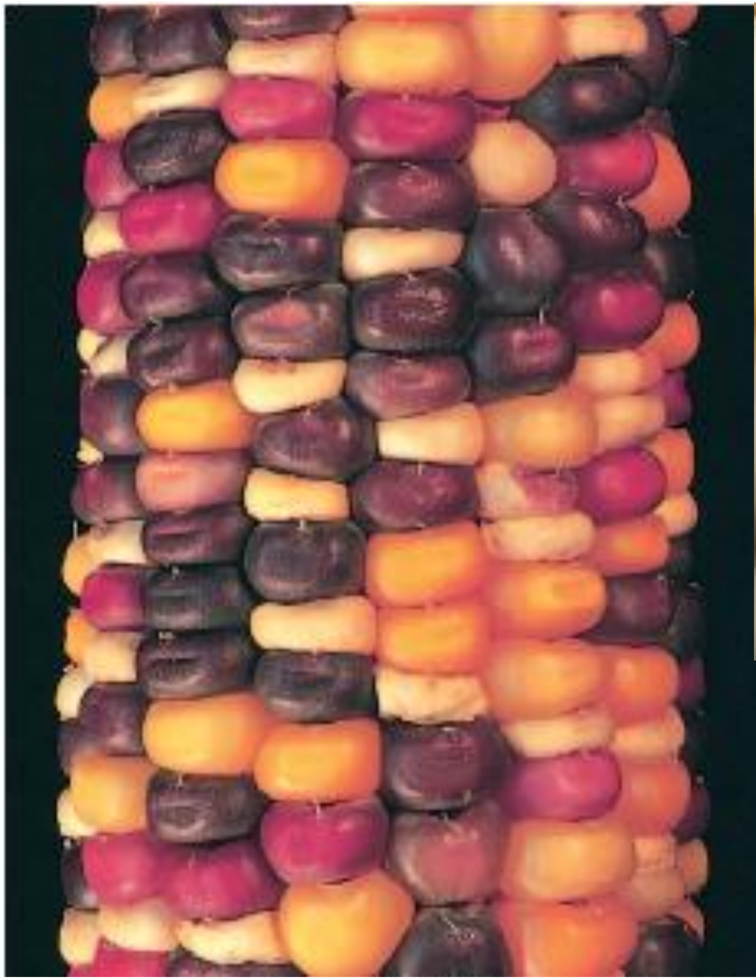
TRANSPOSABLE GENETIC ELEMENTS



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It is a general assumption that genes do not move from one position to the other on the chromosomes. The genetic mapping is also based on the same assumption. Majority of the genes obey this assumption and most of the genes occupy fixed positions on the chromosomes. Thus, overall structure of the genetic map is practically stable.

However, beginning in 1940s scientists have found certain DNA sequences can actually change positions.

Transposable Genetic Elements

Also known as **mobile elements**, **jumping genes**, **genomic parasites**, and **selfish DNA**

Some genes move from place to place inserting themselves into a variety of locations, causing mutations in genes and changing the ways other genes are expressed.

These wanderers are called *transposable elements* (TEs)

In other words……..

Segments of the genome that are capable of moving around to different locations are known as transposable genetic elements or transposons.

The movement is known as transposition.

What do they carry?

-Transposable elements usually are flanked by repeated base sequences.

-They often carry transposase genes that gives the transposition ability.

-They often carry antibiotic resistance genes.

-Transposons can insert into plasmids which can be transferred to recipient cells by conjugation.

Discovery of Transposable Genetic Elements

- **TEs were discovered from corn (Maize) while doing genetical experiments on changing kernel colours.**

- **The genes controlling kernel colour kept moving from one locus to another producing different colour patterns.**

Discoverer

Babara McClintock (1948)

She called them “controlling elements”

Was awarded Nobel Prize in Medicine and Physiology in 1983

Because the transposons can insert within genes or regulatory sequence of a gene, having little selectivity in their choice of insertion sites, this results in the complete disruption of gene function or alteration in the expression of a gene. These disruptions lead to the discovery of transposable elements by Barbara McClintock.

Figure: Example of corn cob showing color variegation due to transposition

Transposition

Transposition is a type of genetic recombination in which certain genetic elements move from one site on DNA to another.

This occurs through recombination between the DNA sequences at the ends of the transposons and a sequence in the host DNA with little sequence selectivity.

Two basic types of TRANSPOSITION

- **Conservative transposition (Cut & Paste)**

An element leaves one location and inserts in another.

