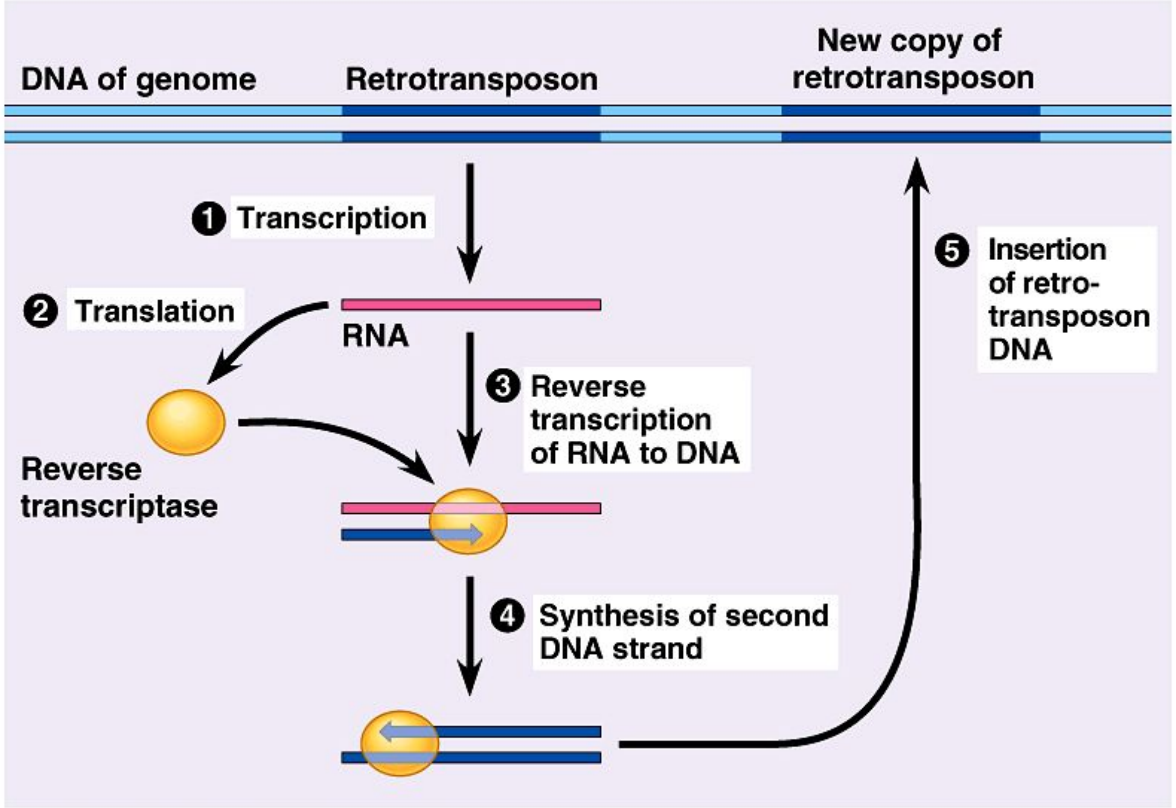
HERV-H, *HERV-K*

Ac/Ds, *hobo*

Tc1, *mos1*

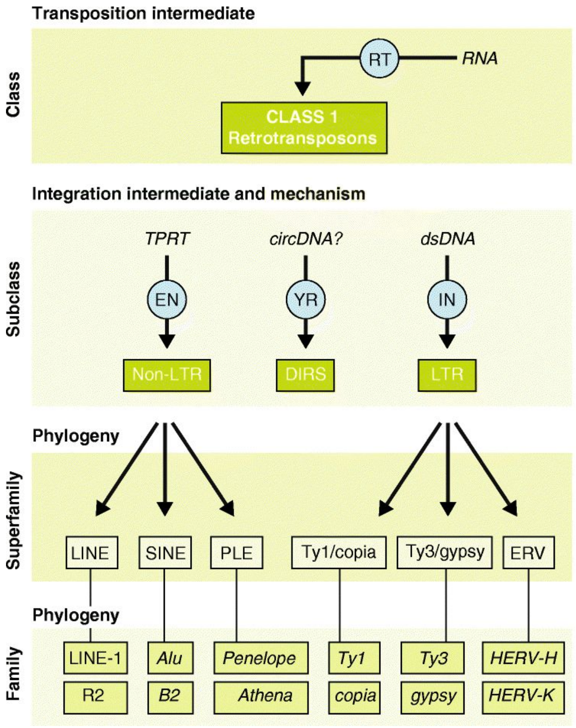
RETRO
ELEMENTS

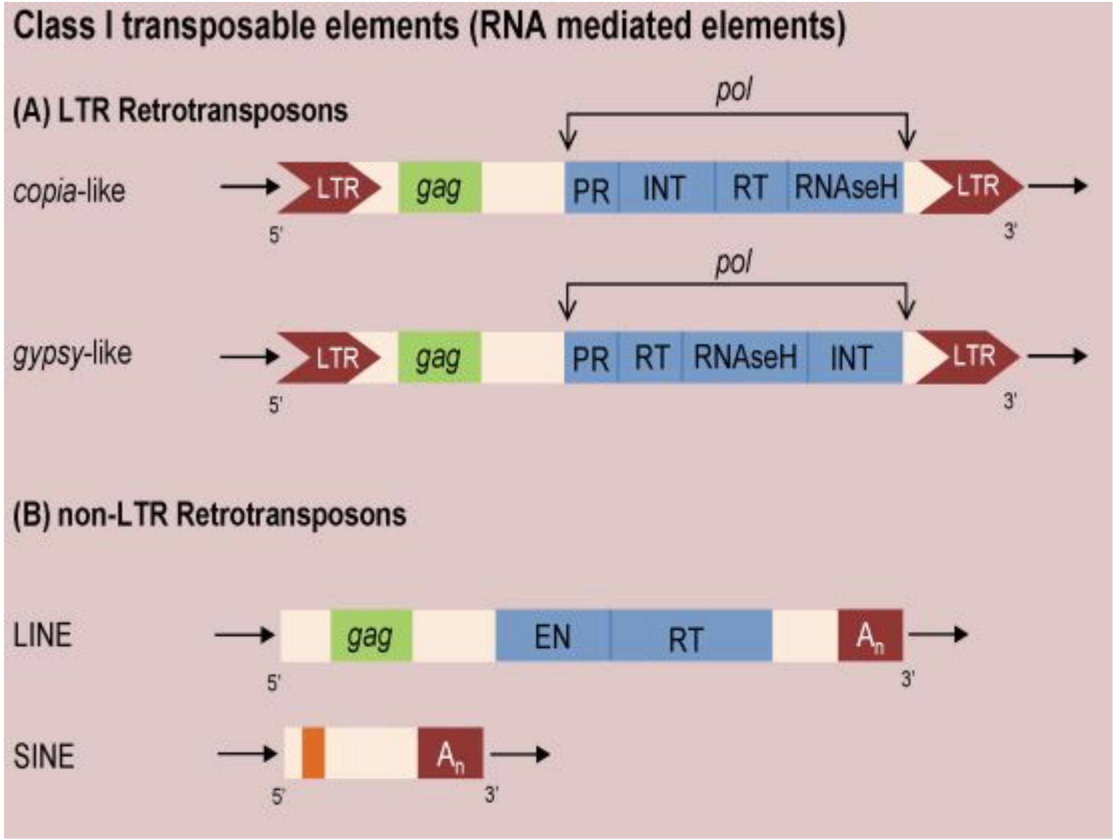
Clase I


DNA of genome
Retrotransposon
New copy of retrotransposon
❶ Transcription
❷ Translation
RNA
❸ Reverse transcription of RNA to DNA
Reverse transcriptase
❹ Synthesis of second DNA strand
❺ Insertion of retro-transposon DNA

Clase I

- **LTR**
- **Non-LTR**
- Penelope
- DIRS





Class I transposable elements (RNA mediated elements)

