MUTATIONS

&

Biology Section 8.7

Revised 2011

KEY CONCEPT

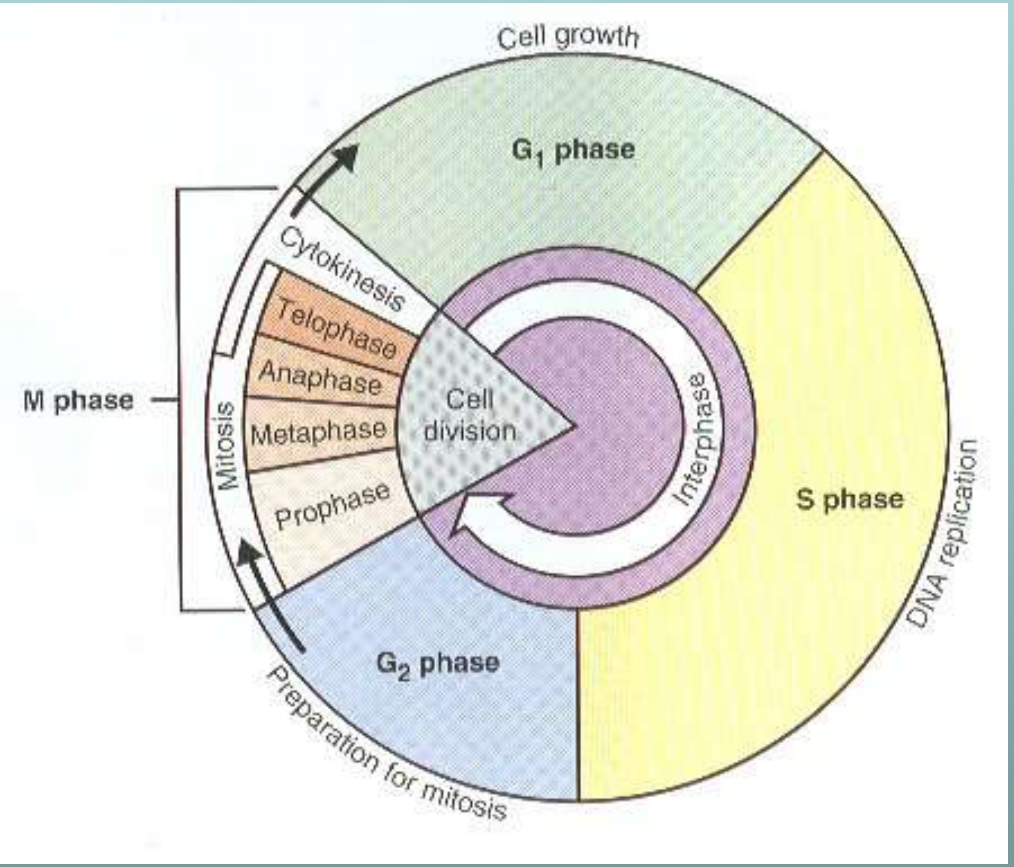
Mutations are changes in DNA that may or may not affect phenotype (phenotype = the physical expression of a gene.)

Two Types of Mutations

Some mutations affect a single gene, while others affect an entire chromosome.

Many kinds of gene mutations can occur, especially during DNA replication.

Chromosomal mutations are more likely to occur during Meiosis



Mutations can be caused by several factors.