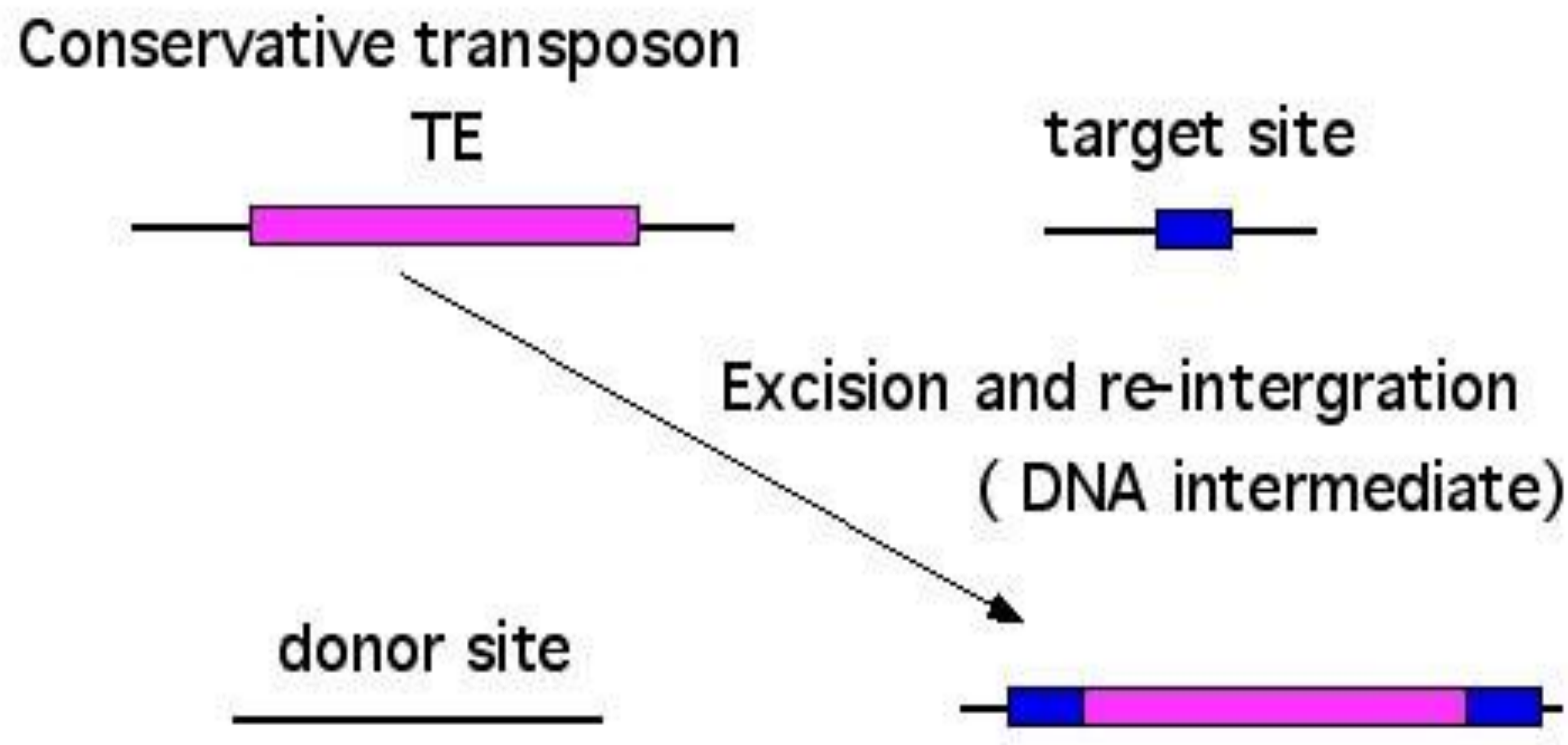
Does not change overall copy number.