(A) LTR Retrotransposons

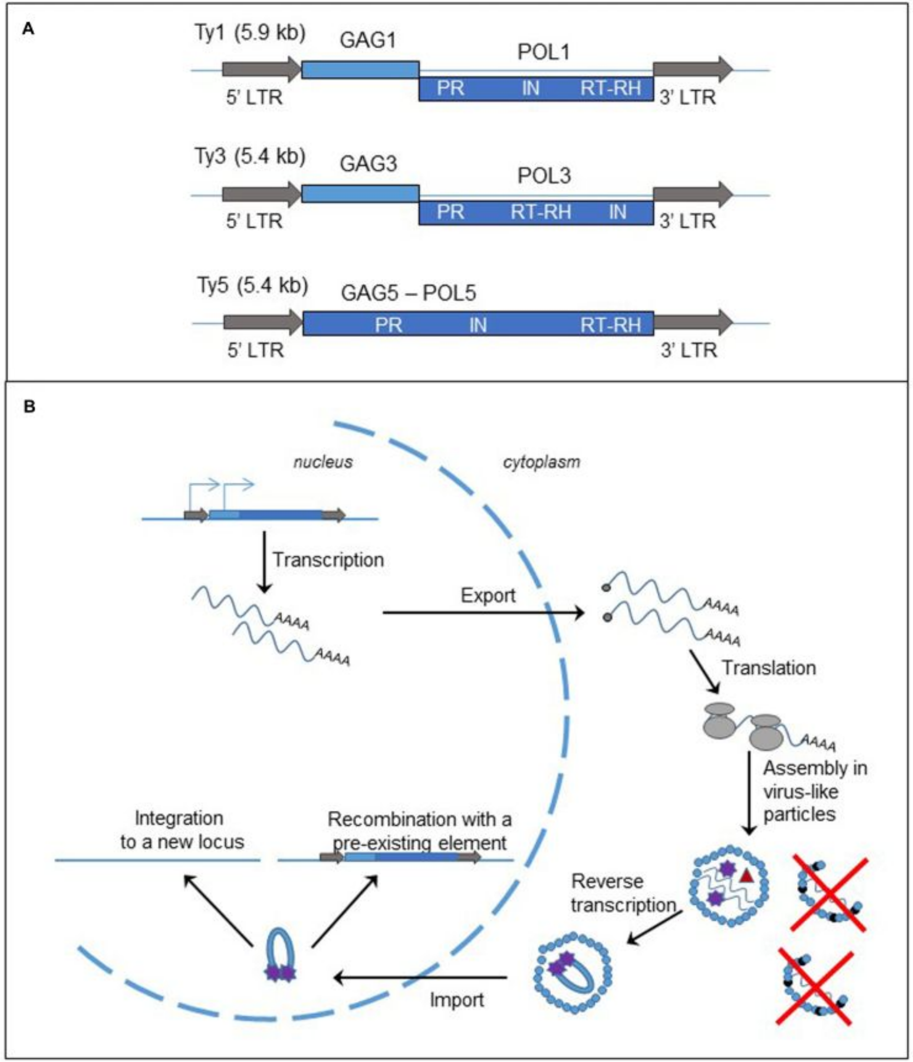
(B) non-LTR Retrotransposons

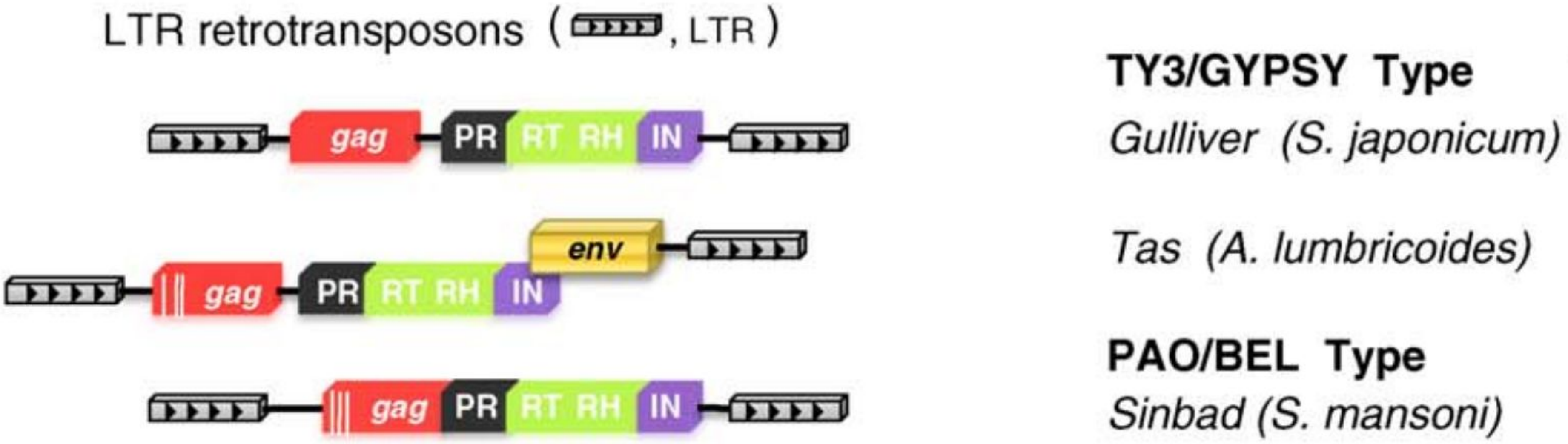
LTR

LTR retrotransoposons
long terminal repeat

3.5 – 10 Kb
LTR ~ 200 to 600pb

protease (PR)
Reverse Transcriptase (RT)
