Transpositions

- Cytologically invisible sequence rearrangement:
  - movement of a segment of DNA from one location to another in the genome.
  - Not a translocation….
- This may be a transfer of DNA or a duplication of DNA.
- The sequences that cause transpostions
  - are called transposable elements,
  - have specific characteristics,
  - notably the potential to propagate themselves.
- Transposable elements are found in virtually all organisms.

Types of Transposable Elements

- Transposons:
  - Move their DNA directly without the requirement of an RNA intermediate.

- Retroposons:
  - Copy and then move the copied DNA
  - via reverse transcription of an RNA intermediate.

Transposons

- Encode an enzyme called Transposase.
- Rather than converting RNA to DNA, this enzyme:
  - directly removes the DNA sequence and
  - inserts it in another location.
- Transposons usually have inverted repeats (IR) on either side upstream and downstream.

Transposons encode transposase enzymes that catalyze events of transposition

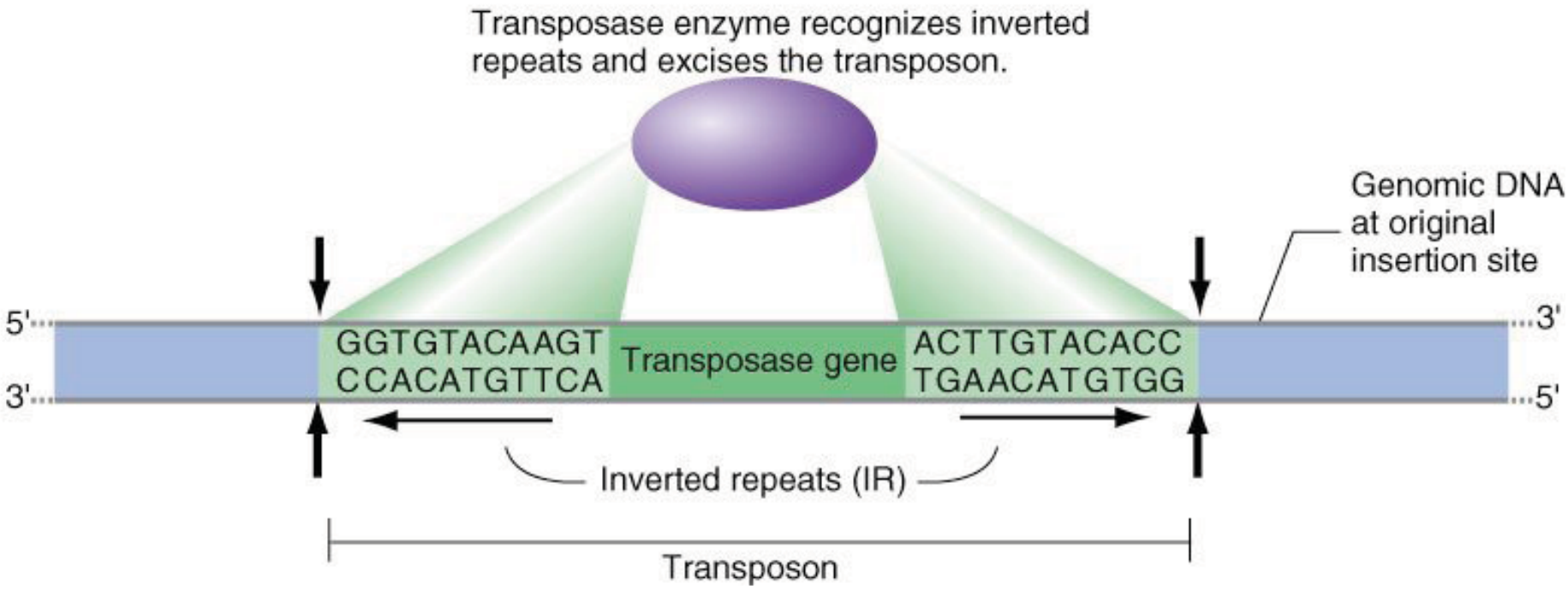


Fig. 13.24 a