- **Replicative transposition**

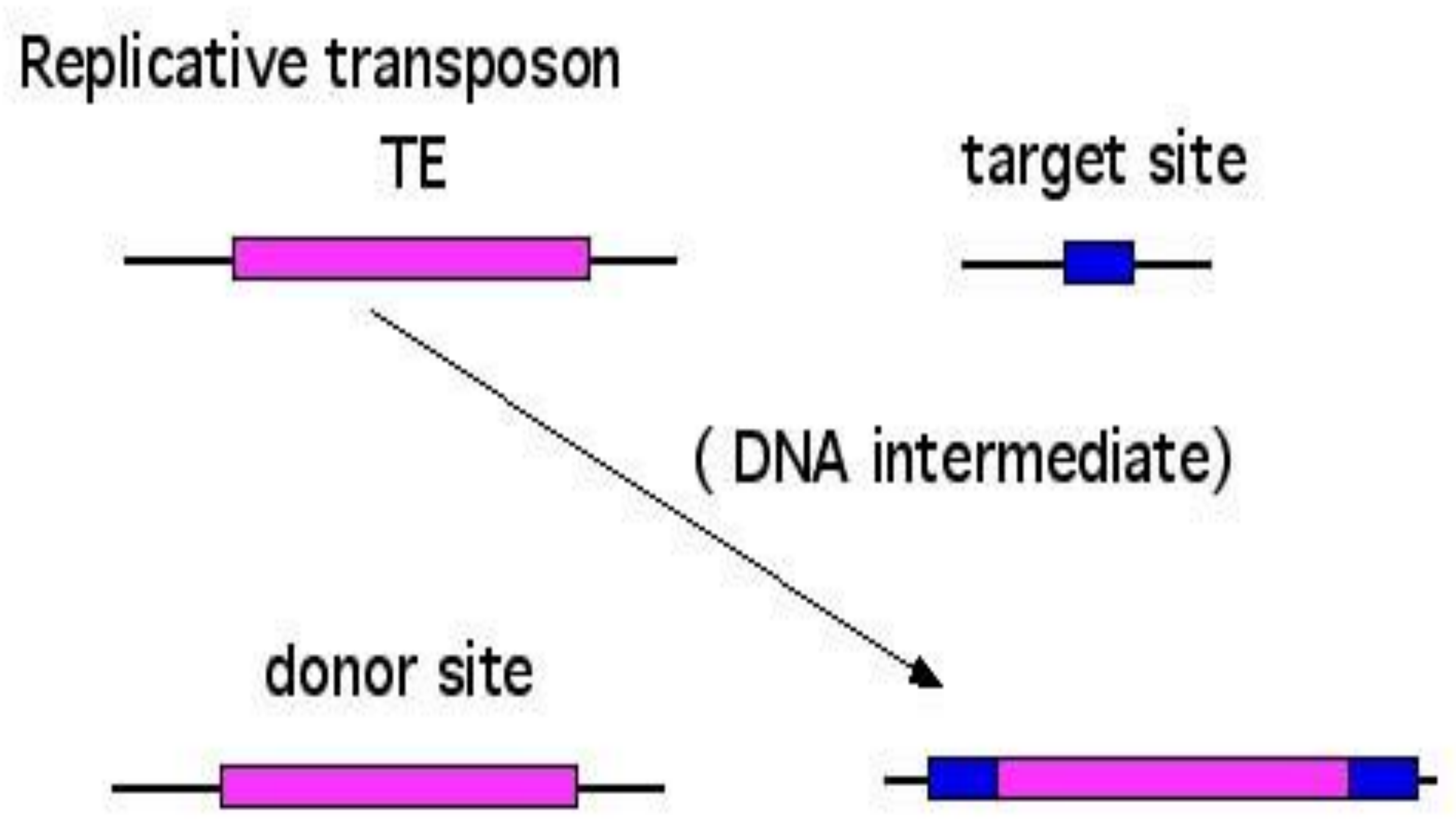
A copy of a transposable element is made and this inserts in a new place.

This is the most common form of transposition.