RnaseH (RH)
Integrase (IN)
Long terminal repeats (direct)





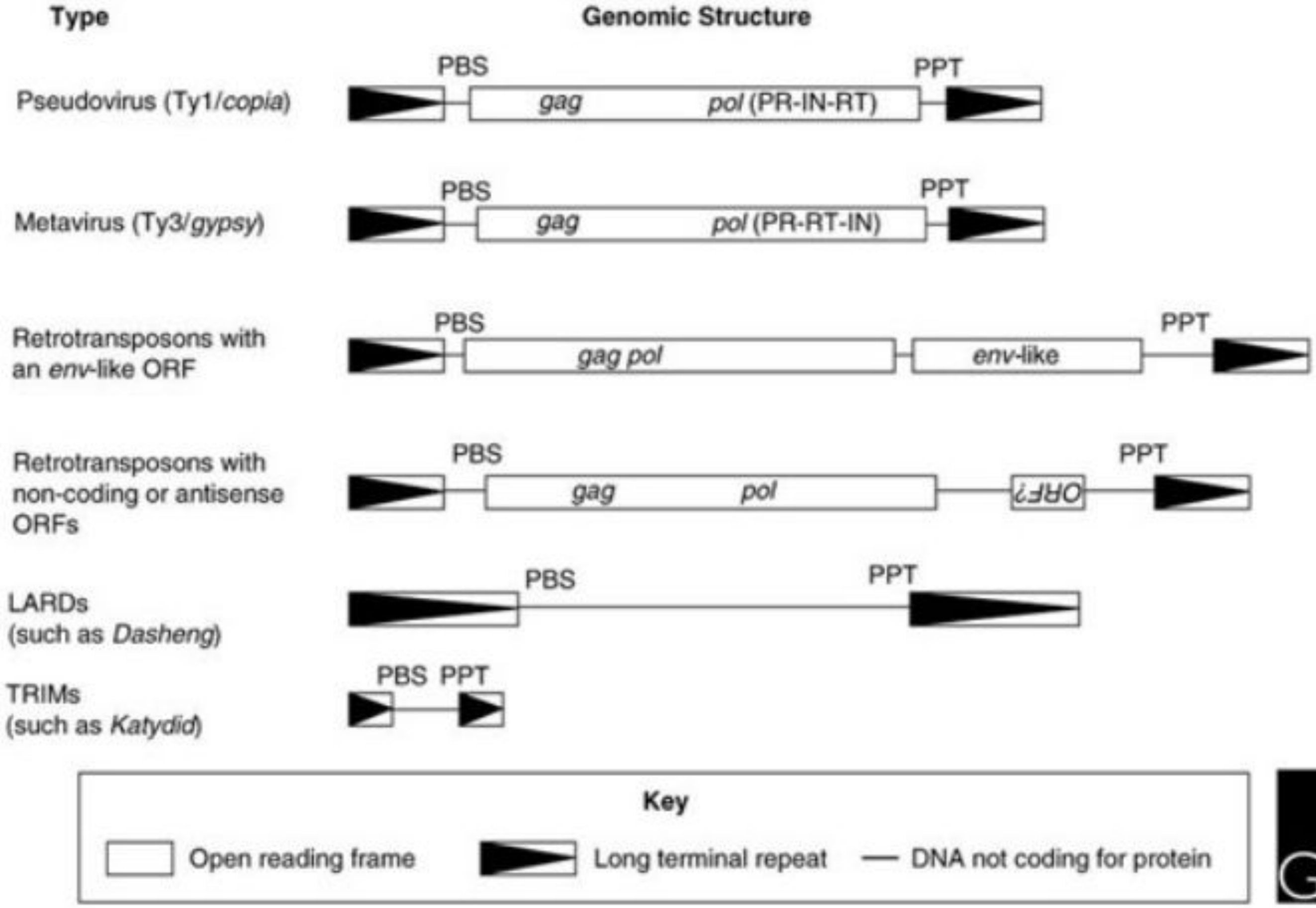
TY3/GYPSY Type

Gulliver (S. japonicum)

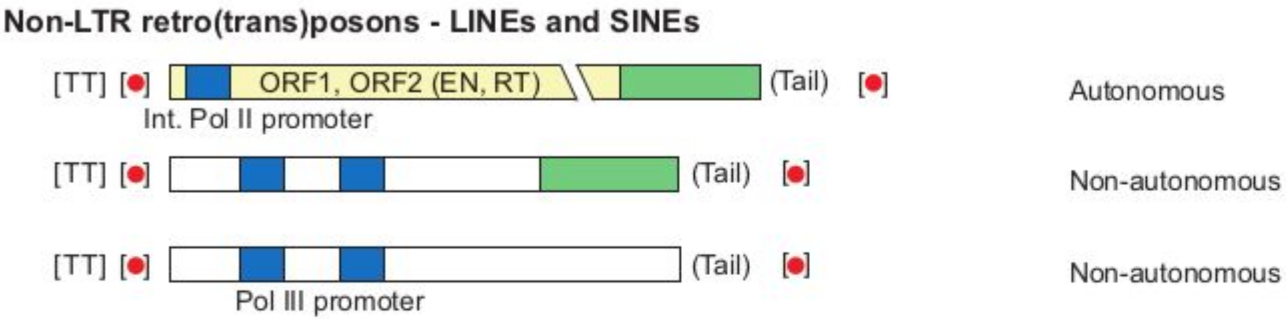
Tas (A. lumbricoides)

PAO/BEL Type

Sinbad (S. mansoni)