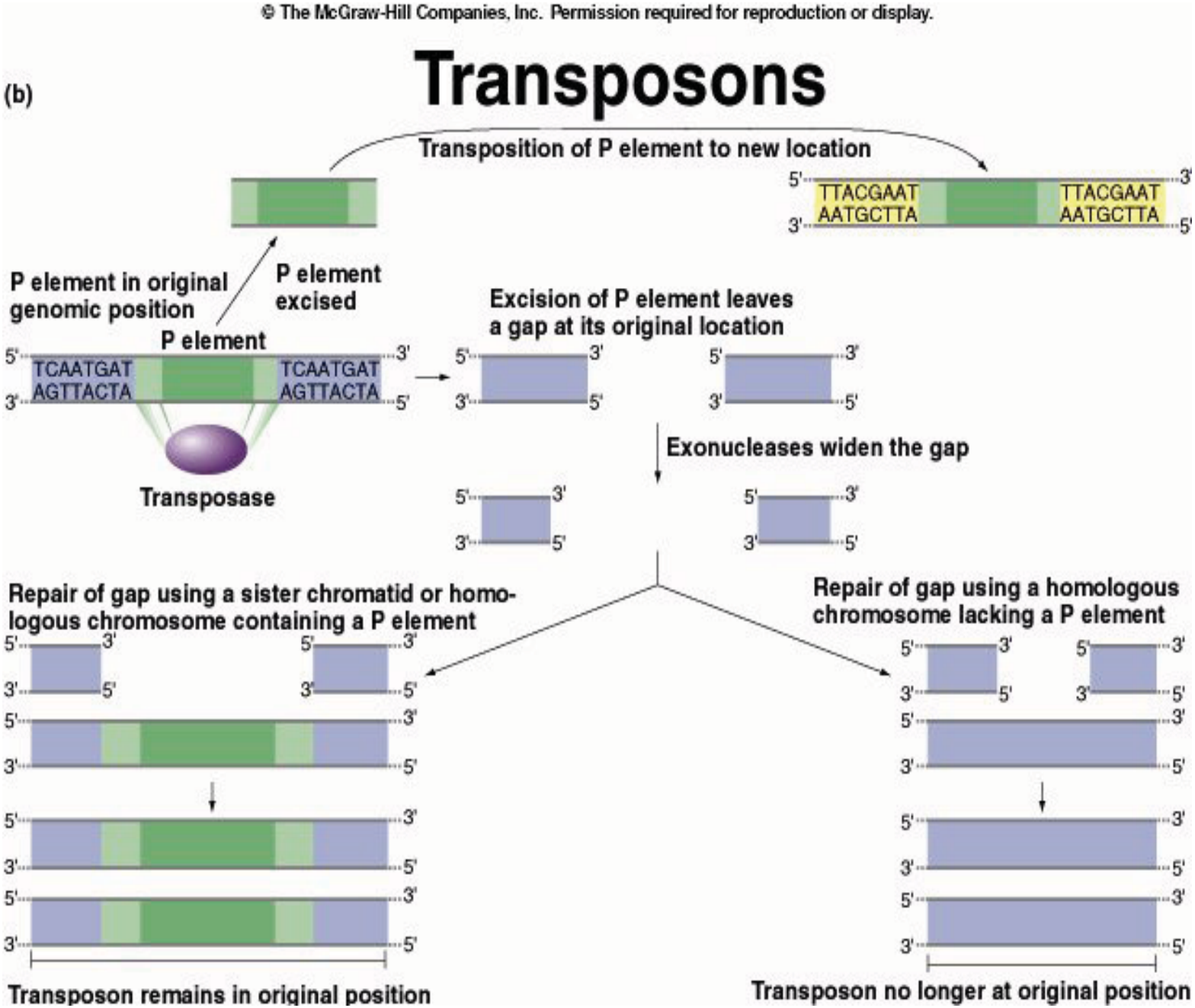
Transposons

- Transposase excises the sequence between the inverted repeats and inserts it into another region of the genome

- The gap created is widened by exonucleases.

- The gap is filled in by repair enzymes that use the sister chromatid or homologous chromosome as a template to fill the gap.
  - If copying from a sister chromatid (also containing the transposon) it will reappear in the original location
    - And hence be copied in the genome

  - If the homologous chromosome is used to replace missing transposon (did not have transposon) it will not be replaced
    - And hence not be copied in the genome



Retroposons

- The DNA sequence in a retroposon codes for a reverse transcriptase,
  - which catalyzes the formation of DNA from an RNA template.
- They always copy DNA and cause a duplication
- Many retroposons also have other polypeptide coding sequences.
- Many retroposons have a poly A tail.
- Others have direct repeat sequences on either side,
  - these are generated because of the way the DNA sequence has been inserted.

