

## 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 - Gln - 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 - Gln - 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 - Gln - 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-G-A-A-A-G-G-G-A-C-C-T' Met - Thr - Asp - <b>Asn - 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

**Irwin Herman Herskowitz**



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## **Table of Contents Mutation Study Guide**

1. Understanding the eBook Mutation Study Guide
  - The Rise of Digital Reading Mutation Study Guide
  - Advantages of eBooks Over Traditional Books
2. Identifying Mutation Study Guide
  - Exploring Different Genres
  - Considering Fiction vs. Non-Fiction
  - Determining Your Reading Goals
3. Choosing the Right eBook Platform
  - Popular eBook Platforms
  - Features to Look for in an Mutation Study Guide
  - User-Friendly Interface
4. Exploring eBook Recommendations from Mutation Study Guide

- Personalized Recommendations
- Mutation Study Guide User Reviews and Ratings
- Mutation Study Guide and Bestseller Lists
- 5. Accessing Mutation Study Guide Free and Paid eBooks
  - Mutation Study Guide Public Domain eBooks
  - Mutation Study Guide eBook Subscription Services
  - Mutation Study Guide Budget-Friendly Options
- 6. Navigating Mutation Study Guide eBook Formats
  - ePub, PDF, MOBI, and More
  - Mutation Study Guide Compatibility with Devices
  - Mutation Study Guide Enhanced eBook Features
- 7. Enhancing Your Reading Experience
  - Adjustable Fonts and Text Sizes of Mutation Study Guide
  - Highlighting and Note-Taking Mutation Study Guide
  - Interactive Elements Mutation Study Guide
- 8. Staying Engaged with Mutation Study Guide
  - Joining Online Reading Communities
  - Participating in Virtual Book Clubs
  - Following Authors and Publishers Mutation Study Guide
- 9. Balancing eBooks and Physical Books Mutation Study Guide
  - Benefits of a Digital Library
  - Creating a Diverse Reading Collection Mutation Study Guide
- 10. Overcoming Reading Challenges
  - Dealing with Digital Eye Strain
  - Minimizing Distractions
  - Managing Screen Time
- 11. Cultivating a Reading Routine Mutation Study Guide
  - Setting Reading Goals Mutation Study Guide
  - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Mutation Study Guide

- Fact-Checking eBook Content of Mutation Study Guide
- Distinguishing Credible Sources
- 13. Promoting Lifelong Learning
  - Utilizing eBooks for Skill Development
  - Exploring Educational eBooks
- 14. Embracing eBook Trends
  - Integration of Multimedia Elements
  - Interactive and Gamified eBooks

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