Type
Genomic Structure
Pseudovirus (Ty1/*copia*)
PBS
PPT
gag
pol (PR-IN-RT)
Metavirus (Ty3/*gypsy*)
PBS
PPT
gag
pol (PR-RT-IN)
Retrotransposons with an *env*-like ORF
PBS
PPT
gag pol
env-like
Retrotransposons with non-coding or antisense ORFs
PBS
PPT
gag
pol
ORF?
LARDs (such as *Dasheng*)
PBS
PPT
TRIMs (such as *Katydid*)
PBS
PPT
Key
Open reading frame
Long terminal repeat
DNA not coding for protein

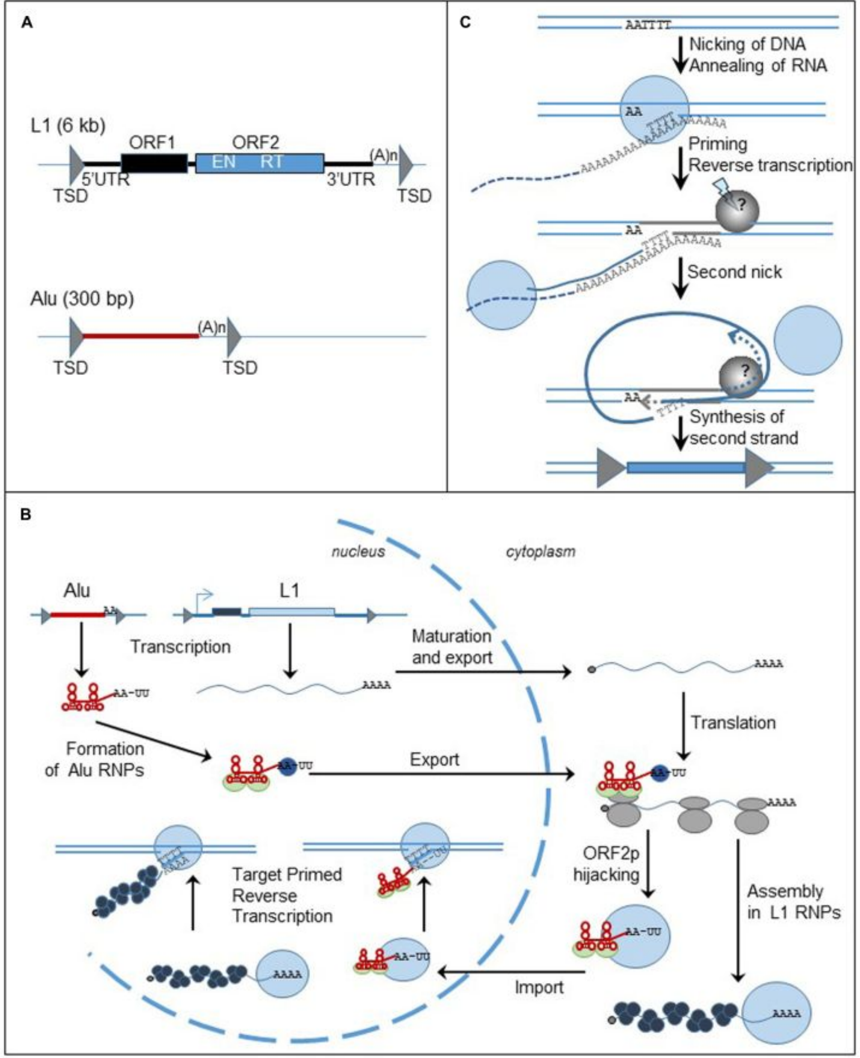
Non-LTR

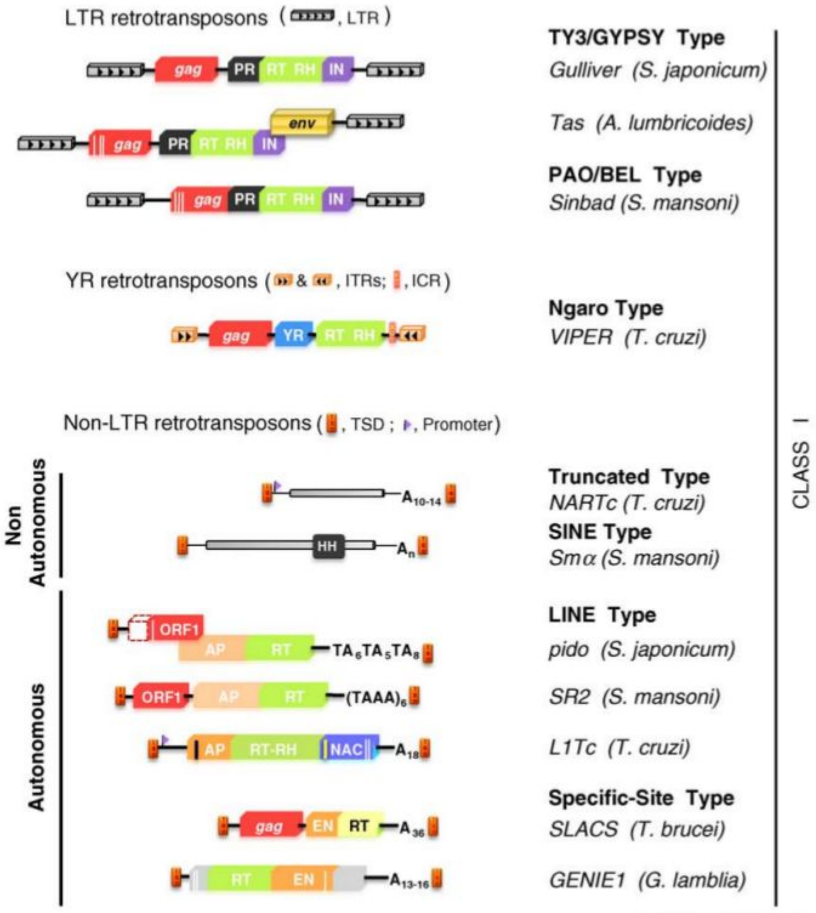

Non-LTR retro(trans)posons - LINEs and SINEs
[TT]
ORF1, ORF2 (EN, RT)
(Tail)
Int. Pol II promoter
Autonomous
[TT]
(Tail)
Non-autonomous
[TT]
(Tail)
Pol III promoter
Non-autonomous