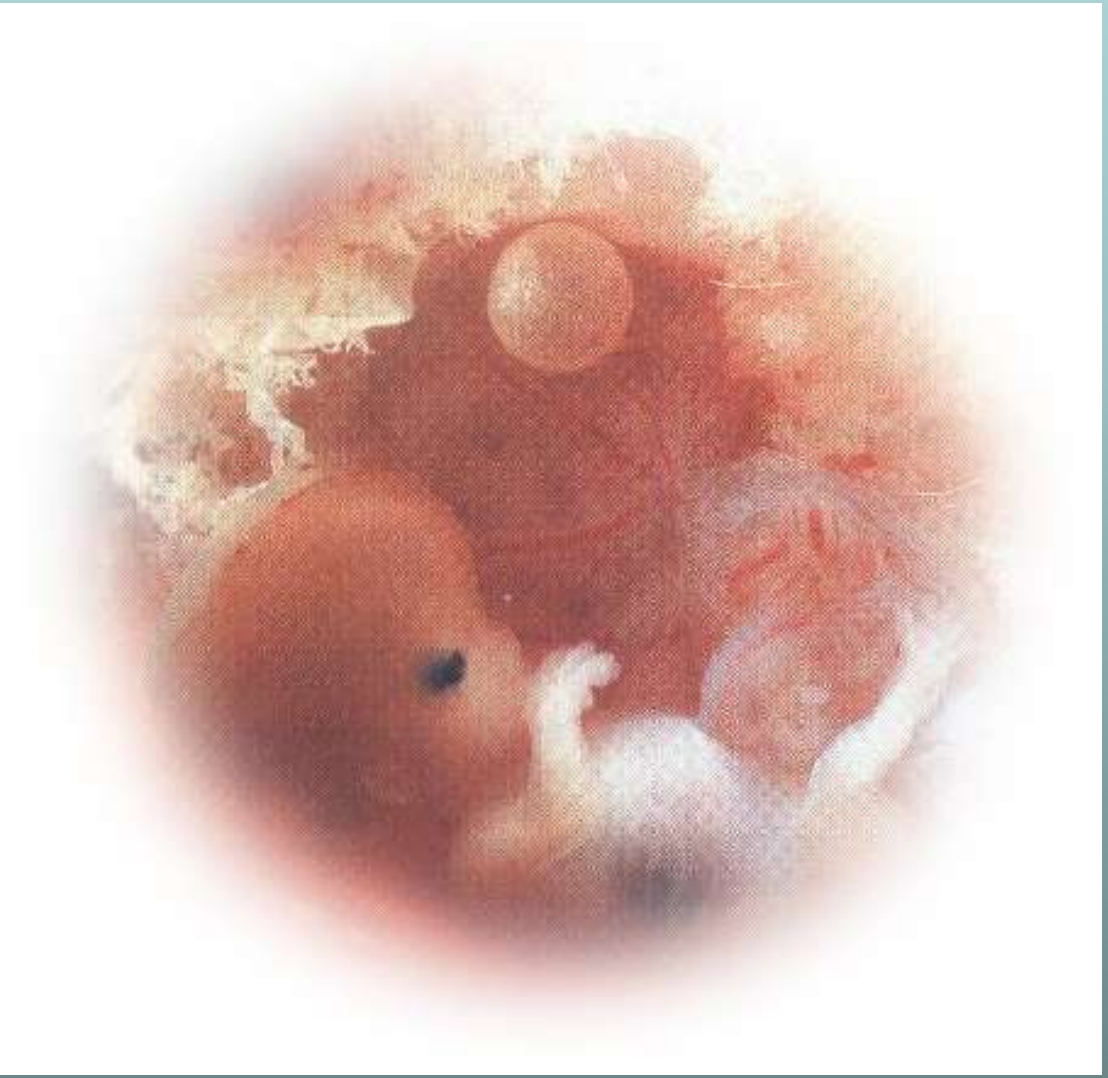
- 1. Replication errors can cause mutations.
- 2. Mutagens, such as UV rays and chemicals, can cause mutations.
- 3. Some cancer drugs use mutagenic properties to kill cancer cells.

Rachel Carson, ecologist who warned about use of pesticides causing mutatio in song birds.

