

### 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.
    - ii. AGG stands for arginine (Arg)

# Mutation Study Guide

**Peter A. Rosenbaum**



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