Non-LTR

LINE / SINE

Long and short interspersed elements





TRENDS in Parasitology

Clase I

- **LTR**
- **Non-LTR**

  Penelope
  - Contienen RT y EN
  - Generan TSD
  - Codifican para un ORF
- DIRS
  - Codifican para una RT
  - tienen una INT de la familia de las tyrosine recombinasas
  - Gran distribución en eucariotas (antiguos)


LTR
GAG
POL
ENV
LTR

Retrovirus a partir de ET

- Se asume que los retrovirus se formar a partir de la familia Ty3/Gypsi
- Sus secuencias se encuentran en la mayoría de los organismos
- Está asociada a la adquisición del gen ENV

Hypothesis on the Origin of Viruses from Transposons

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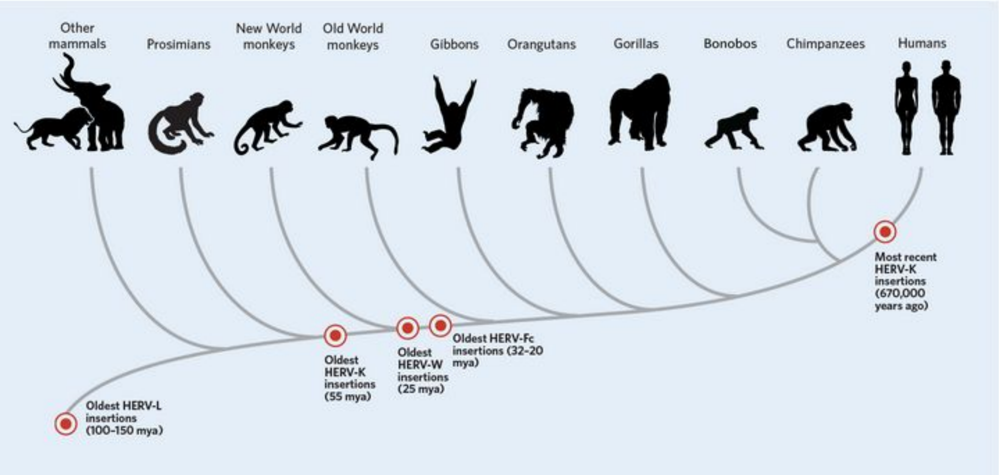
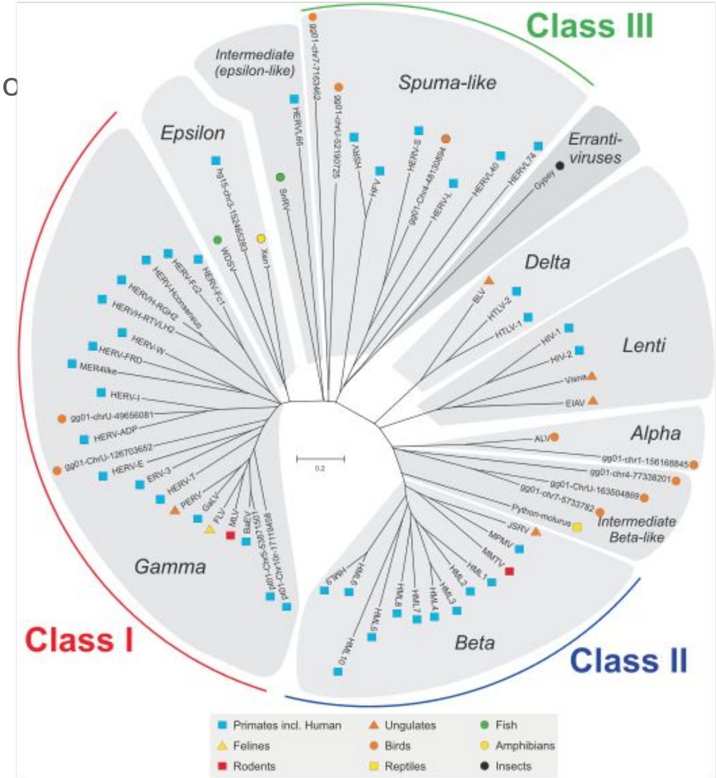
Otros virus a partir de otros ET?

- Virus de ADN se pueden haber originado de ET de ADN
- Debido a la cantidad de mutaciones, muchos virus pueden haber perdido su rastro hacia ET
- Esta hipótesis plantea que al mismo tiempo que perdieron enzimas para integración en el genoma, adquirieron virulencia y capacidad de transmitirse a otras células y organismos

