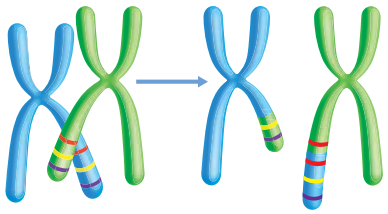
Chromosomal Mutations

Chromosomal mutations are changes in either chromosome segments or whole chromosomes. These mutations may change the amount of genetic material or change the structure of a chromosome, and they usually occur during mitosis and meiosis.

Gene Duplication

During meiosis, homologous chromosomes exchange DNA segments through crossing over. If the chromosomes do not align with each other, a segment of one chromosome may break off and attach itself to the other chromosome, resulting in one chromosome with two copies of a gene or genes. This process is called gene duplication. The chromosome that lost the segment has undergone gene deletion.

FIGURE 10: The douc langur has digestive enzymes that evolved as the result of a gene duplication event. These enzymes allow douc langurs to digest leaves and fruits.



Mutations can have harmful effects, but they can also increase genetic variation, or the variety of traits among individuals within a population. Gene duplication has occurred many times in the evolution of eukaryotic organisms. When gene duplication occurs, multiple copies of a gene are present. As a result, one copy of the gene can encode functional proteins, while the other copies are "free" to accumulate mutations. Mutated genes may encode proteins with new structures, which may take on new functions in the organism.

Model Draw a model illustrating how gene duplication and mutations can lead to a gene with a new function over the course of several generations.

Engineering

Sometimes the entire genome is duplicated. This type of error can lead to polyploidy, or multiple copies of the genome. Genome duplication has occurred in the evolution of many crop plants, such as strawberry, wheat, and mustard plants.

Scientists can use chemicals to artificially induce polyploidy in cells. These chemicals interfere with the formation of microtubules, disrupting the separation of chromosomes during mitosis. As a result, one daughter cell receives a double set of chromosomes. This technique has been used to manipulate traits such as flower size to make plants more desirable to customers.

Analyze Suppose you wanted to chemically induce polyploidy to make a plant with larger fruit. Write a list of questions you would ask to define and delimit the problem.

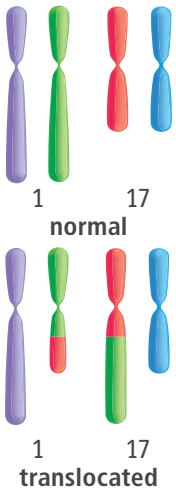
FIGURE 11: Polyploidy in Plants

Common Name	Chromosome Number
Banana	3N= 33
Potato	4N = 48
Common wheat	6N = 42
Boysenberry	7N = 49
Strawberry	8N = 56

Gene Translocation

Translocation is another type of chromosomal mutation. In translocation, a segment of one chromosome moves to a nonhomologous chromosome. Translocations are often reciprocal, which means that the two nonhomologous chromosomes exchange segments with each other. In Figure 12, a translocation occurs between chromosome 1 and chromosome 17. This is known as a balanced translocation because the swapping of segments did not break up any genes, and there was no gain or loss of material.

FIGURE 12: Gene Translocation



Predict Many people with balanced translocation mutations are not aware they have them until they try to have children. How might this be possible?

Nondisjunction Mutations

Nondisjunction mutations occur when one or more homologous chromosomes do not separate during anaphase of meiosis. The resulting gametes do not have the same number of chromosomes and can have more or fewer chromosomes than the parent cell.

FIGURE 13: A karyotype can be used to identify a nondisjunction mutation.

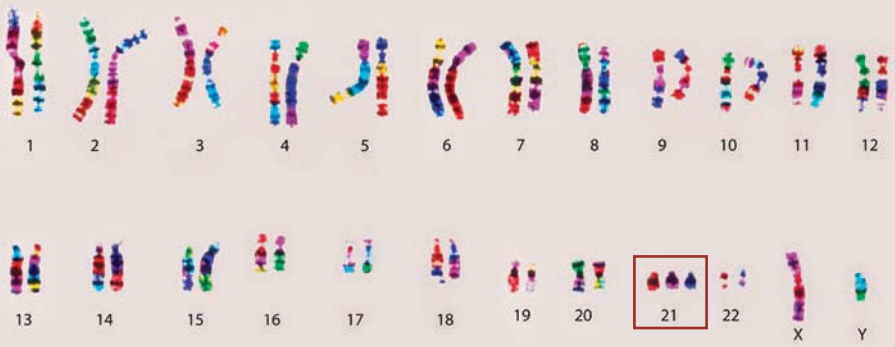


FIGURE 14: People with Down syndrome have three copies of chromosome 21, called trisomy 21.

Examples of human disorders caused by nondisjunction include Down syndrome and Klinefelter disorder. Down syndrome occurs in people with three copies of chromosome 21. Klinefelter disorder is caused by an extra X chromosome in the cells of males. Recall that males have one X and one Y chromosome. A male with this disorder would have three chromosomes: XXY. This mutation affects the learning ability and sexual development of males. Turner syndrome is another example of a disorder caused by nondisjunction. Females with this syndrome have only one X chromosome instead of two. This missing X chromosome interferes with the development of secondary sexual characteristics in females.

Model Draw a model to illustrate how a nondisjunction mutation could occur during either anaphase I or anaphase II of meiosis.

Explain Make a chart to organize and describe the main types of mutations you have learned about so far. Then use your chart to help you write an explanation for these questions: When is a mutation likely to increase genetic variation? When is a mutation likely to have harmful effects?