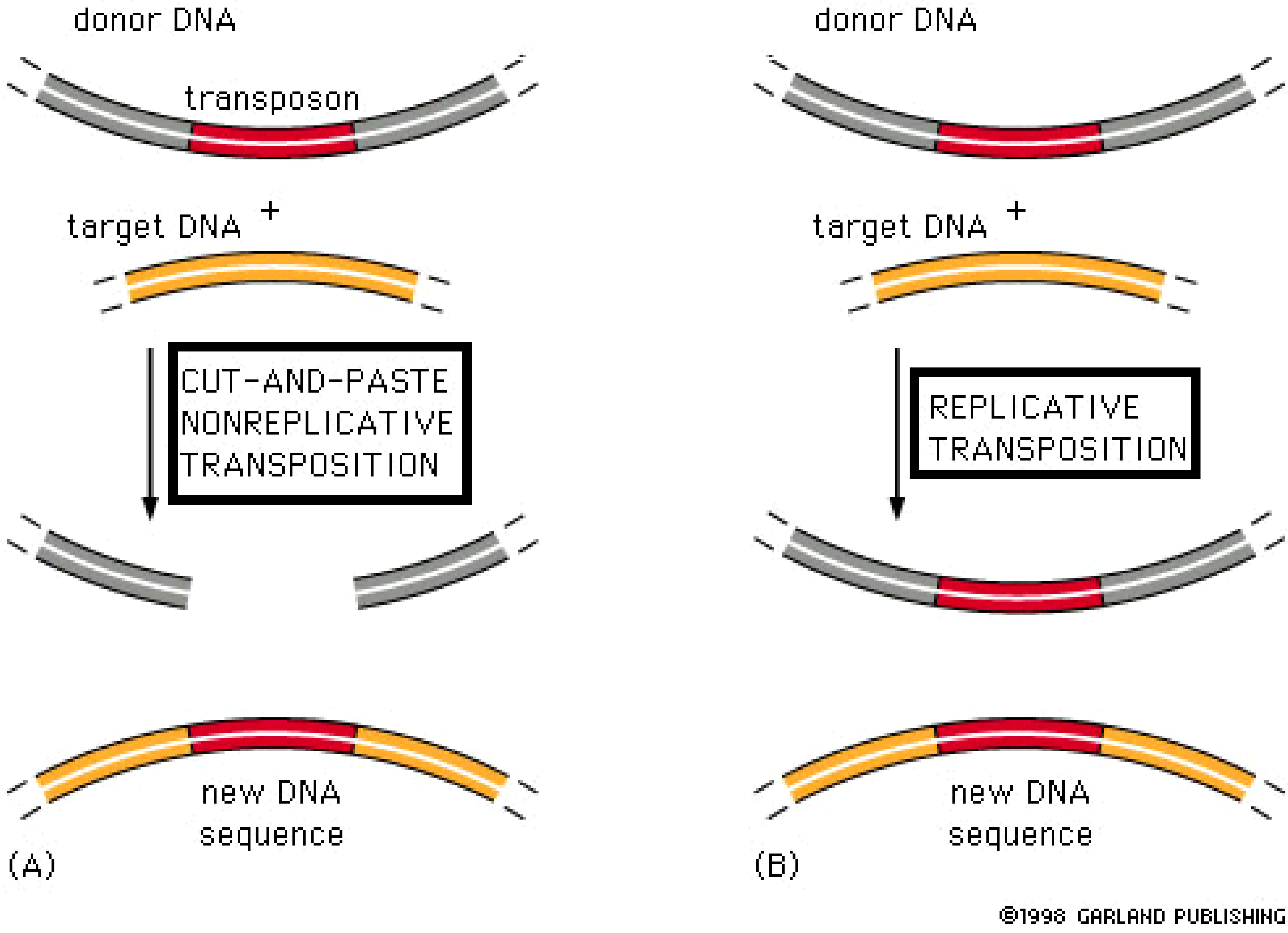
CONSERVATIVE TRANSPOSITION



REPLICATIVE TRANSPOSITION



CONSERVATIVE AND REPLICATIVE TRANSPOSITIONS



Transposable Elements based on Mechanism of action or pathway can be:

DNA TRANSPOSONS

Which Directly synthesize a DNA copy of themselves

RETROTRANSPOSONS

That Generate an RNA intermediate which is then reverse-transcribed back into DNA

DNA TRANSPOSONS

- They transpose via DNA intermediates.
- A common feature of DNA transposons is its flanking by short inverted repeat sequences.
- The enzyme transposase recognizes these sequences, creates a stem/loop structure, and excises the loop from the region of the genome.
- The excised loop can then be inserted into another region of the genome.