Mutations affect the code for Proteins

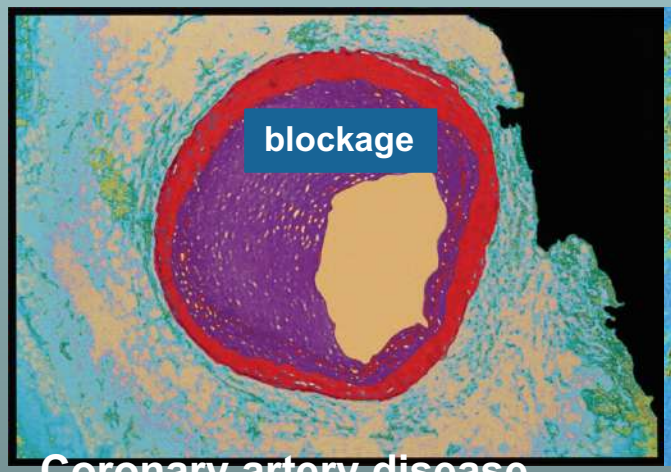
Proteins are key to everything cells do.

- **There are both Functional** (enzymes) and **structural proteins**

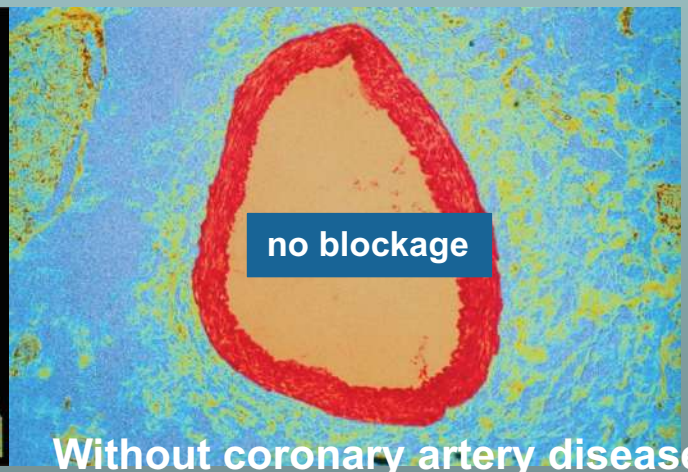


End Result: Mutations may or may not affect the organism.

- **Some gene mutations** change phenotype. (Strucure or function)
- Examples of changes from a gene mutation:
  - A mutation may cause a premature stop codon.
  - A mutation may change protein shape or the active site.
  - A mutation may change gene regulation.



Coronary artery disease



Without coronary artery disease

A gene mutation can help prevent cornornary artery disease like the picture on the lright.

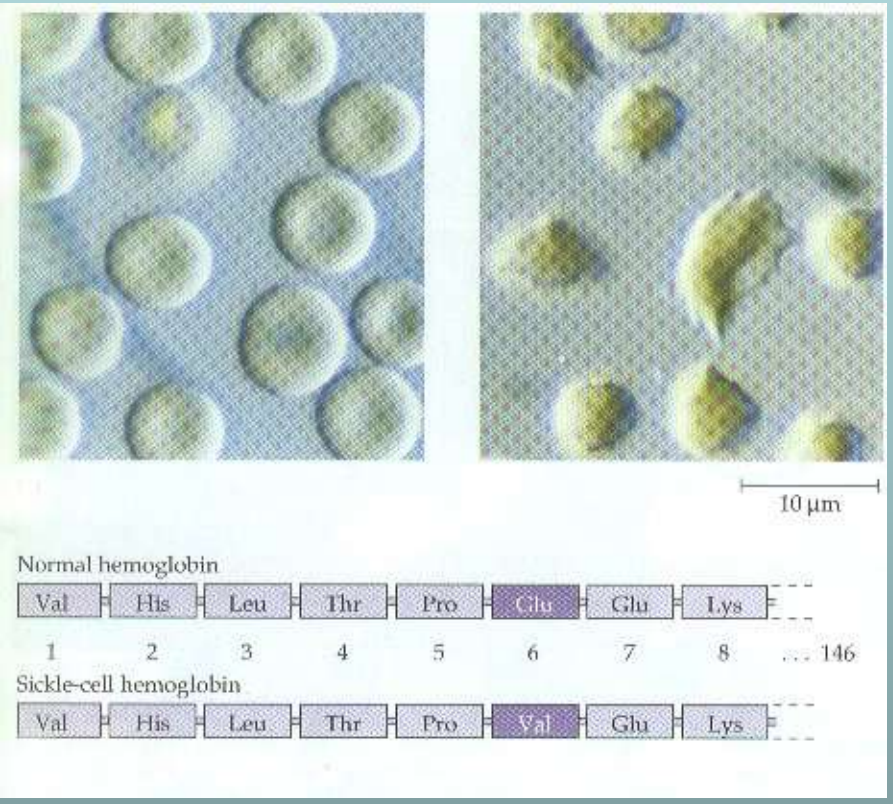
Cystic Fibrosis is caused
a gene deletion (frameshi