ERV Retrovirus Endógenos

Son elementos virales endógenos que se han insertado en el geno

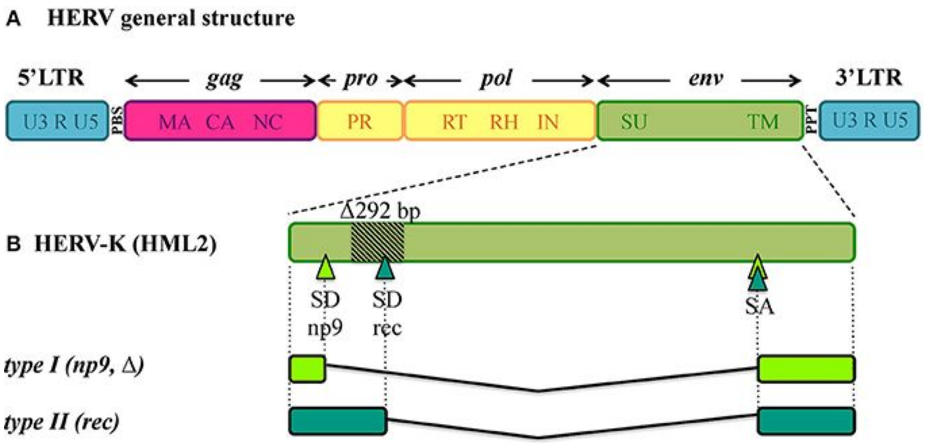


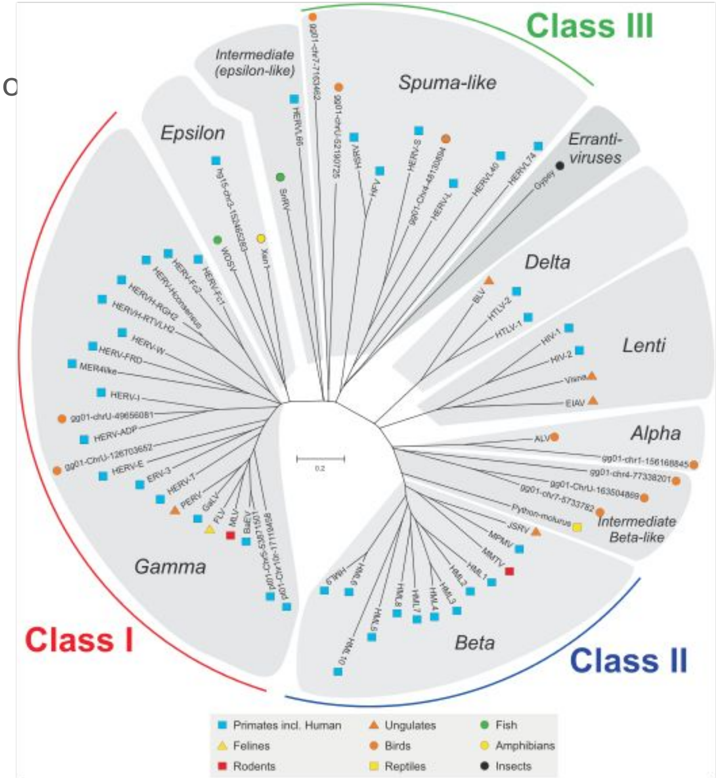


ative unrooted Pol neighbor joining (NJ) dendrogram

ERV Retrovirus Endógenos

Son elementos virales endógenos que se han insertado en el geno





ative unrooted Pol neighbor joining (NJ) dendrogram

Grandi and Tramontano, Front Micriobiol 2018

ERV Retrovirus Endógenos

Research | Open Access | Published: 16 December 2019

HIV-1 Rev interacts with HERV-K RcREs present in the human genome and promotes export of unspliced HERV-K proviral RNA

Laurie R. Gray, Rachel E. Jackson, Patrick E. H. Jackson, Stefan Bekiranov, David Rekosh & Marie-Louise Hammarskjöld

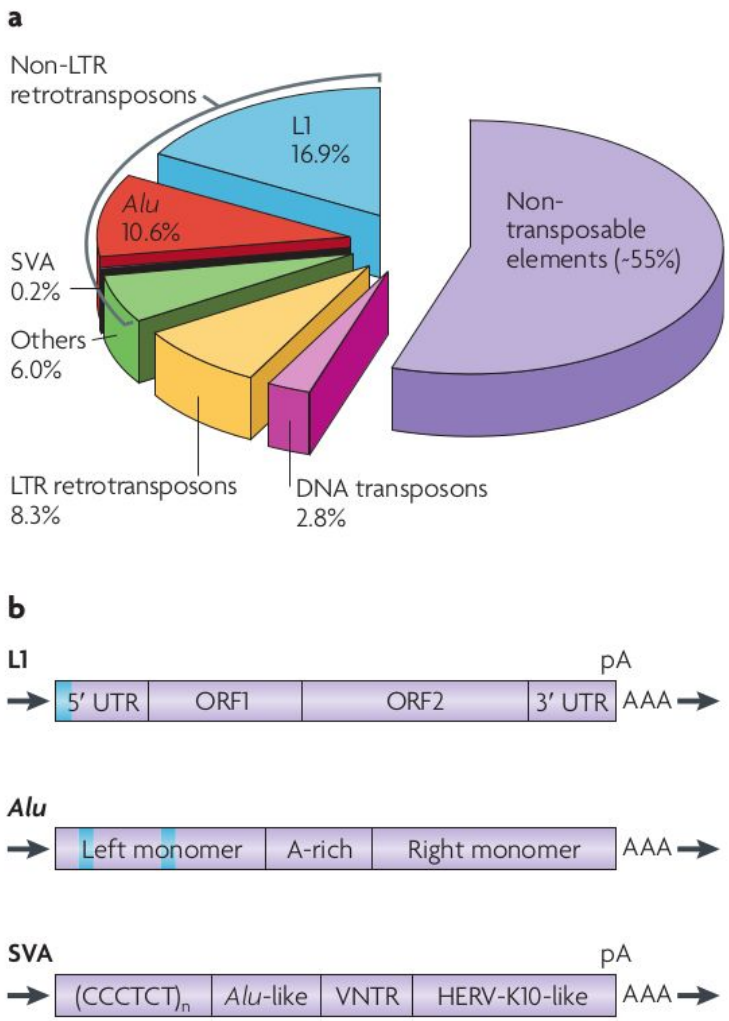
Retrovirology **16**, Article number: 40 (2019) | Cite this article