RETROTRANSPOSONS

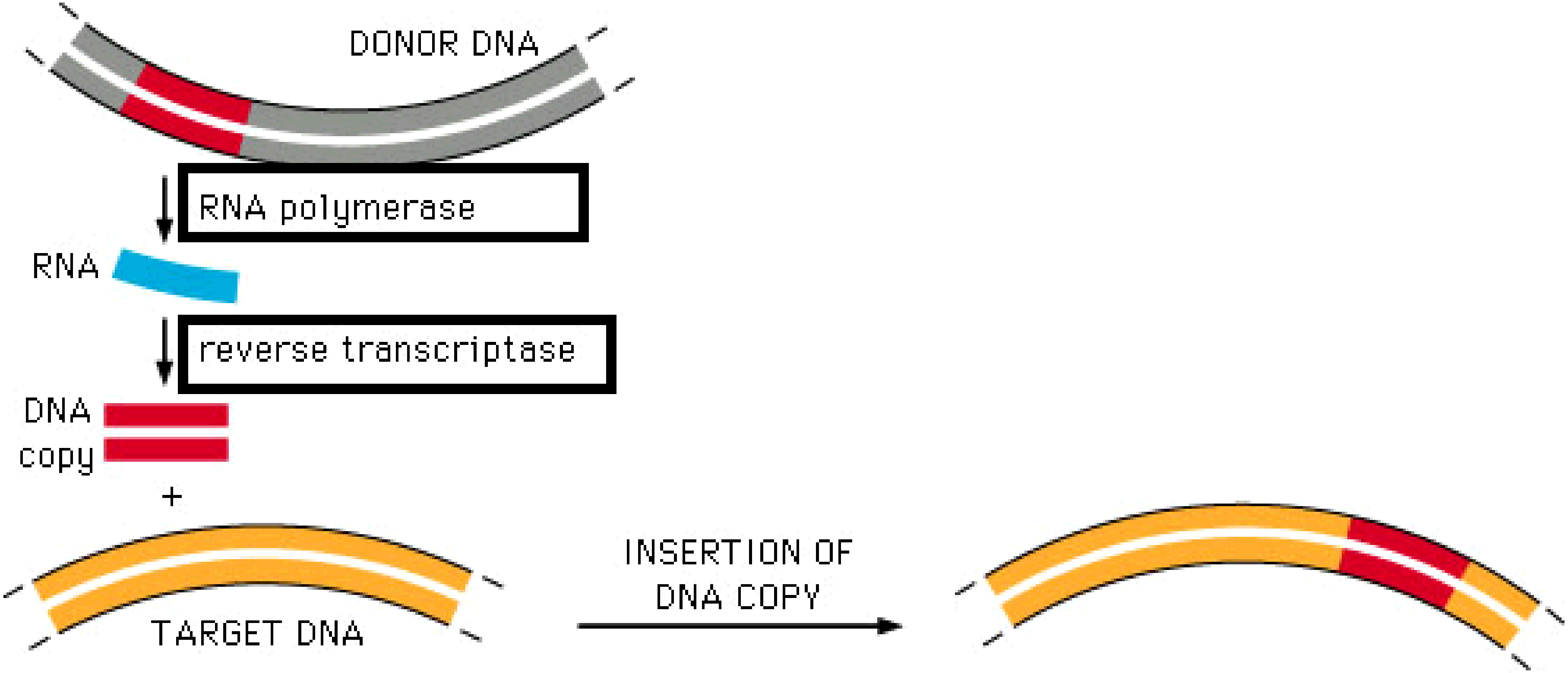
- **They transpose via RNA intermediates.**

- **The RNA is copied by the enzyme reverse transcriptase into DNA.**

- **The DNA integrates into the genome.**

- **Retroelements are found in all eukaryotes such as *Tos* in rice, *copia* in animals and *Ty1* in yeast.**

<u>Retrotransponons</u> – Which are perhaps unique to eukaryotes.



Transposable Genetic Elements based on their Mechanism of Action can be:

Autonomous

Can move independently within the genome

Non-autonomous

Require other elements to move

1. **Autonomous transposons: carry a pair of terminal inverted repeats and a transposase gene; can function independently.**

2. **Nonautonomous transposons: carry the terminal inverted repeats but not the functional transposase; need the transposase encoded by autonomous transposons to enable transposition**

