Topic 5 – Mutations and Genetic Variation
Pre-Class Reading Assignment

1. Read pgs 687-690

2. Define the following terms

a. Point mutations
b. Gene mutations
c. Silent mutation
d. Missense mutation
e. Nonsense mutation
f. Deletion
g. Insertion
h. Frameshift mutation
i. Translocation
j. Inversion
k. Spontaneous mutation
l. Mutagenic agent
m. Induced mutation

3. Explain how mutations may be of benefit to an organism, and describe how these beneficial mutations are maintained in a species. Identify the biological process that influences which mutations stay in a population over time. (Read pg 690)

Topic 5 – Mutations and Genetic Variation

- A mutation occurs when the sequence of bases in a DNA molecule is altered

- Mutations can occur in any cell in the body
  - Mutations of somatic cells are not passed on to offspring
  - Mutations of germ cells can be passed on to offspring

- Mutations can be beneficial or harmful
  - Beneficial mutations that occur in germ cells are passed on
  - Harmful mutations reduce the ability of an organism to survive and usually are not passed on

Point mutations –
- A mutations which alters only a single gene

- Can occur when a nucleotide is changed, deleted (deletion) or added (insertion)

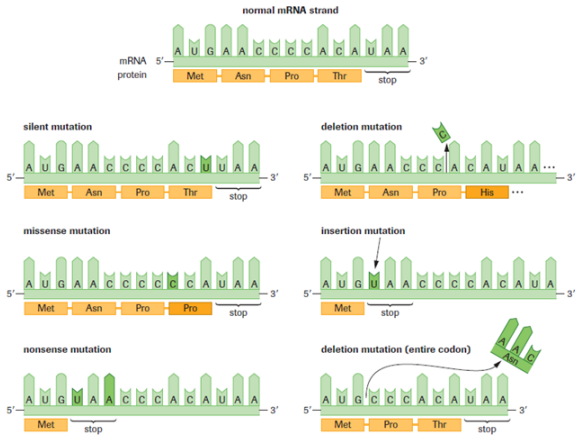
- There are many kinds of point mutations
  - **Silent** mutation – does not result in a change to the amino acid being coded for

  - **Missense** mutation – results in the single substitution of one amino acid in the polypeptide

  - **Nonsense** mutation – converts a codon for an amino acid into a stop codon

  - **Insertion** mutation – placement of an extra nucleotide in a DNA sequence

  - **Deletion** mutation – removal of a nucleotide from the DNA sequence

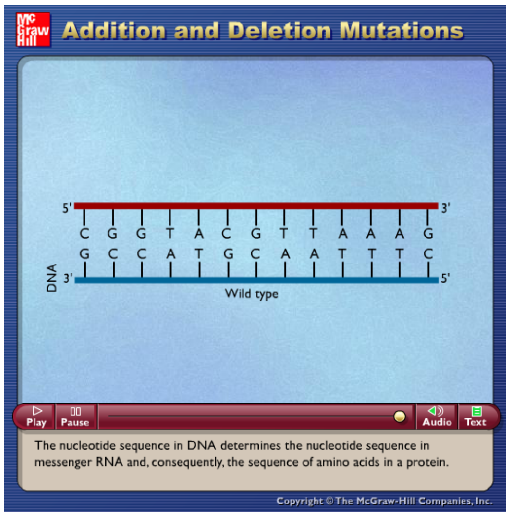


- Frameshift mutation

  - DNA is read in frames of three

  - A frame shift mutation occurs when one or more base pairs is added or deleted from the DNA sequence

  - As a result the reading of the frames is affected

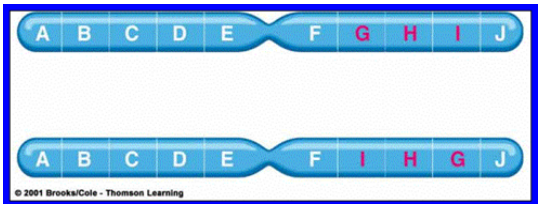


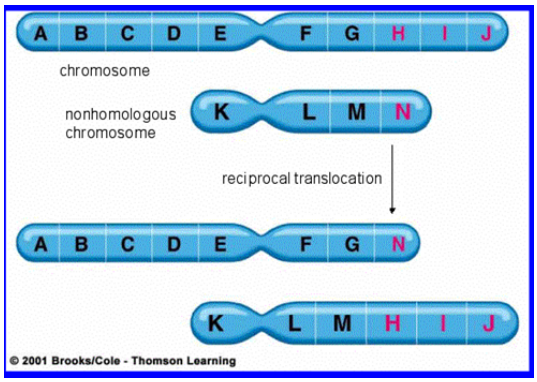
Chromosomal Mutations
- A mutation which affects large sections of DNA

