

7.3 Mutation

Lesson Objectives

- Identify causes of mutation.
- Compare and contrast types of mutations.
- Explain how mutations may affect the organisms in which they occur.

Vocabulary

- chromosomal alteration
- frameshift mutation
- genetic disorder
- germline mutation
- mutagen
- mutation
- point mutation
- somatic mutation

Introduction

A change in the sequence of bases in DNA or RNA is called a **mutation**. Does the word mutation make you think of science fiction and bug-eyed monsters? Think again. Everyone has mutations. In fact, most people have dozens or even hundreds of mutations in their DNA. Mutations are essential for evolution to occur. They are the ultimate source of all new genetic material in a species. Although most mutations have no effect on the organisms in which they occur, some mutations are beneficial. Even harmful mutations rarely cause drastic changes in organisms.

Causes of Mutation

Mutations have many possible causes. Some mutations seem to happen spontaneously without any outside influence. They occur when mistakes are made during DNA replication or transcription. Other mutations are caused by environmental factors. Anything in the environment that can cause a mutation is known as a **mutagen**. Examples of mutagens are pictured in **Figure 7.12**. For a video about mutagens, go the link below. <http://www.youtube.com/watch?v=OwrNxCGKCws&feature=related> (0:36)

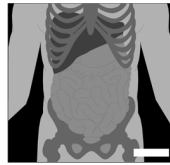
Types of Mutations

There are a variety of types of mutations. Two major categories of mutations are germline mutations and somatic mutations.

- **Germline mutations** occur in gametes. These mutations are especially significant because they can be transmitted to offspring and every cell in the offspring will have the mutation.
- **Somatic mutations** occur in other cells of the body. These mutations may have little effect on the organism because they are confined to just one cell and its daughter cells. Somatic mutations cannot be passed on to offspring.

Radiation

UV Radiation
both natural sunlight
and tanning beds



X-Rays
medical, dental,
airport security screening

Chemicals

Cigarette Smoke
contains dozens of
mutagenic chemicals



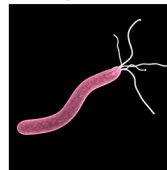
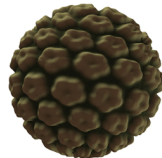
Benzoyl Peroxide
common ingredient
in acne products

**Nitrate and Nitrite
Preservatives**
in hot dogs and
other processed meats

Barbecuing
creates mutagenic
chemicals in foods

Infectious Agents

**Human Papillomavirus
(HPV)**
sexually transmitted virus



Helicobacter pylori
bacteria spread through
contaminated food

Figure 7.12: Examples of Mutagens. Types of mutagens include radiation, chemicals, and infectious agents. Do you know of other examples of each type of mutagen shown here?

Mutations also differ in the way that the genetic material is changed. Mutations may change the structure of a chromosome or just change a single nucleotide.

Chromosomal Alterations

Chromosomal alterations are mutations that change chromosome structure. They occur when a section of a chromosome breaks off and rejoins incorrectly or does not rejoin at all. Possible ways these mutations can occur are illustrated in **Figure 7.13**. Go to this link for a video about chromosomal alterations: http://www.youtube.com/watch?v=0rXRSqa_3lU&feature=related (2:18).

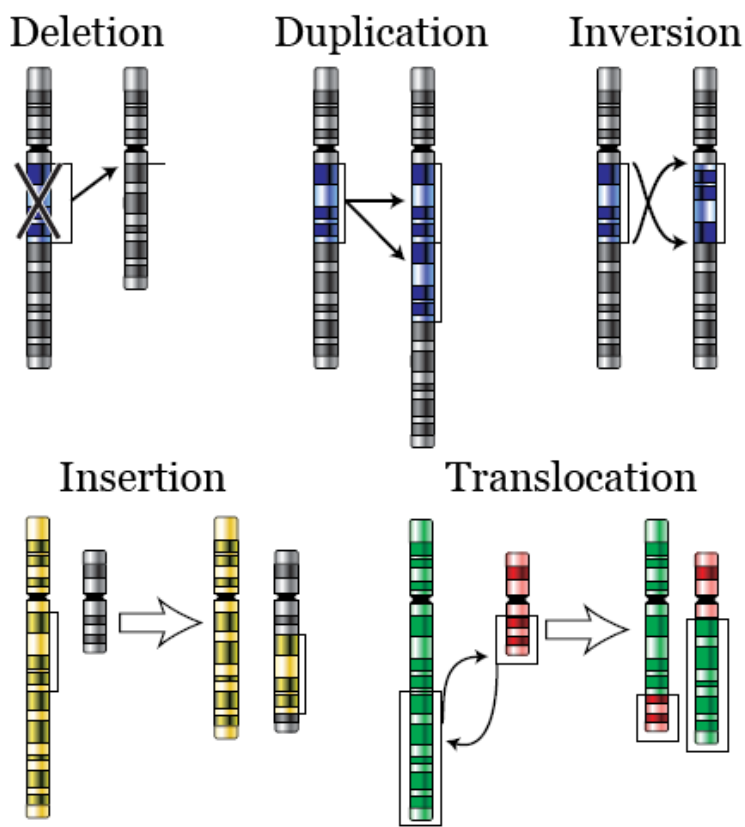


Figure 7.13: Chromosomal Alterations. Chromosomal alterations are major changes in the genetic material.