- Mutations in **body cells** do not affect offspring.

- Mutations in **sex cells** can be harmful or beneficial to offspring.

Gene Mutations- result from changes in a single gene

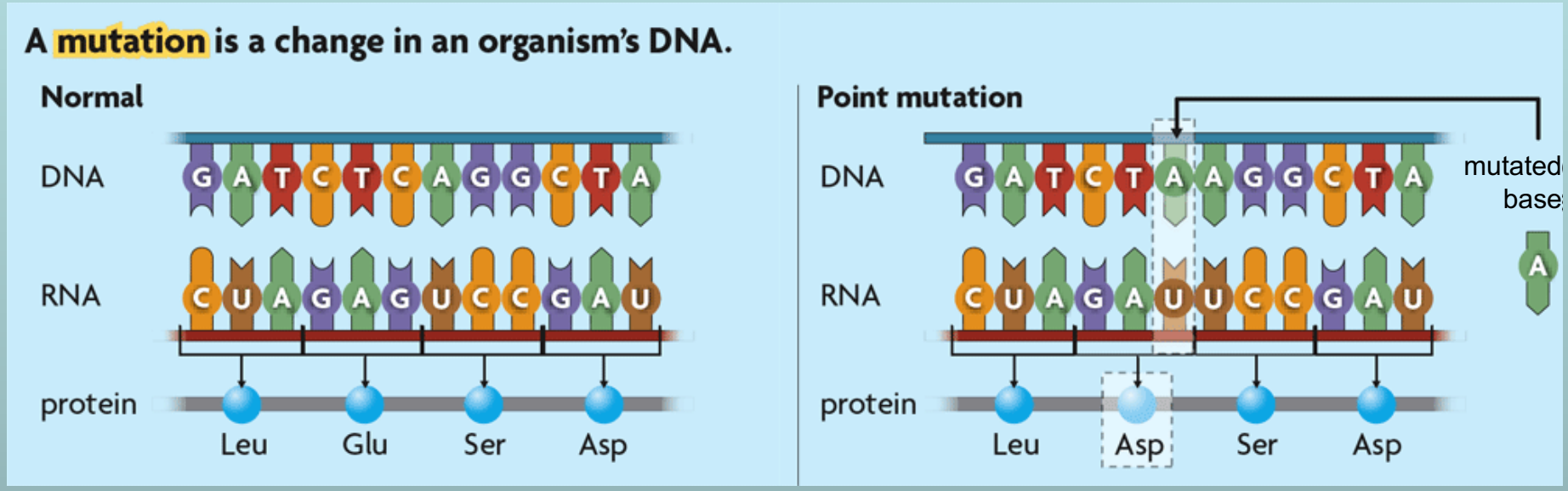
1. **Several types-** some involve several nucleotides, but most affect only one



a. **Point mutation**- occurs at a single point in DNA sequence. Generally change in one of the amino acids making up the protein

