Lesson

2 Mutations

Key Words

mutation: a change in form

gene mutation: permanent change in the DNA of a gene

chromosome mutation: permanent change in the number or structure of

chromosomes of a cell

carrier: person who has a recessive gene for a trait but does

not show the trait

sex-linked mutation: mutation carried on either the X or Y chromosome

KEY IDEAS

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

Technology has provided methods for making changes to the genetic code of an organism. Plant breeders use these methods to develop useful traits in plants. They change the genetic code of the parent plant's cells to produce helpful mutations. Through experiments and field study, plant breeders "create" new plants. These plants can be made to be more resistant to disease, to produce more offspring, or to taste better than the plants from which they came. Many of the tomatoes you buy at the supermarket have been altered in these ways.

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** (myoo-TAY-shuhn) is a change in something's form. An error can occur during replication that changes DNA. A permanent change in the DNA of a gene is called **gene mutation** (jeen myoo-TAY-shuhn). Sickle cell anemia is a deadly blood disorder. The disease is the result of a change in one base of a base triplet in the DNA molecule.

Mistakes that occur during cell division may cause a **chromosome mutation** (KROH-muh-sohm myoo-TAY-shun). 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.

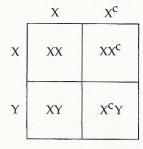
Other genetic disorders are caused by gene mutations. Sickle cell anemia results from a gene mutation. The mutation causes the wrong amino acid to join a protein. The changed protein causes cells to be sickle shaped instead of round. The sickle cells cause blood clots and deprive the body's organs of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

Fig. 12-1 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 (KAR-ee-uhr) has the recessive gene for a trait but does not actually show the trait. Carriers of the sickle cell trait produce both normal and sickle-shaped cells. Because the carriers have enough normal cells, they do not get the disease.

Fig. 12-1

S
S
S
A
AS
AS
S
S
S
S
S
S

A = normal gene S = gene for sickle cell Fig. 12-2



Sex-linked Mutation. In humans, certain genes are carried on either the X or Y chromosomes. A change in the DNA of these genes produces a **sex-linked mutation** (sehks-lihngkt myoo-TAY-shuhn). For example, the trait of colorblindness is a sex-linked mutation. The genes for color vision are carried on the X chromosome. The trait for normal color vision is dominant. The gene for colorblindness is recessive.

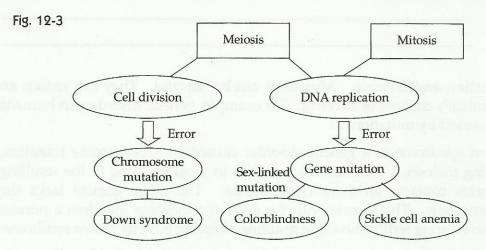
A female who is homozygous dominant has normal color vision. A female who is heterozygous has normal color vision, but is a carrier of the trait. However, a male will be colorblind only if his mother is a carrier of the trait. There is no gene for color vision on the Y chromosome. The inheritance of colorblindness is shown in Fig. 12-2. X^c shows the X chromosome with the gene for colorblindness.



2. What is a sex-linked mutation? ____



Fig. 12-3 shows how mutations can occur during mitosis and meiosis.



Check Your Understanding

Complete each sentence with a term from the list below.

cell division gene		Down syndrome sickle cell anemia	DNA
During replicati	on, (3)	_ in the nucleus o	f a parent cell
duplicates. If	f an error occurs during	this process,	the resulting
(4)	is changed. A pern	nanent change in	the DNA of a
gene is called a (5) Or		ne 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
	omosome mutation is (9)
10.	What is a gene mutation and how is it caused?
11.	What is a chromosome mutation and how is it caused?
12.	How could 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?