Chromosomal alterations are very serious. They often result in the death of the organism in which they occur. If the organism survives, it may be affected in multiple ways. An example of a human chromosomal alteration is the mutation that causes Down Syndrome. It is a duplication mutation that leads to developmental delays and other abnormalities.

Point Mutations

A **point mutation** is a change in a single nucleotide in DNA. This type of mutation is usually less serious than a chromosomal alteration. An example of a point mutation is a mutation that changes the codon UUU to the codon UCU. Point mutations can be silent, missense, or nonsense mutations, as shown in **Table 7.1**. The effects of point mutations depend on how they change the genetic code. You can watch an animation about nonsense mutations at this link: http://www.biostudio.com/d_%20Nonsense%20Suppression%20I%20Nonsense%20Mutation.htm.

Table 7.1: Point Mutations and Their Effects

Type	Description	Example	Effect
Silent	mutated codon codes for the same amino acid	CAA (glutamine) → CAG (glutamine)	none
Missense	mutated codon codes for a different amino acid	CAA (glutamine) → CCA (proline)	variable
Nonsense	mutated codon is a premature stop codon	CAA (glutamine) → UAA (stop) usually	serious

Frameshift Mutations

A **frameshift mutation** is a deletion or insertion of one or more nucleotides that changes the reading frame of the base sequence. Deletions remove nucleotides, and insertions add nucleotides. Consider the following sequence of bases in RNA:

AUG-AAU-ACG-GCU = start-asparagine-threonine-alanine

Now, assume an insertion occurs in this sequence. Let's say an **A** nucleotide is inserted after the start codon **AUG**:

AUG-AAA-UAC-GGC-U = start-lysine-tyrosine-glycine

Even though the rest of the sequence is unchanged, this insertion changes the reading frame and thus all of the codons that follow it. As this example shows, a frameshift mutation can dramatically change how the codons in mRNA are read. This can have a drastic effect on the protein product.

Effects of Mutations

The majority of mutations have neither negative nor positive effects on the organism in which they occur. These mutations are called neutral mutations. Examples include silent point mutations. They are neutral because they do not change the amino acids in the proteins they encode. Many other mutations have no effect on the organism because they are repaired before protein synthesis occurs. Cells have multiple repair mechanisms to fix mutations in DNA. One way DNA can be repaired is illustrated in **Figure 7.14**. If a cell's DNA is permanently damaged and cannot be repaired, the cell is likely to be prevented from dividing.

Beneficial Mutations

Some mutations have a positive effect on the organism in which they occur. They are called beneficial mutations. They lead to new versions of proteins that help organisms adapt to changes in their environment. Beneficial mutations are essential for evolution to occur. They increase an organism's chances of surviving or reproducing, so they are likely to become more common over time. There are several well-known examples of beneficial mutations. Here are just two:

1. Mutations in many bacteria that allow them to survive in the presence of antibiotic drugs. The mutations lead to antibiotic-resistant strains of bacteria.
2. A unique mutation is found in people in a small town in Italy. The mutation protects them from developing atherosclerosis, which is the dangerous buildup of fatty materials in blood vessels. The individual in which the mutation first appeared has even been identified.