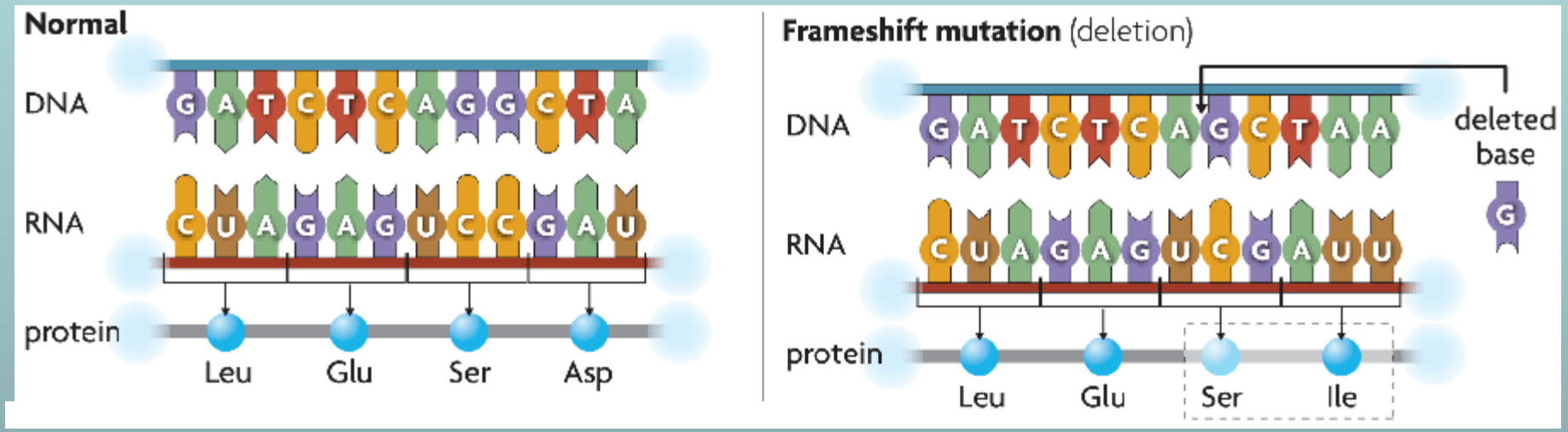
b. Ex. Sickle Cell Anemia – caused by a point mutation

- A **point mutation substitutes** one nucleotide for another.

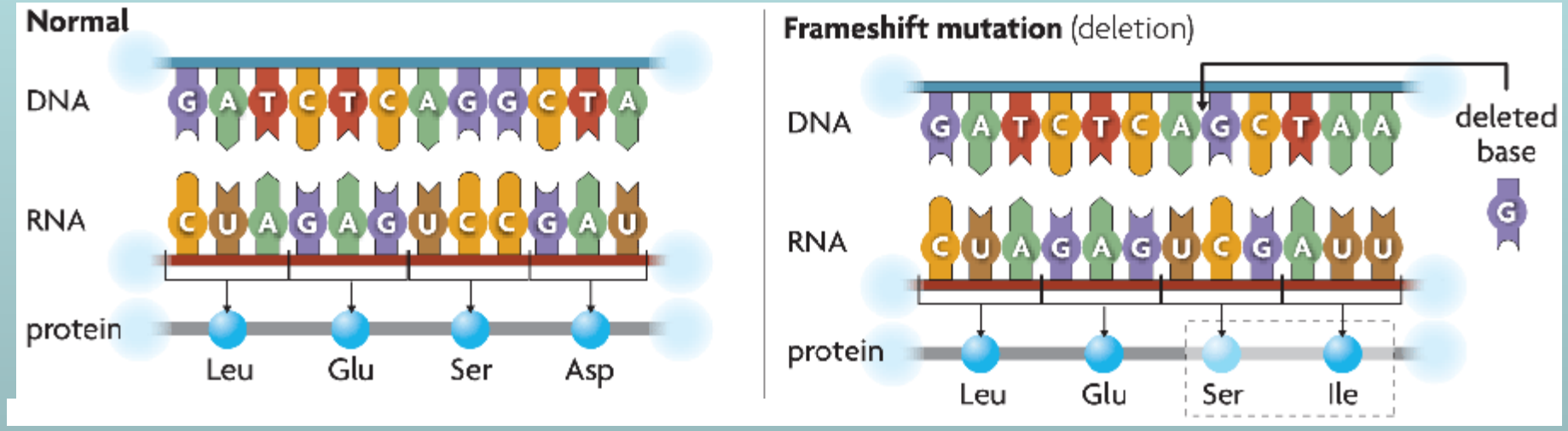


Frameshift mutation- insertion or deletion of nucleotide. **Causes bigger changes!**