2025 Accesses | **4** Citations | **5** Altmetric | Metrics

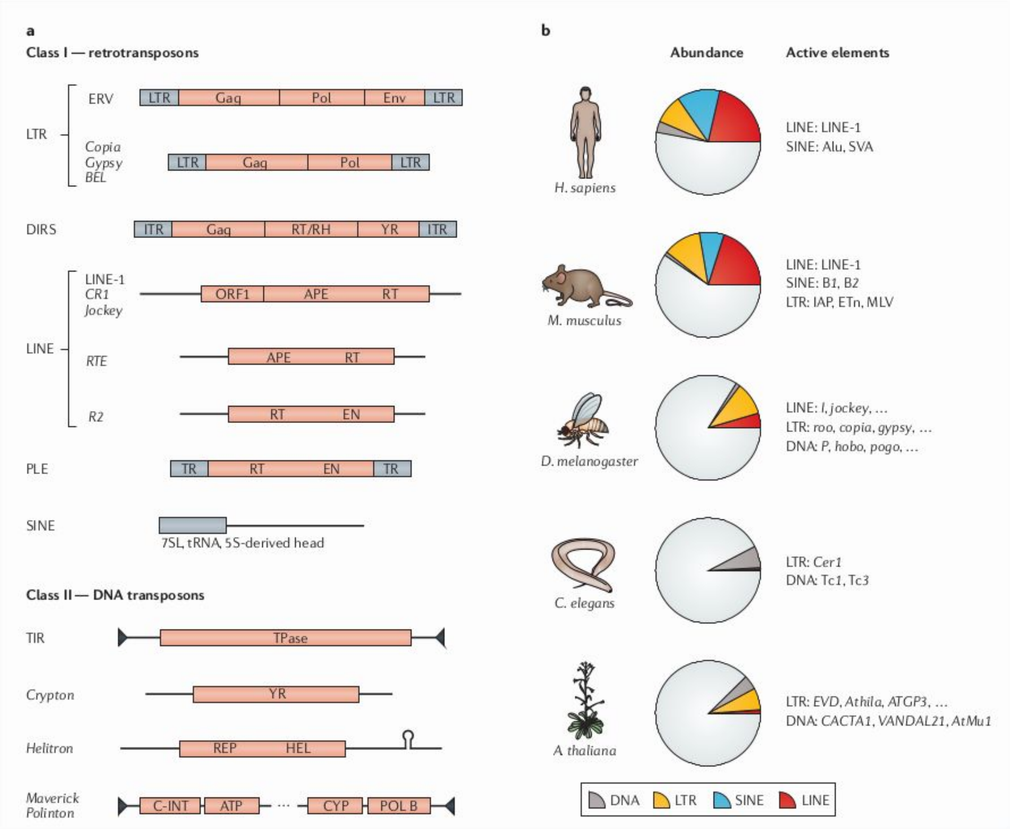
Genoma humano

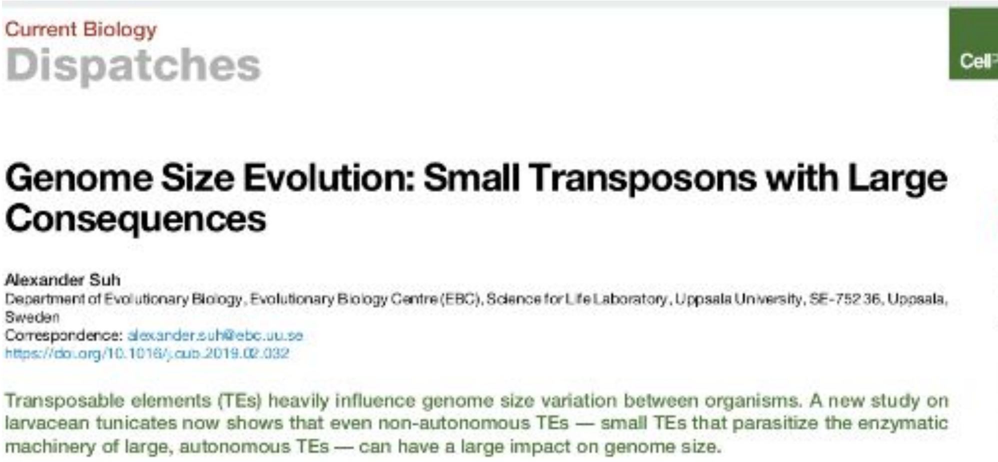
The impact of retrotransposons on human genome evolution

Richard Cordaux and Mark A. Batzer‡*



Moduladores del tamaño genómico





Current Biology
Dispatches

Genome Size Evolution: Small Transposons with Large Consequences

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Correspondence: alexander.suh@ebc.uu.se
https://doi.org/10.1016/j.cub.2019.02.032

Transposable elements (TEs) heavily influence genome size variation between organisms. A new study on larvacean tunicates now shows that even non-autonomous TEs — small TEs that parasitize the enzymatic machinery of large, autonomous TEs — can have a large impact on genome size.

Blommaert et al. BMC Genomics (2019) 20:466
https://doi.org/10.1186/s12864-019-5859-y
BMC Genomics

RESEARCH ARTICLE Open Access

Small, but surprisingly repetitive genomes: transposon expansion and not polyploidy has driven a doubling in genome size in a metazoan species complex

J. Blommaert[1], S. Riss[1], B. Hecox-Lea[2], D. B. Mark Welch[2] and C. P. Stelzer[1*]

genes
MDPI

Review

Genome Size Diversity and Its Impact on the Evolution of Land Plants

Jaume Pellicer *, Oriane Hidalgo, Steven Dodsworth and Ilia J. Leitch

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Received: 10 January 2018; Accepted: 5 February 2018; Published: 14 February 2018

Current Biology
Report

Massive Changes of Genome Size Driven by Expansions of Non-autonomous Transposable Elements

Magali Naville,[1,5] Simon Henriet,[2,5] Ian Warren,[1] Sara Sumic,[2] Magnus Reeve,[2,4] Jean-Nicolas Volff,[1,*] and Daniel Chourrout[2,3,6,*]

“Molecular domestication”

- Una de las funciones más importantes de los ET en la evolución de los organismos
- Dada su diversidad y la variedad de los mecanismos de transposición, codifican para muchas proteínas con funciones o dominios involucrados en la catálisis o interacción con ADN, ARN y otras proteínas
- Los ET pueden suministrar módulos funcionales para generar nuevos genes

TOP 10: Telomerasa

TERT Telomerase reverse transcriptasa

- Es uno de los eventos de domesticación más antiguos
- Lo comparten la mayoría de los eucariotas
- Responsable de los telómeros y los cromosomas lineales!

