

37. Use a diagram to show why, for each pregnancy, the chances of giving birth to either a boy or a girl is 50-50. Explain the results shown in your diagram.
38. Explain why hemophilia occurs more often in males than in females. Use a diagram to illustrate your answer.

GENETIC MUTATIONS

Changes in the genetic material are called **mutations**. Mutations in body cells can be passed on to new cells of the individual as a result of mitosis, but they cannot be transmitted to offspring by sexual reproduction. However, mutations in sex cells *can* be transmitted to the next generation. Mutations may involve alterations in chromosomes or alterations in the chemical makeup of genes.

Chromosomal Alterations

Chromosomal alterations involve a change in the structure or number of chromosomes. The effects of chromosomal alterations are often seen in the phenotype of an organism because each chromosome contains many genes.

Nondisjunction. During meiosis, the two chromosomes of each homologous pair separate from each other; each gamete produced by the division receives only one member of each homologous pair. The separation of homologous chromosomes is called *disjunction*. The term *nondisjunction* refers to a type of chromosomal alteration in which one or more pairs of homologous chromosomes fails to separate normally during meiotic cell division (Figure 7-7).

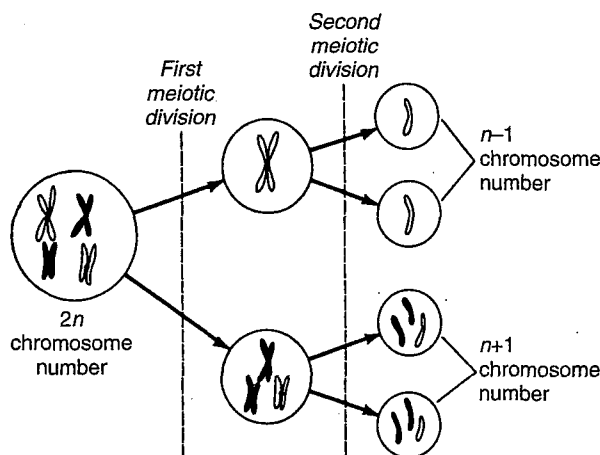


Figure 7-7. Nondisjunction of chromosomes.

As a result of nondisjunction, one of the gametes produced contains both members of the homologous pair, while another gamete contains neither chromosome. Nondisjunction results in the production of some gametes with more chromosomes than normal and some gametes with fewer chromosomes than normal. If one of these abnormal gametes is involved in fertilization, the resulting zygote will have either more than or less than the normal ($2n$) number of chromosomes.

Down syndrome in humans is caused by the presence of an extra chromosome number 21. Nondisjunction during gamete production in one of the parents produces a gamete with an extra chromosome 21. As a result of fertilization, this extra chromosome is transmitted to the offspring.

Polyploidy. Occasionally during gamete formation, a complete set of chromosomes fails to undergo disjunction, and a gamete is produced that contains the diploid ($2n$) chromosome number. If a diploid gamete unites with a normal (n) gamete during fertilization, the resulting zygote will have a $3n$ chromosome number. If two $2n$ gametes fuse, a $4n$ zygote results. The inheritance of one or more complete extra sets of chromosomes is called *polyploidy*. This condition is common in plants but rare in animals. In plants, polyploid individuals are usually larger or more vigorous than the normal, diploid varieties. Certain strains of wheat, potatoes, alfalfa, apples, tobacco, and zinnias are polyploid. Some polyploid plants produce seedless fruit and are sterile.

Changes in Chromosome Structure. Changes in the makeup of chromosomes may result from random breakage and recombination of chromosome parts. *Translocation* occurs when a segment of one chromosome breaks off and reattaches to a nonhomologous chromosome. *Addition* occurs when a segment breaks off one chromosome and reattaches to the homologous chromosome. *Inversion* occurs when a segment breaks off and reattaches in reverse on the same chromosome. *Deletion* occurs when a segment breaks off and does not reattach to any other chromosome.

Gene Mutations

A random change in the chemical makeup of the DNA (genetic material) is a *gene mutation*. The effects of some gene mutations, such as albinism, are noticeable, but other gene mutations may not produce noticeable effects.

Inheritable gene mutations tend to be harmful to the individual. For example, sickle-cell anemia and Tay-Sachs disease are caused by gene

mutations. Fortunately, most gene mutations are recessive and are hidden by the normal, dominant allele. However, if both parents carry the same recessive mutant gene, there is a chance that their offspring will be homozygous recessive and show the harmful trait.

Occasionally, random gene mutations produce changes that make an individual better adapted to the environment. Over time, such helpful mutant genes tend to increase in frequency within a population.