Can alter protein- making it unable to perform normal functions

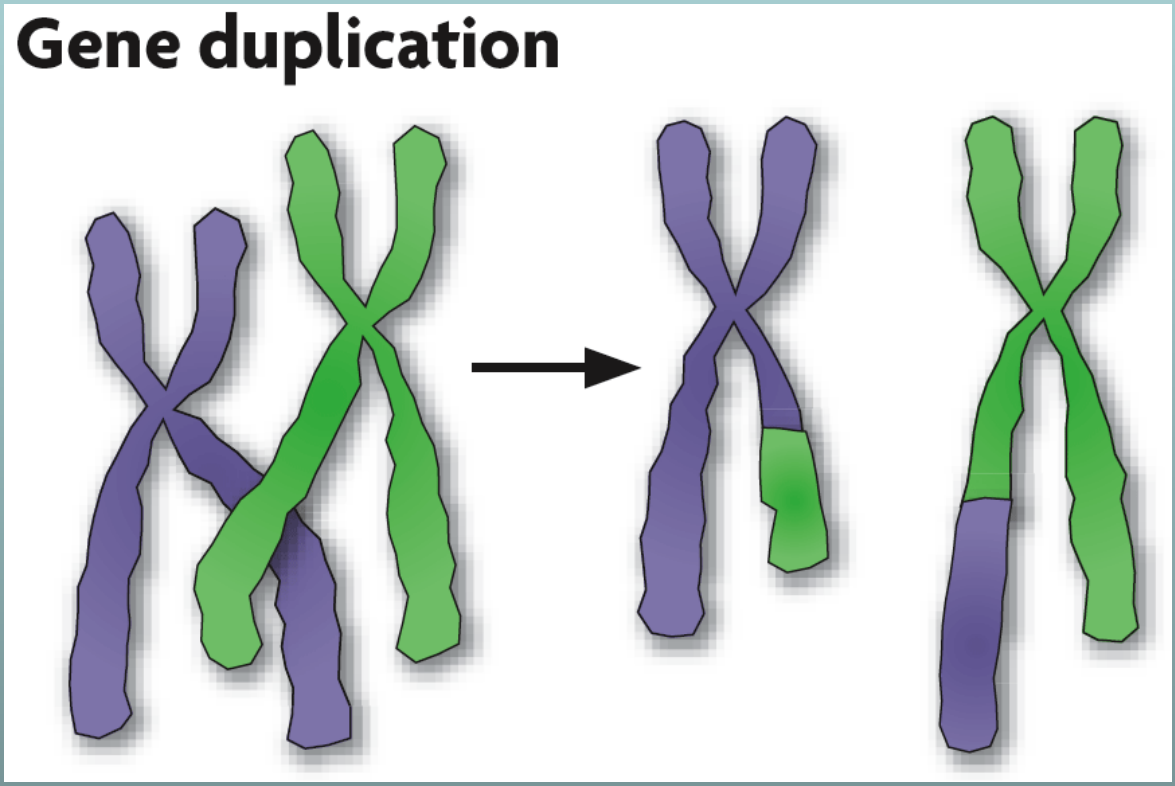


– A **frameshift mutation inserts or deletes** a nucleotide in the DNA sequence.

