Mutations
Adapted from Success in Science: Basic Biology

Key Words

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
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<tbody>
<tr>
<td>Mutations</td>
<td>A change in form</td>
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<tr>
<td>Gene mutation</td>
<td>Permanent change in the DNA of a gene</td>
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<tr>
<td>Chromosome mutation</td>
<td>Permanent change in the number or structure of chromosomes of a cell</td>
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<td>Carrier</td>
<td>Person who has a recessive gene for a trait but does not show the trait</td>
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<tr>
<td>Sex-linked mutation</td>
<td>Mutation carried on either the X or Y chromosome</td>
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Key Ideas

The genetic code is passed from the parents to offspring through gene replication and cell division. A change in a gene or chromosome can occur during either of these processes. Either the type of change can change the genetic code of a cell. If the changed cell is a gamete, then all cells formed will differ from the original parent cell.

Mutation. In mitosis, DNA contained in the nucleus of a parent cell makes copies of itself. This process, called replication, ensures that the resulting daughter cells will contain the same genetic code as the parent cell.

A mutation is a change in something’s form. An error can occur during replication that changes DNA. A permanent change in DNA is a deadly bloody disorder. The disease is the result of a change on one base of a base triplet in the DNA molecule.

Mistakes that occur in cell division may cause a chromosome mutation. Sometimes an error occurs when a chromosome copies itself. The chromosomes in the resulting daughter cells are then different from the parent cell.

In some cases, a portion of a chromosome may not be copied. People who are missing a small part of chromosome 11 have a condition called aniridia. Aniridia is the absence of the iris of the eye. Other times, the cell does not divide evenly. This results in daughter cells with a different number of chromosomes from the parent cell. Down syndrome is an example of this type of mutation.
If a mutation occurs in a gamete, the changed gene or chromosome is passed to the offspring. All cells formed from the gamete contain the mutation. Many types of mutations have little or no harmful effect on the resulting organism. Some mutations can be helpful to an organism. They also add variety to a species.

1. How are gene mutations and chromosome mutations alike?

__________________________________

__________________________________

**Mutation and Disease.** Mutations can be harmful. They can reduce an organism’s chances of survival. For example, genetic disorders in humans are caused by mutations.

Down syndrome is a genetic disorder caused by chromosome mutation. During meiosis, a chromosome pair fails to separate. One of the resulting gametes contains an extra chromosome. The other gamete lacks the chromosome. The gamete with the extra chromosome may join a normal gamete during fertilization. The resulting offspring inherits Down syndrome.

Other genetic disorders are caused by gene mutations. Sickle cell anemia results from a gene mutation. The mutation causes the wrong amino acid to join a protein. The changed protein causes cells to be sickle shaped instead of round. The sickle cells cause clots and deprive the body’s organs of needed oxygen.

The figure shows the genes involved in sickle cell anemia. A person with the sickle cell trait must inherit a mutated gene from both parents. A person who inherits one normal gene and one altered gene is a carrier of the disorder. A carrier has the recessive gene trait but does not actually show the trait. Carriers of the sickle cell trait produce both normal cells, they do not get the disease.

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A = normal allele

S - sickle cell allele

**Sex-linked Mutation.** In humans, certain genes are carried on either the X or Y chromosome. A change in the DNA of these produces a sex-linked mutation. For example, the trait of colorblindness is sex-linked mutation. The genes for color vision are carried on the X chromosome. The trait for normal color vision is carried on the Y chromosome. The gene for colorblindness is recessive.
A female who is homozygous dominant has normal color vision. A female who is heterozygous has normal color vision, but is a carrier of the trait. However, a male will be colorblind only if his mother is a carrier of the trait. There is no gene for color vision on the Y chromosome. The inheritance of colorblindness is shown in the figure.

2. What is a sex-linked mutation?

Check your Understanding

Complete the following with a term from the list below.

**Cell division** chromosome mutation **Down Syndrome** DNA
Gene gene mutation **sickle cell anemia**

During replication, (3) ________________ in the nucleus of a parent cell duplicates. If an error occurs during this process, the resulting (4) ________________ is changed. A permanent change in the DNA of a gene is called a (5) ____________. One genetic disorder caused by a gene mutation is (6) ________________. Errors can also occur during (7) _____________. If a parent cell does not divide evenly a (8) ________________ may occur. One genetic disorder caused by a chromosome mutation is (9) ________________.
10. **WHAT** is a gene mutation?

    **HOW** is it caused?

11. **WHAT** is a chromosome mutation?

    **HOW** is it caused?

12. **HOW** does a mutation HELP a species?

13. Does a carrier for sickle cell anemia show the trait? **EXPLAIN**

14. How are sex-linked mutations and gene mutations **alike**?

    How are they **different**?

15. Why are most colorblind people male?