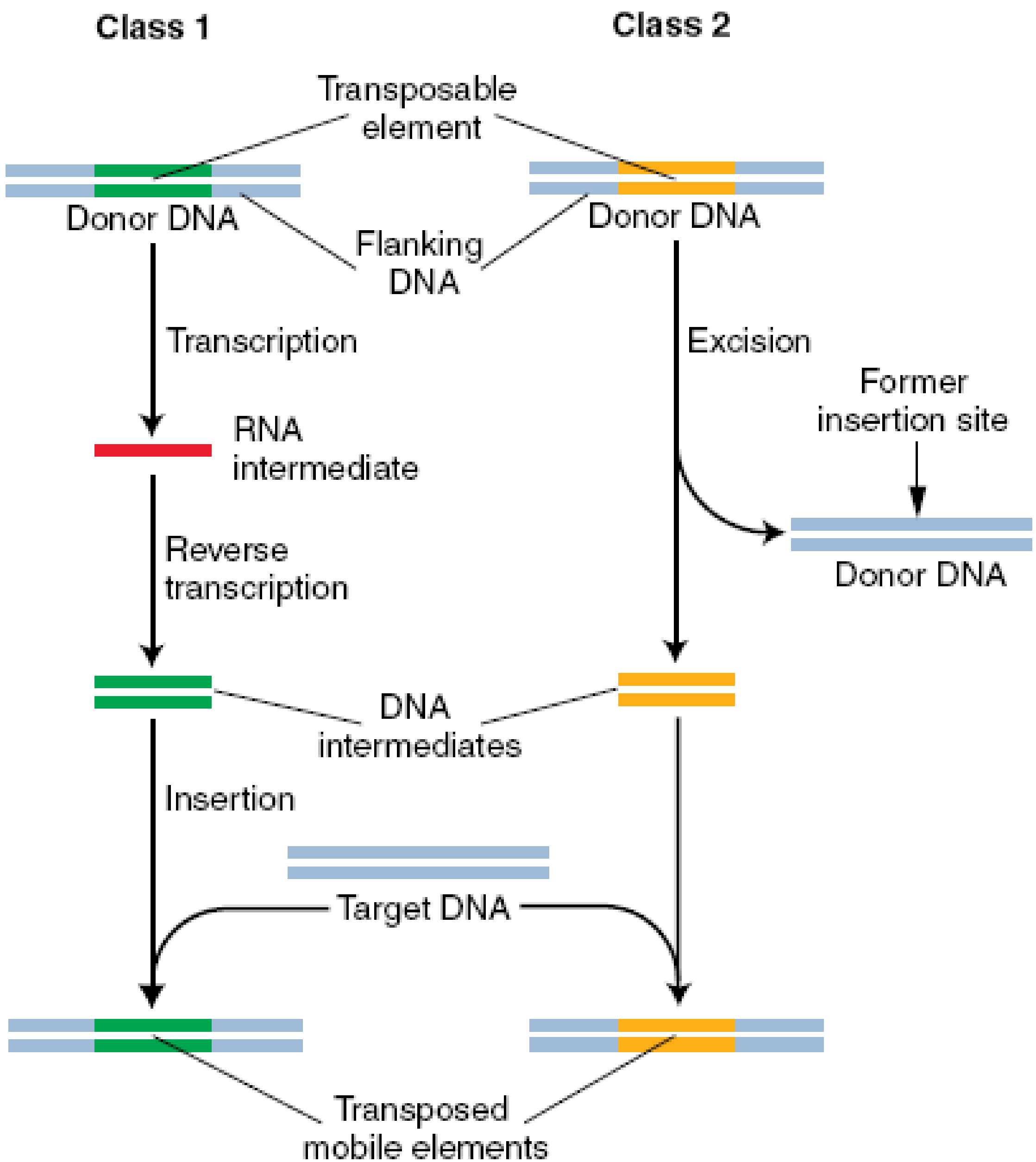
TWO MORE CLASSES OF TEs

Class 1 Transposons

Remain permanently once inserted; they cannot be excised from the donor site, but they are still considered to be mobile because their copies can insert into new target DNA

Class 2 Transposons

Known as DNA elements, they move by excision from one site in the genome to another. First transposable elements discovered genetically in maize were class 2 elements.



Class 1
Class 2
Transposable element
Donor DNA
Flanking DNA
Donor DNA
Transcription
Excision
Former insertion site
RNA intermediate
Donor DNA
Reverse transcription
DNA intermediates
Insertion
Target DNA
Transposed mobile elements

There are three principle groups of transposable elements

1. DNA transposons
2. Viral-like retrotransposons including the retrovirus, which are also called LTR (Long Terminal Repeat) retrotransposons
3. Poly-A retrotransposons, also called nonviral or nonLTR retrotransposons.

ANOTHER CLASS OF TEs

“Degenerate” transposons

- **many naturally occurring transposable elements have suffered mutation and are no longer active.**

- **some of these may have cis-acting end mutations and cannot be mobilized.**

- **others may have intact ends but no transposase: these can be mobilized by an element called “autonomous” element.**

The Biological Relevance of Transposons

1. **Transposons are present in the genomes of all life-forms.**

2. **Almost half of the human genome is derived from transposable elements.**

3. **Transposon-related sequences can make up huge fractions of the genome of an organism (50% of human and maize genome).**

4. **The transposon content in different genomes is highly variable.**

Significance of transposons

For mutagenesis studies:

- The insertion and excision of transposable elements results in changes to the DNA at the transposition site.

- The transposition can be identified when a known DNA sequence or selection markers are inserted within the elements.

Significance of transposons

The genetic recombination mechanisms of transposition are also used for other functions than the movement of transposons, such as integration of some virus into the host genome and some DNA rearrangement to alter gene expression .