Retroposons: The process of LTR transposition

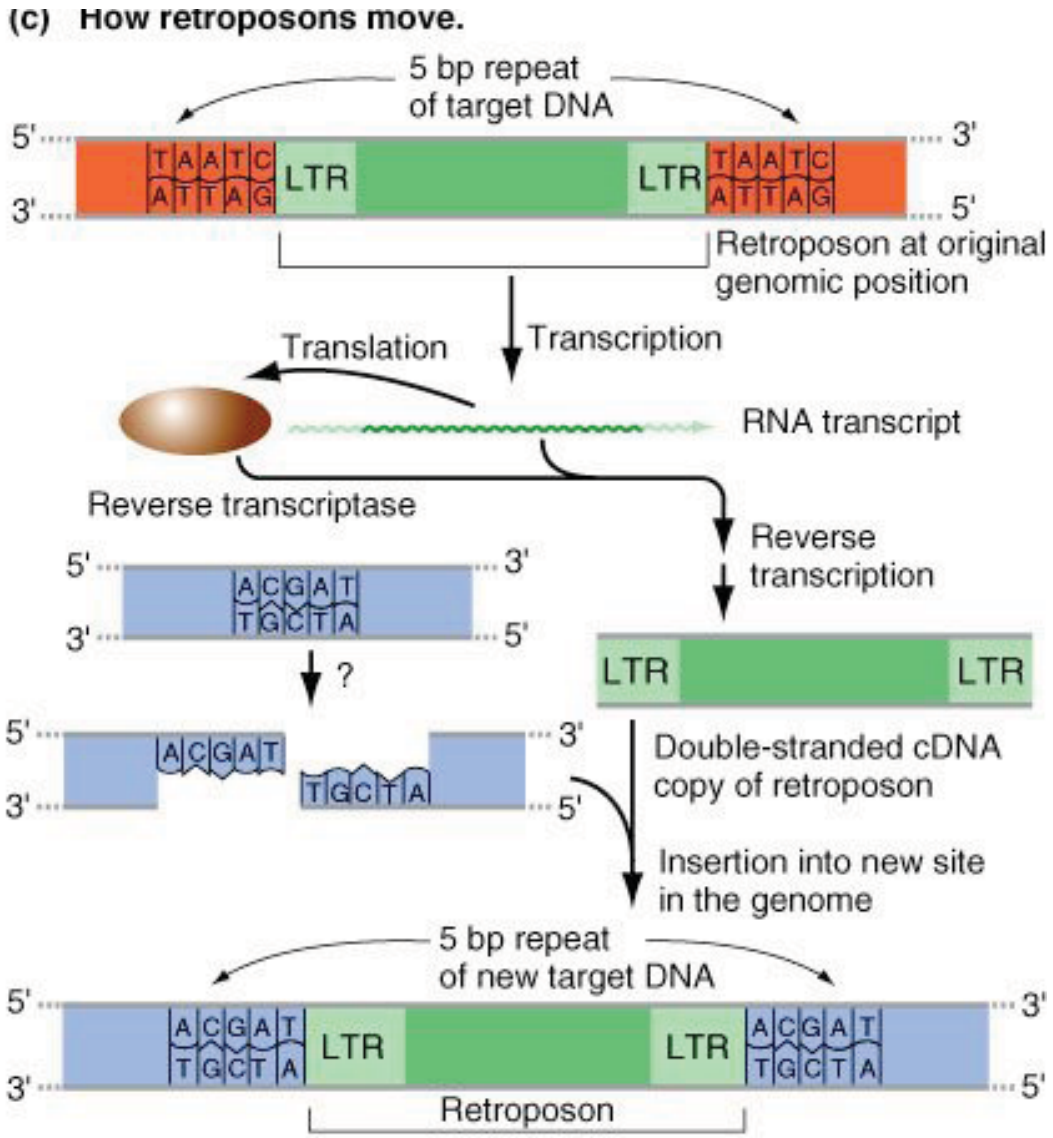
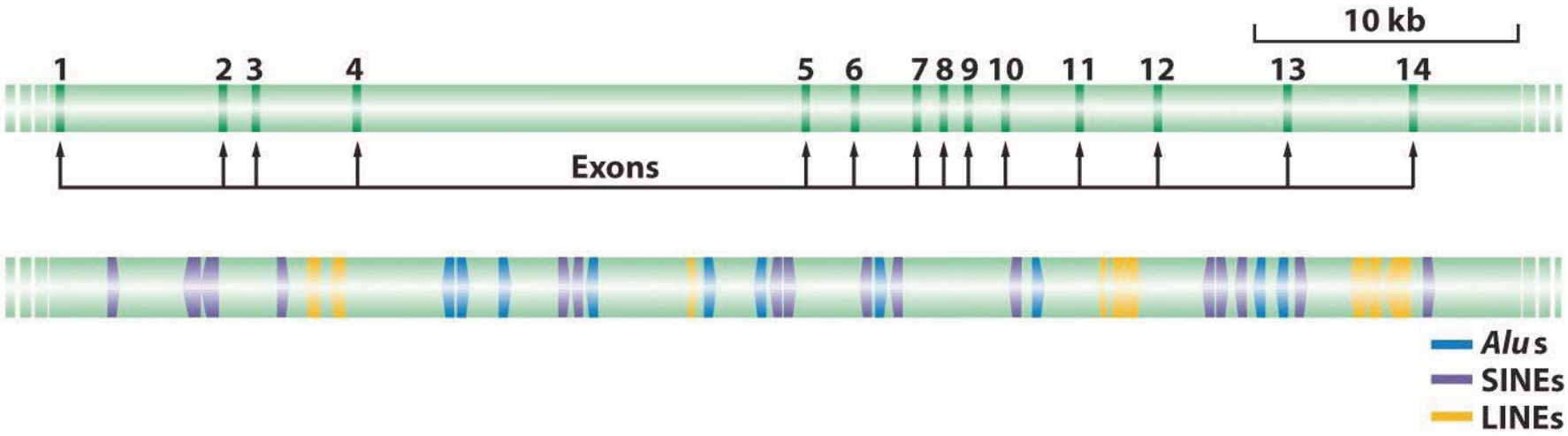