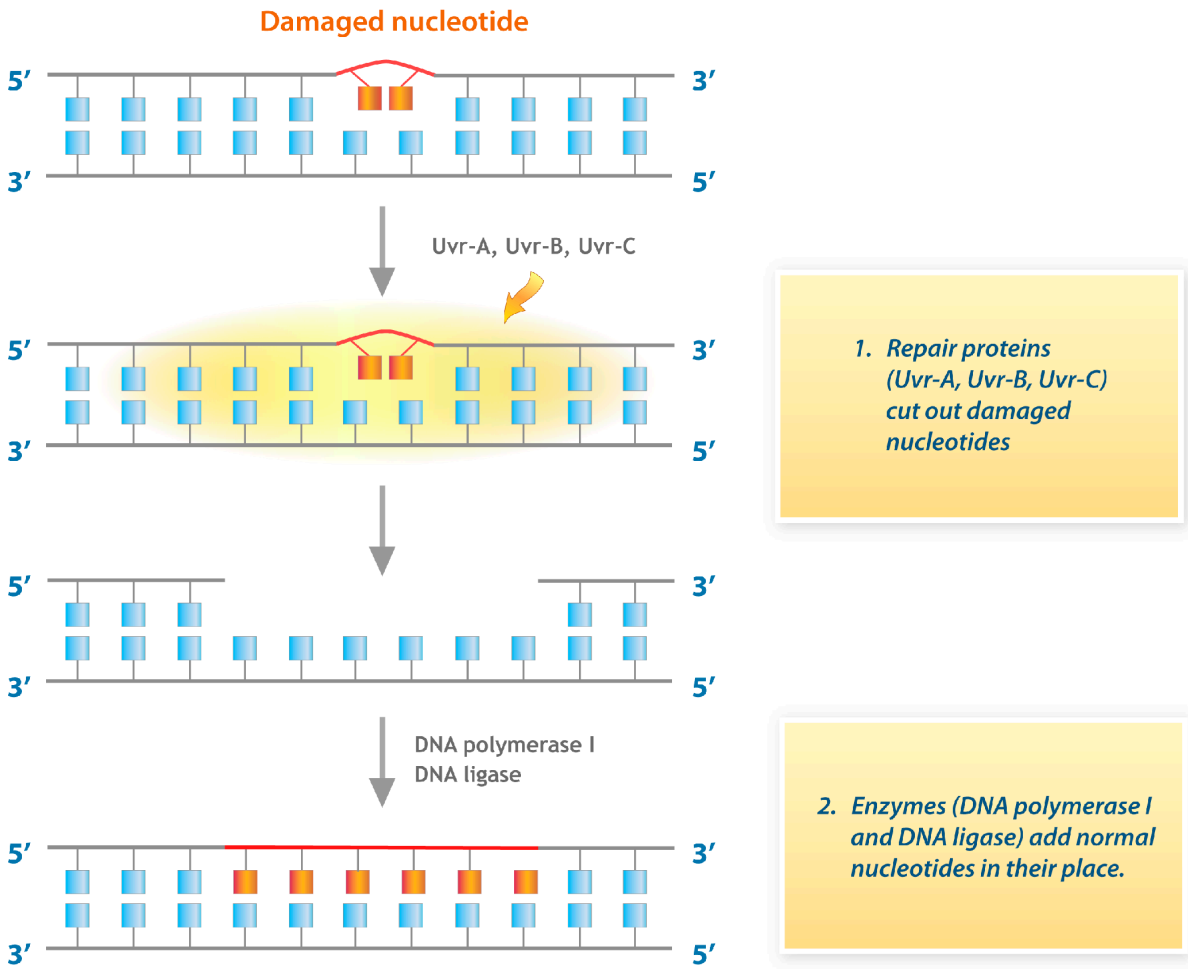


Figure 7.14: DNA Repair Pathway. This flow chart shows one way that damaged DNA is repaired in *E. coli* bacteria.

Harmful Mutations

Imagine making a random change in a complicated machine such as a car engine. The chance that the random change would improve the functioning of the car is very small. The change is far more likely to result in a car that does not run well or perhaps does not run at all. By the same token, any random change in a gene's DNA is likely to result in a protein that does not function normally or may not function at all. Such mutations are likely to be harmful. Harmful mutations may cause genetic disorders or cancer.

- A **genetic disorder** is a disease caused by a mutation in one or a few genes. A human example is cystic fibrosis. A mutation in a single gene causes the body to produce thick, sticky mucus that clogs the lungs and blocks ducts in digestive organs. You can watch a video about cystic fibrosis and other genetic disorders at this link: http://www.youtube.com/watch?v=8s4he3wLgkM#38;feature=Playlist#38;p=397710758E9BCB24#38;playnext_from=PL#38;playnext=1#38;index=17 (9:31).
- Cancer is a disease in which cells grow out of control and form abnormal masses of cells. It is generally caused by mutations in genes that regulate the cell cycle. Because of the mutations, cells with damaged DNA are allowed to divide without limits. Cancer genes can be inherited. You can learn more about hereditary cancer by watching the video at the following link: <http://www.youtube.com/watch?v=LWk5FplsKwM> (4:29)

Lesson Summary

- Mutations are caused by environmental factors known as mutagens. Types of mutagens include radiation, chemicals, and infectious agents.
- Germline mutations occur in gametes. Somatic mutations occur in other body cells. Chromosomal alterations are mutations that change chromosome structure. Point mutations change a single nucleotide. Frameshift mutations are additions or deletions of nucleotides that cause a shift in the reading frame.
- Mutations are essential for evolution to occur because they increase genetic variation and the potential for individuals to differ. The majority of mutations are neutral in their effects on the organisms in which they occur. Beneficial mutations may become more common through natural selection. Harmful mutations may cause genetic disorders or cancer.

Lesson Review Questions

Recall

1. Define mutation and mutagen.
2. List three examples of mutagens.
3. Identify three types of chromosomal alterations.
4. Distinguish among silent, missense, and nonsense point mutations.
5. What is a frameshift mutation? What causes this type of mutation?

Apply Concepts

6. Assume that a point mutation changes the codon AUU to AUC. Why is this a neutral mutation?

7. Look at the mutation shown below. The base A was inserted following the start codon AUG. Describe how this mutation affects the encoded amino acid sequence.

AUG-GUC-CCU-AAA → AUG-AGU-CCC-UAA-A

Think Critically

8. Compare and contrast germline mutations and somatic mutations.
9. Why are mutations essential for evolution to occur?

Points to Consider

Sometimes even drastic mutations do not affect the proteins produced by a particular type of cell. The reason? The genes affected by the mutations are not normally used to make proteins in that type of cell. In all cells, some genes are turned off - they are not transcribed - while other genes are turned on.

- How do cells control which genes are turned on and used to make proteins?
- Can you think of a mechanism that might prevent transcription of a gene?