

**Module 5: Mutations Study Guide**

1. A mutation is a permanent and heritable change in the **DNA sequence**. It can be neutral, beneficial (rare), or harmful.
2. Mutation that changes 1 DNA base is called a **point mutation**. (base substitution)
3. If change in a DNA base did not change the resulting amino acid, it's called a **silent mutation**; if the change in a DNA base change one resulting amino acid, it's called a **missense mutation**; if the change in a DNA base results in creation of a Stop codon, it's called a **nonsense mutation**.
4. Inversion of DNA sequences can result in **missense mutation** or **nonsense mutation**.
5. Frameshift mutation is caused by **deletion** or **insertion** of nucleotides not in multiple of 3. All amino acids after the mutation change. This mutation almost always results in **truncated/shortened** proteins with decreased or no function.
6. (This will be on the exam) You need to know how to compare the original DNA sequence and the mutated sequence, transcribe to RNA then translate to amino acid sequence, and determine what type of mutation it is. (see **module 6 homework questions** for example)
  - a. **Silent mutations**
    - i. First, you must convert the DNA sequence into the mRNA sequence.
    - ii. If the coding sequence is provided, the simplest way to transcribe is to take the coding sequence directly because codons are read from 5' to 3'.
    - iii. Then duplicate the GAG sequence to get your mRNA sequence, which is also from 5' to 3' but is now being converted to GAA.
    - iv. GAG, which codes for glutamic acids, is the **original sequence** on the fourth codon (Glu)
    - v. It has been renamed GAA, which also stands for glutamic acids (Glu)
    - vi. Because the amino acids are the same, the mutation had no effect on the resulting amino acid.
    - vii. A silent mutation occurs when there are no changes in the resulting amino acid.
  - b. **Missense mutations**
    - i. First, determine the mRNA sequence.
      1. AGG has been changed to AAG in the sequence copied from the coding strand.
      2. AGG stands for arginine (Arg)

# Mutation Study Guide

**Peter A. Rosenbaum**

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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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