-

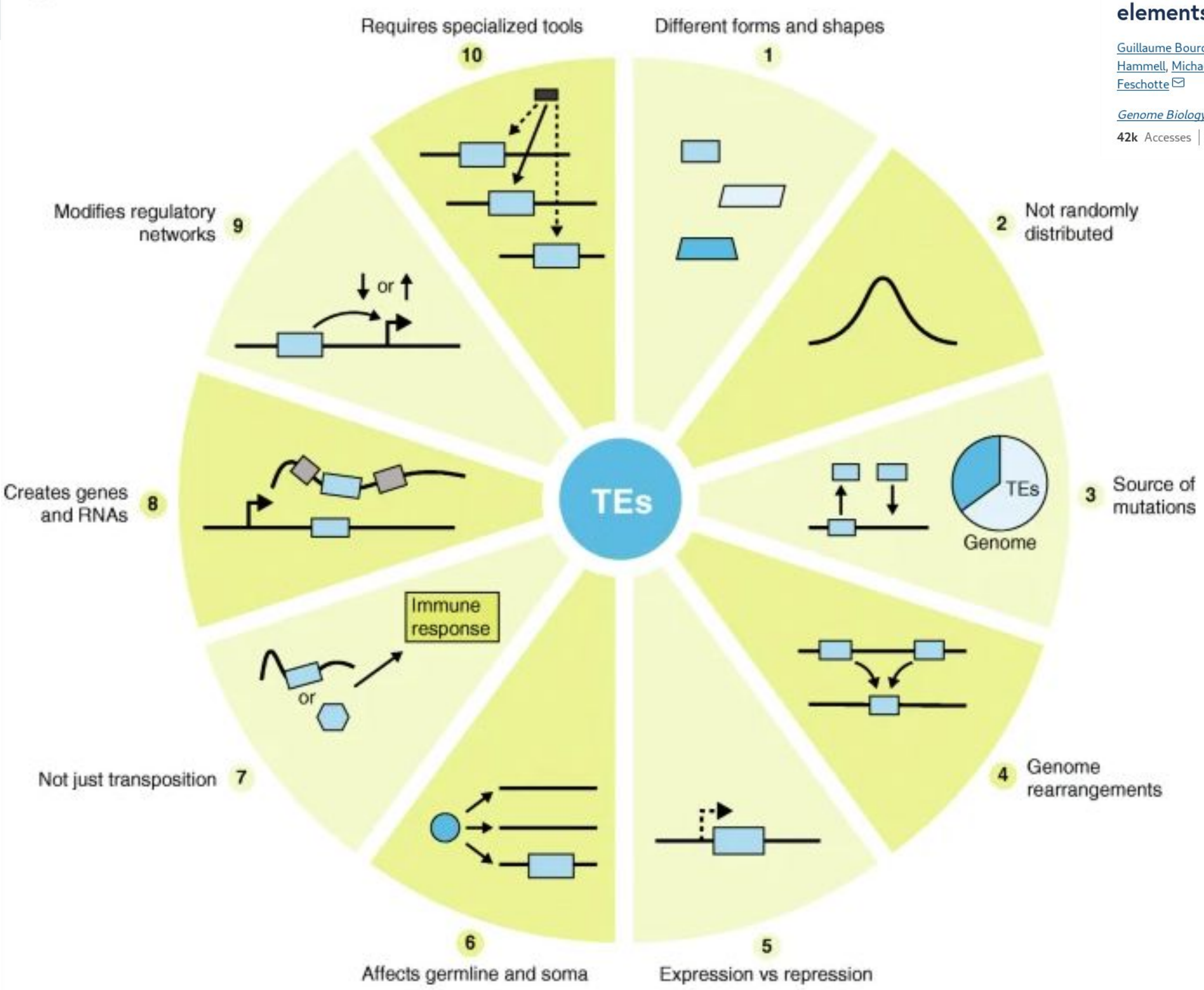
> Proc Natl Acad Sci U S A. 2007 May 29;104(22):9352-7. doi: 10.1073/pnas.0702741104. Epub 2007 May 4.

Telomere-associated endonuclease-deficient Penelope-like retroelements in diverse eukaryotes

Eugene A Gladyshev [1], Irina R Arkhipova

Affiliations + expand

Fig. 2



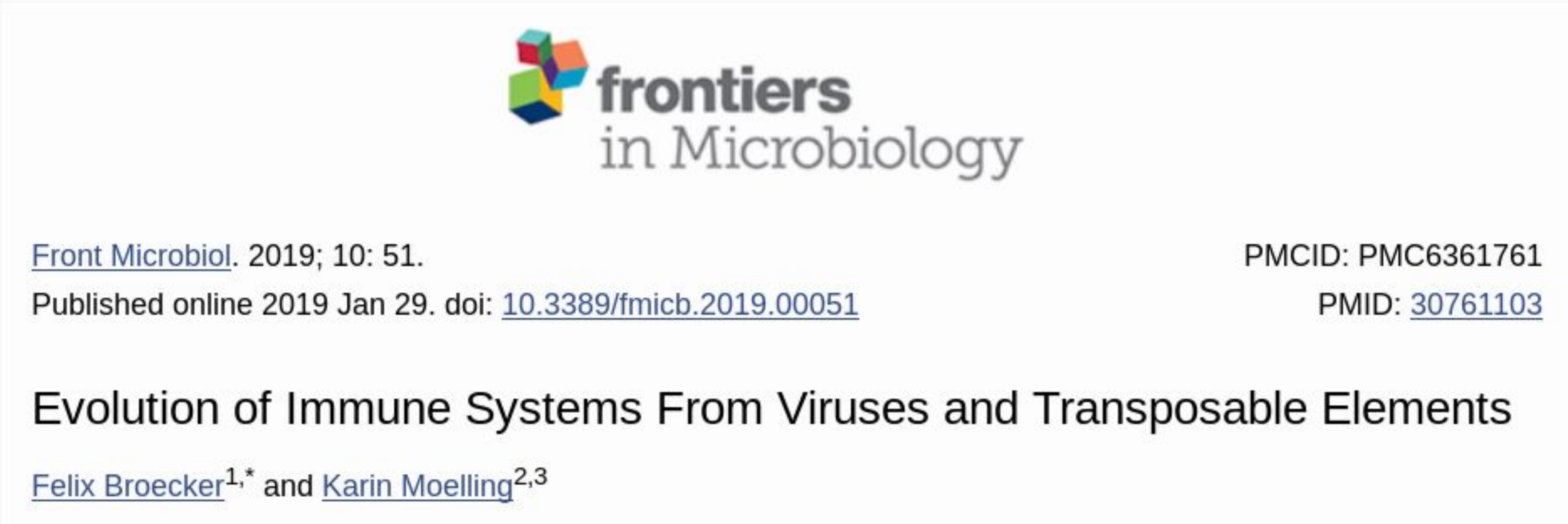

Review | Open Access | Published: 19 November 2018

Ten things you should know about transposable elements

Guillaume Bourque, Kathleen H. Burns, Mary Gehring, Vera Gorbunova, Andrei Seluanov, Molly Hammell, Michaël Imbeault, Zsuzsanna Izsvák, Henry L. Levin, Todd S. Macfarlan, Dixie L. Mager & Cédric Feschotte

Genome Biology **19**, Article number: 199 (2018) | Cite this article

42k Accesses | **73** Citations | **261** Altmetric | Metrics



frontiers
in Microbiology

Front Microbiol. 2019; 10: 51.
Published online 2019 Jan 29. doi: 10.3389/fmicb.2019.00051

PMCID: PMC6361761
PMID: 30761103

Evolution of Immune Systems From Viruses and Transposable Elements

Felix Broecker[1,*] and Karin Moelling[2,3]

Continuará