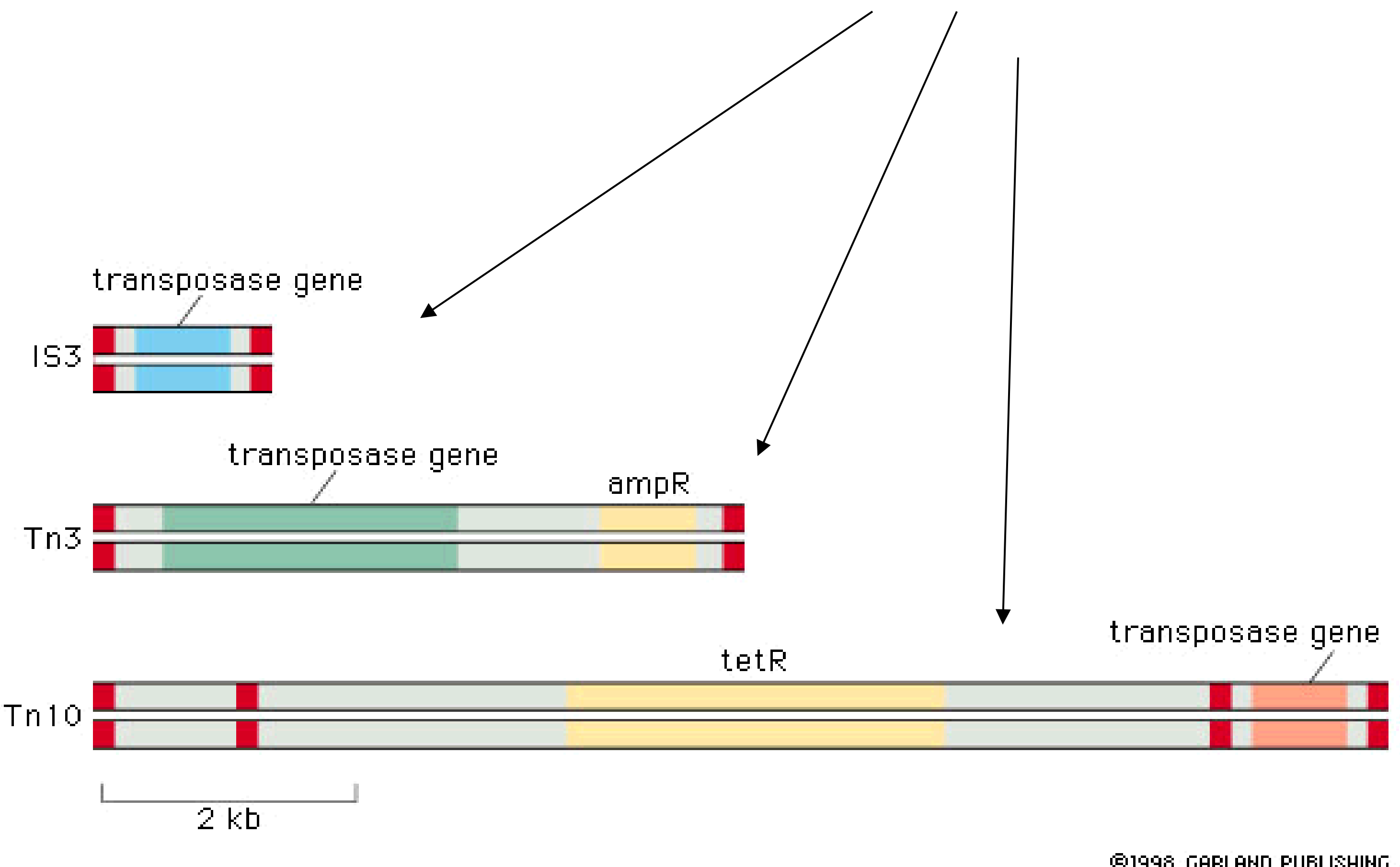
Significance of transposons…..

Evolution of genomes has been accelerated by transposable elements as they are a significant source of mutation.

They provide opportunities for rearrangements of the genome by serving as targets of recombination.

In some organisms, spontaneous mutations result due to insertion into coding sequences of a gene or into its regulatory region.

Transposable Elements (transposons) create more genetic diversity. Range from several hundred to tens of thousands of base pairs. Typical lab *E. coli* contains 10-20 different transposons, with many having multiple copies. Transposons move within a DNA molecule by using a special recombination enzymes - transposases



Significance of transposons:

- **Transposable elements in grasses are responsible for genome size differences. The grasses, including barley, rice, sorghum, and maize, arose from a common ancestor about 70 million years ago. Since that time, the transposable elements have accumulated to different levels in each species. Chromosomes are larger in maize and barley, whose genomes contain large amounts of LTR retrotransposons.**

Transposons as Molecular Biology Tools

- **Transposons can be used to facilitate cloning of genes, identify regulatory elements, and produce transgenic organisms.**