- May be harmful depending on where the mutations occur

- There are two types:
    - Translocation
        - relocation of a section of DNA from one chromosome to another
        - Usually occurs between non-homologous chromosomes

    - Inversion
        - Section of chromosome has reversed its position in a chromosome





Causes of Mutations
- Spontaneous mutations
    - Occur as a result of DNA replication
    - Usually an enzyme checks the new DNA strands for errors in the replication process (but it can miss some)

- Induced mutations
    - Arise from exposure to mutagenic agents (something that causes a mutation)
    - Eg. UV radiation, X-rays, certain chemicals

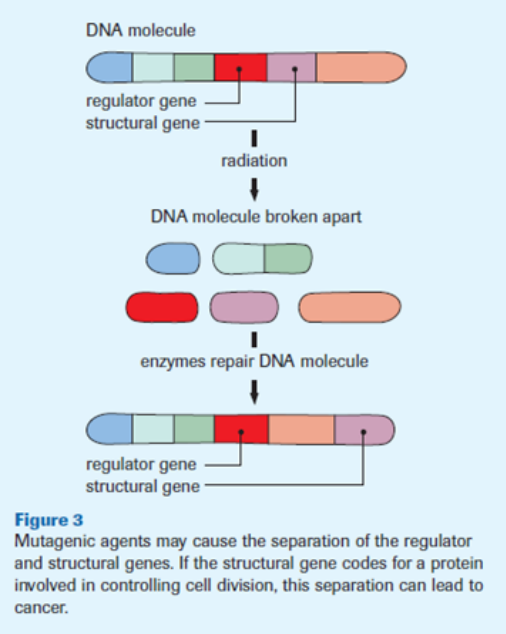


Figure 3
Mutagenic agents may cause the separation of the regulator and structural genes. If the structural gene codes for a protein involved in controlling cell division, this separation can lead to cancer.

Topic 5 – Mutations and Genetic Variation
Review Sheet

1. Clearly define the following terms and give an example of each using a strand of DNA: mutation, frameshift mutation, point mutation, nonsense mutation, missense mutation.

2. Explain why mutations, such as insertions or deletions, are often much more harmful than nitrogen-base substitutions

3. Identify the type of mutation that has occurred in the strands below. Describe the effect on the protein. The original strand is

AUG UUU UUG CCU UAU CAU CGU

Determine whether or not the following mutations would be harmful to an organism. Translate the mRNA sequence into protein to help you decide. The mutation is underlined.

(a) AUG UUU UUG CCU UAU CAU CGU
AUG UUU UUG CCU UAC CAU CGU

(b) AUG UUU UUG CCU UAU CAU CGU
AUG UUU UUG CCU UAA CAU CGU

(c) AUG UUU UUG CCU UAU CAU CGU
AUG UUU CUU GCC UUA UCA UCG U

(d) AUG UUU UUG CCU UAU CAU CGU
AUG UUU UUG CCU AUC AUC GU

(e) AUG UUU UUG CCU UAU CAU CGU
UGC UAC UAU UCC GUU UUU GUA

4. Which of the following amino acid changes can result from a single base-pair substitution?
(a) arg to leu (b) cys to glu (c) ser to thr (d) ile to ser

Insulin is a protein hormone. It has been hypothesized that a change in the 57th amino acid of this hormone from asparagine to another amino acid will result in an increased risk for developing IDDM.

i. Write a DNA triplet that codes for asparagine. **(1 mark)**

ii. Show how a single base change in this DNA triplet would code for an amino acid other than asparagine. Identify the amino acid coded for by the mutated DNA triplet. **(1 mark)**