Fig. 13.23

Example of Transposable Elements Found in the Human Chromosome



Types of transposable elements in the human genome

Element	Transposition	Structure	Length	Copy number	Fraction of genome
LINEs	Autonomous	ORF1 ORF2 (*pol*) AAA	1–5 kb	20,000–40,000	21%
SINEs	Nonautonomous	AAA	100–300 bp	1,500,000	13%
DNA transposons	Autonomous	← transposase →	2–3 kb	300,000	3%
	Nonautonomous	← →	80–3000 bp		

Transposable elements move around the genome and are not susceptible to excision or mismatch repair

Why?

They are not damaged bases and they are not mismatches

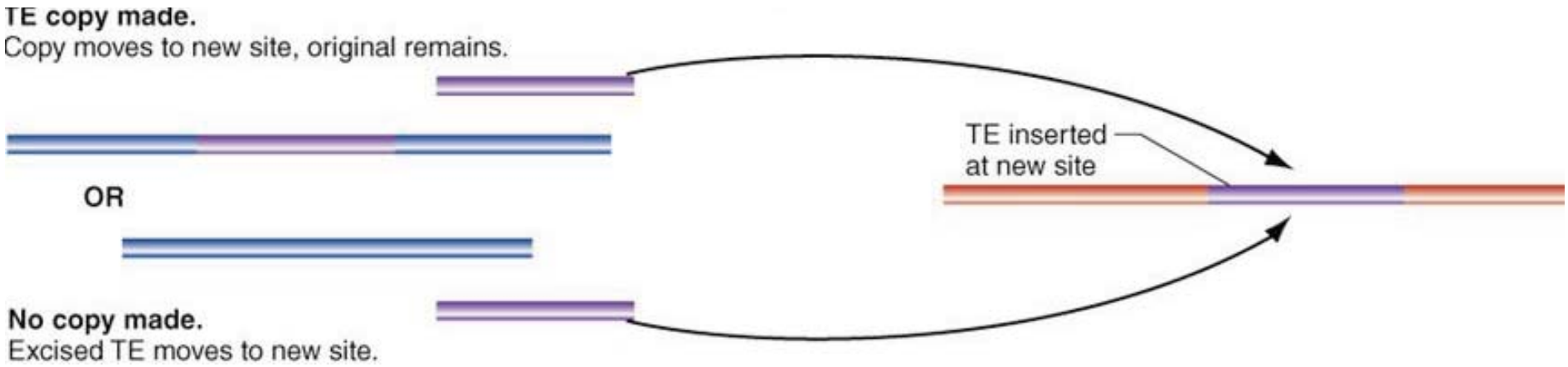


Fig. 7.10 e