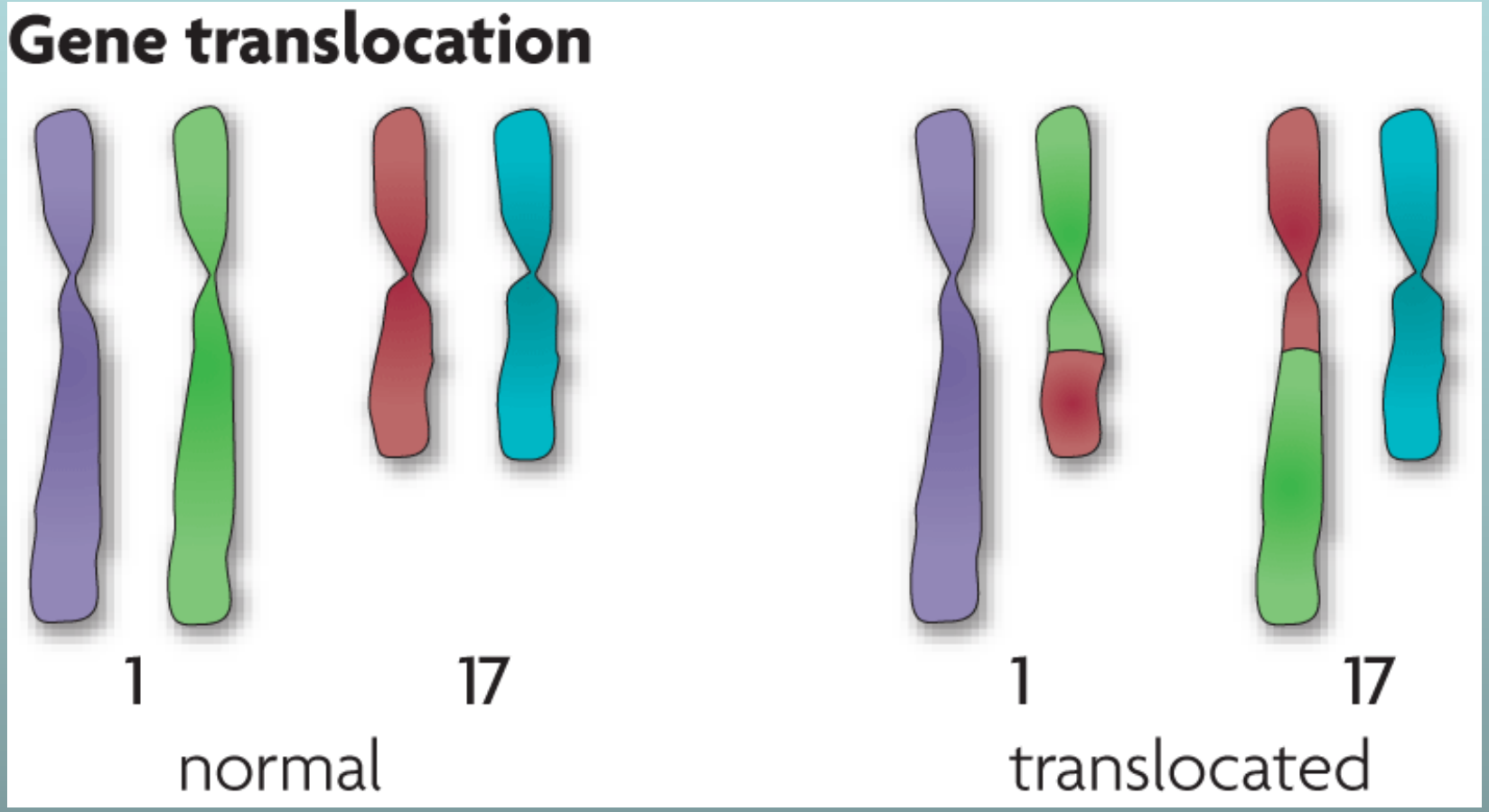


Chromosomal mutations tend to have a **big** effect.

