

## Chap. 19 Gene Mutation & DNA Repair

### Learning Outcomes:

**Mutation:** a heritable change in the genetic material; the structure of the DNA is permanently changed and that change is passed on to daughter cells in cell division and from parent to offspring in reproductive cells. (not about the outcome).

- We have previously discussed changes in chromosome numbers and structure (chromosome mutations and genome mutations). These are considered mutations because they are heritable.
- Now we are considering mutations in single genes and the consequences of these mutations.

Gene mutations are molecular changes in the DNA sequence of a gene.

- **Point mutation:** a change in a single base pair within DNA.
  - **Base substitution:** a point mutation in which one base is substituted for another.
    - **Transition:** A base substitution of a pyrimidine for another **pyrimidine** (C to T) or a purine for another **purine** (A to G).
      - This is more common.
    - **Transversion:** A base substitution of a **purine** for a **pyrimidine**.
- Another type of mutation is when short sequences of base pairs are deleted or added to the chromosomal DNA.

Mutations in the coding sequence of a gene may alter a polypeptide function.

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TABLE 16.1 Consequences of Point Mutations Within the Coding Sequence				
Type of Change	Mutation in the DNA	Example*	Amino Acids Altered	Likely Effect on Protein Function
None	None	5'-A-T-G-A-C-C-G-A-C-C-C-G-A-A-A-G-G-G-A-C-C-T' Met - Thr - Asp - Pro - Cys - Gly - Thr -	None	None
Silent	Base substitution	5'-A-T-G-A-C-C-G-A-C-C-C-G-A-A-A-G-G-G-A-C-C-T' Met - Thr - Asp - Pro - Cys - Gly - Thr -	None	None
Missense	Base substitution	5'-A-T-G-A-C-C-G-A-C-C-C-G-A-A-A-G-G-G-A-C-C-T' Met - <b>Pro</b> - Asp - Pro - Cys - Gly - Thr -	One	Neutral or inhibitory
Nonsense	Base substitution	5'-A-T-G-A-C-C-G-A-C-C-C-G-T-A-A-G-G-G-A-C-C-T' Met - Thr - Asp - Pro - <b>STOP</b>	Many	Inhibitory
Frameshift	Addition/deletion	5'-A-T-G-A-C-C-G-A-C-C-C-C-C-G-A-A-G-G-G-A-C-C-T' Met - Thr - Asp - <b>Ala - Glu - Arg - Asp -</b>	Many	Inhibitory

\*DNA sequence is the coding strand. Note that this sequence is the same as the mRNA sequence except that the DNA contains uracil (U) instead of thymine (T). The 3-base codons are shown in alternating black and red colors. Mutations are shown in green.

# Mutation Study Guide

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