OTHER USES OF TRANSPOSONS

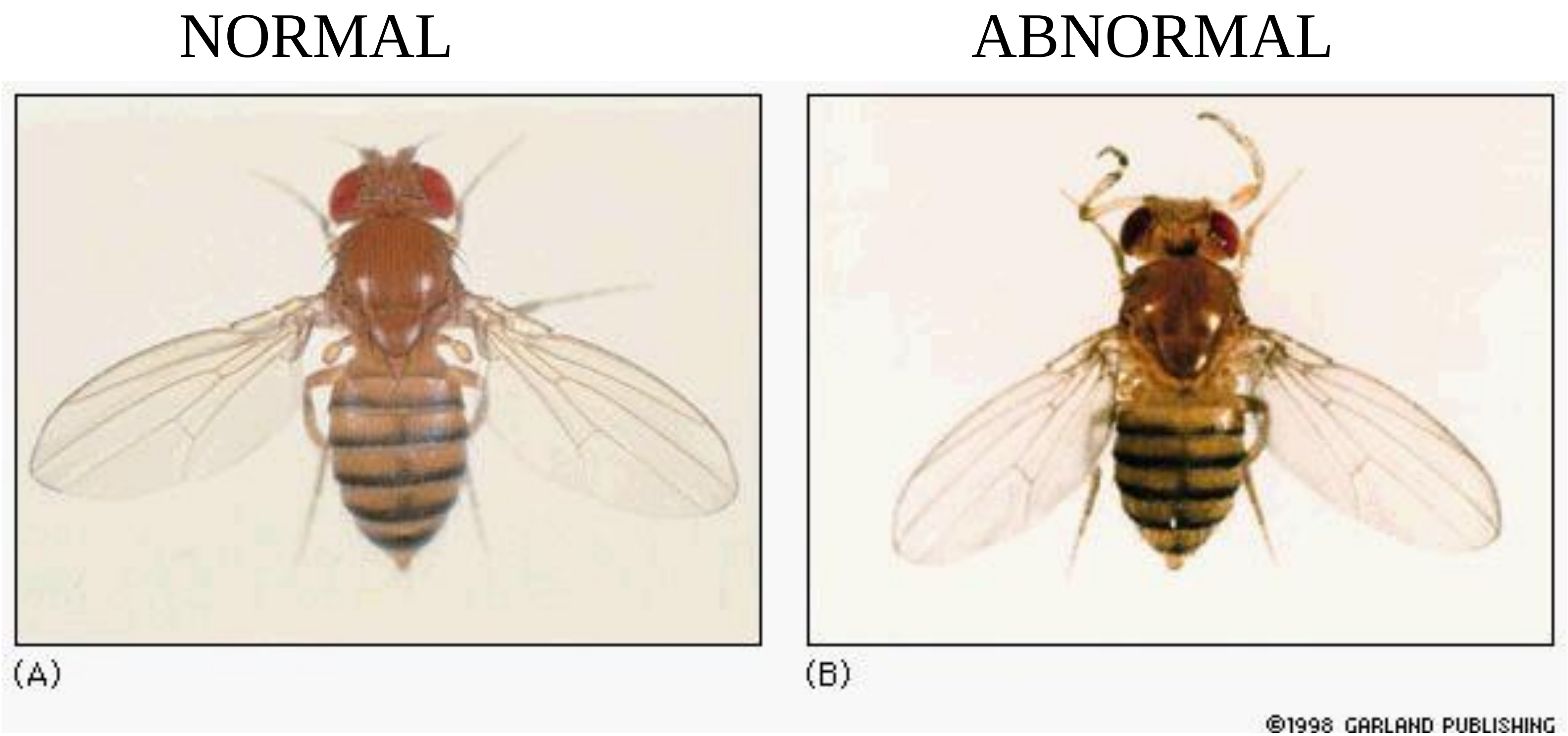
- In prokaryotes: as conventional antibiotic-resistance marker.

- In eukaryotes: for generation of insertion mutations, gene mapping, gene cloning, producing transgenic organisms etc.

TO SUM UP: TRANSPOSONS-

- are “mobile genetic elements”.
- comprise 45% of human chromosomal DNA which is “middle repetitive DNA”.
- contribute to spontaneous mutation, genetic rearrangements, horizontal transfer of genetic material.
- help in speciation and genomic change (in bacteria they are often associated with antibiotic resistance genes).
- cells must depress transposition to insure genetic stability.

DNA rearrangement caused by a transposable element can produce a dramatic change in the organism.



Thanks