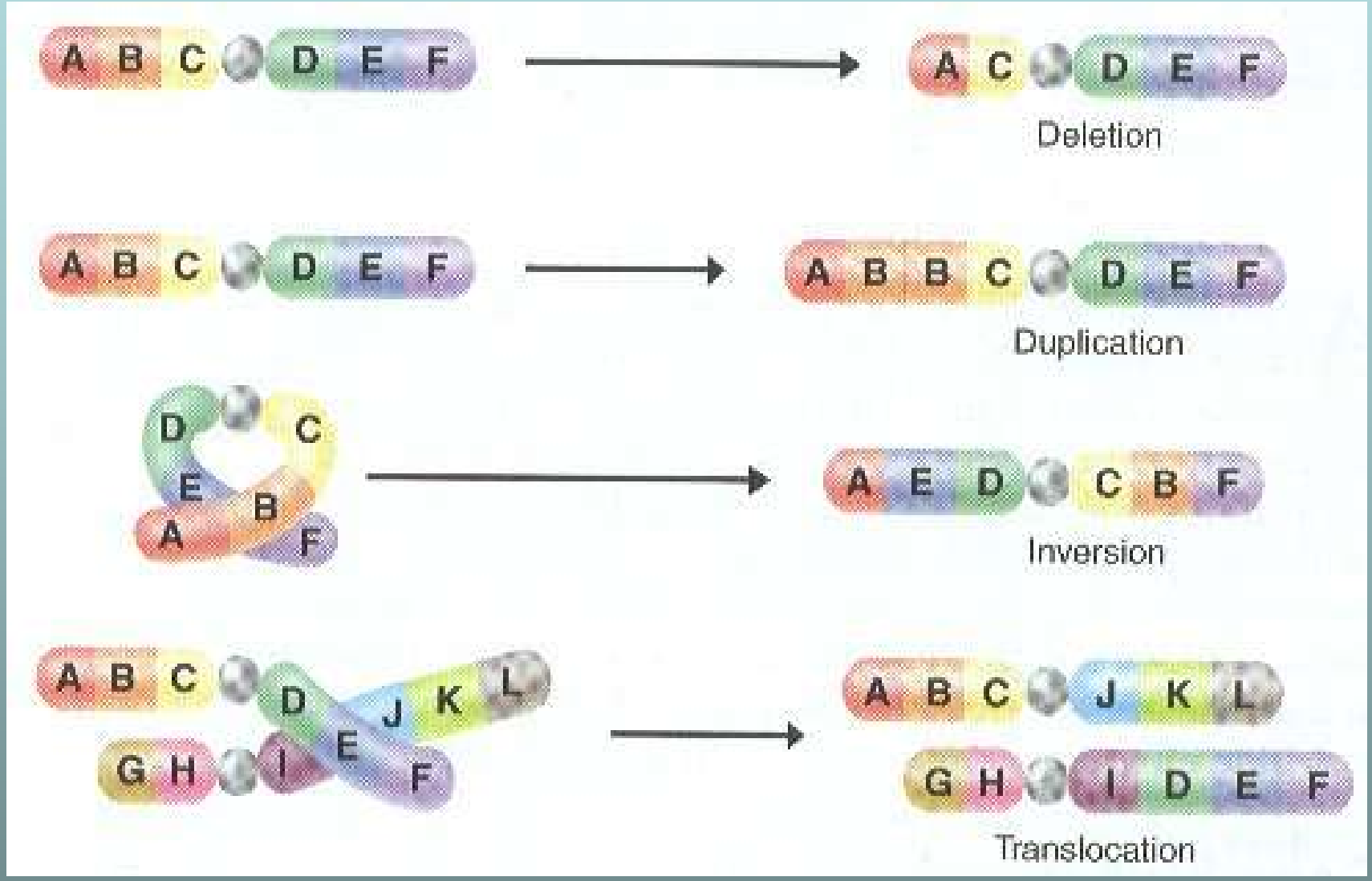
- **Chromosomal mutations** affect **many genes**.
- Chromosomal mutations **may occur** during **crossing over**
  - Chromosomal mutations affect many genes.
  - **Gene duplication** results from unequal crossing over.



- **Translocation** results from the exchange of DNA segments between nonhomologous chromosomes.



B. Chromosomal Mutations- involves changes in the number and structure of chromosomes



- Some gene mutations do not affect phenotype.
  - A mutation may be **silent =** does not affect the resulting protein. (Some substitutions can still result in the same amino acid being put in the protein. WHY?
  - A mutation may occur in a noncoding region.
  - A mutation may not affect protein folding or the active site.