

# General Biology II (101-HTK) Mutations

## Concepts and Learning Outcomes

Topic	Concept	Learning Outcomes
<b>What are mutations?</b>	1. Mutations are heritable changes in DNA (alterations of the DNA nucleotide sequence)—changes that are passed on to daughter cells.	1. Define mutation
<b>Mutations in multicellular organisms</b>	2. Multicellular organisms have two types of mutations: somatic (passed on during mitosis to daughter cells, but not to subsequent generations) and germ line (occur in cells that give rise to gametes) mutations.	2. Name and contrast the 2 types of mutations that may occur in multicellular organisms
<b>Phenotypic effects of mutations</b>	3. Phenotypically, mutations may affect proteins and their functions: (1) silent mutation (no effect on protein structure and function), (2) loss of function mutation (mutation coding for non-functional protein), (3) gain of function mutation (mutation coding for a protein with new function), and (4) conditional mutation (phenotype expressed under certain restrictive conditions).	3. Identify and compare the 4 different phenotypic classes of mutations, and explain the effects that each has on mRNA and resulting amino acid sequence
<b>Types of mutations</b>	4. At the molecular level, mutations can be classified as being (1) point mutations (affecting single genes) or chromosomal mutations (changes in the arrangements of chromosomal DNA segments). 5. Point mutations are changes in single nucleotides and can be of 4 different types: (1) silent mutation (no change in amino acid sequence specified by codons), (2) missense (change in amino acid sequence specified by codons), (3) nonsense mutation (shortened amino acid sequence due to substitution causing	4. Identify the 2 types of mutations, and explain the molecular effects that each has on base sequences of DNA 5. Identify and give examples of the 4 different classes of point mutations, and explain the molecular effects that each has on base sequences of DNA 6. Identify and give examples of the 4 different classes of chromosomal mutations, and explain the effects that each has on DNA segments

a stop codon to form somewhere in the mRNA), and (4) frame-shift mutation (addition/deletion of nucleotide resulting in codon frame-shift).

6. Chromosomal mutations involve large regions of chromosomes and can be divided into 4 categories: (1) deletion (loss of chromosomal segment), (2) duplication (deletion of segment from a chromosome and its reattachment to its homologous chromosome), inversion (reinsertion of a broken chromosomal segment in reverse order), and (4) translocation (exchange of chromosomal segments between non-homologous chromosomes).

**Causes of mutations**

7. Mutations can be the result of spontaneous (endogenous permanent changes caused by tautomeric shifts, chemical reactions, errors in DNA replication, etc.) or induced changes in DNA (exogenous permanent changes caused by mutagens such as chemicals and radiation).

7. Compare spontaneous and induced mutations and give examples of each

**Overall effect of mutations**

8. Mutations can be beneficial or harmful. Germ line mutations provide genetic diversity and are therefore the raw material of evolution, while some somatic mutations can cause cancer.

8. Explain the overall effects of mutations and give examples of beneficial and harmful effects of mutations