Mutagenic Agents

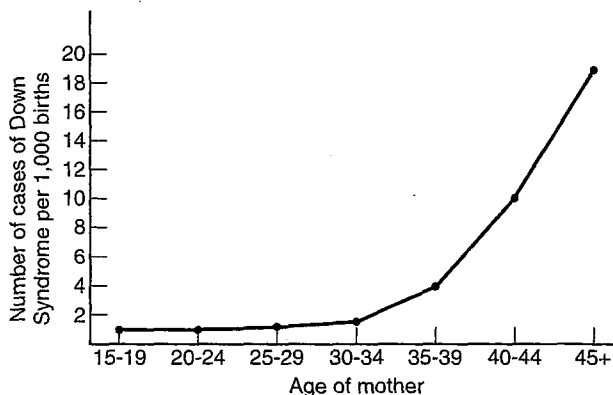
Although mutations occur spontaneously, the rate of mutation can be increased by exposure to certain chemicals and forms of radiation that act as *mutagenic agents*. For example, forms of mutagenic radiation include x-rays, ultraviolet rays, radioactive substances, and cosmic rays. Mutagenic chemicals include formaldehyde, benzene, and asbestos fibers.

QUESTIONS

PART A

39. Which phrase best describes most mutations? (1) dominant and disadvantageous to the organism (2) recessive and disadvantageous to the organism (3) recessive and advantageous to the organism (4) dominant and advantageous to the organism
40. The failure of a pair of homologous chromosomes to separate during meiotic cell division is called (1) nondisjunction (2) translocation (3) addition (4) deletion
41. The condition in which a gamete contains the $2n$ or $3n$ number of chromosomes is called (1) translocation (2) a gene mutation (3) polydactyly (4) polyploidy
42. The presence of only one X chromosome in each body cell of a human female produces a condition known as Turner syndrome. This condition most probably results from the process called (1) polyploidy (2) crossing-over (3) nondisjunction (4) hybridization
43. A random change in the chemical structure of DNA produces (1) polyploidy (2) a translocation (3) nondisjunction (4) a gene mutation
44. Down syndrome in humans is characterized by the presence of an extra chromosome 21 in all cells of the body. The number of chromosomes present in the body cells of individuals with this condition is (1) $n + 1$ (2) $3n$ (3) $2n + 1$ (4) $4n$

45. The graph below shows the relationship between maternal age and the number of children born with Down syndrome per 1000 births.



According to the graph, the incidence of Down syndrome (1) generally decreases as maternal age increases (2) is about nine times greater at age 45 than at age 30 (3) stabilized at 2 per 1000 births after age 35 (4) is greater at age 15 than at age 35

46. Ultraviolet rays, x-rays, and certain other forms of radiation can increase the rate of gene mutation. These forms of radiation are said to act as (1) mutagenic agents (2) catalysts (3) enzymes (4) indicators
47. The large size and exceptional vigor of certain varieties of wheat, apples, and zinnias are due to the possession of extra sets of chromosomes, which result from (1) incomplete dominance (2) gene mutations (3) nondisjunction of complete sets of chromosomes (4) nondisjunction of chromosome number 21 only
48. A type of chromosomal alteration in which a segment of chromosome breaks off and does not reattach to any chromosome is called (1) addition (2) inversion (3) deletion (4) translocation
49. Changes in the genetic code of a human can be transmitted to offspring if they occur in (1) cancer cells (2) gametes (3) cell membranes (4) antibodies

PART B-2

Base your answers to questions 50 through 53 on the following information about an experiment and on your knowledge of biology.

Two groups of 100 lima beans each were used. Group A was exposed to natural light for a period of 24 hours and then planted.

Group B was exposed to microwave energy for 24 hours and then planted under the same conditions as Group A. When the seeds germinated, the plants were observed for growth over a period of two weeks. The results are summarized in the table below.

Group	Number of Plants	
	Normal Growth	Stunted and/or Pale
A	83	17
B	54	46

50. What hypothesis was most likely being tested in this experiment?
51. Describe the results of the experiment.
52. Based on the data, propose a conclusion for the experiment.
53. What are some of the implications of the data?

PART C

54. Explain how it is possible for an individual to inherit an extra chromosome. List two or more human genetic disorders caused by the inheritance of an abnormal number of chromosomes.
55. Mutagens are agents that increase the rate of gene mutations in cells. Identify three types of mutagenic agents and briefly explain how each one causes mutations. Describe how people may reduce their chances of being harmed by these particular agents.

HEREDITY AND THE ENVIRONMENT

The development and expression of inherited traits can be influenced by environmental factors such as nutrients, temperature, sunlight, and so on. The relationship between gene action and environmental influence can be seen in the following examples.

Temperature affects fur color in the Himalayan rabbit. Under normal circumstances, these rabbits are white with black ears, nose, tail, and feet. (The black fur helps the rabbit absorb more heat in its extremities.) However, when some of the white fur on a Himalayan rabbit's back is shaved off and the area kept covered with an ice pack, the new hairs grow in black. The artificial change in temperature produces a change in fur color.

Experiments have shown that the production of chlorophyll requires exposure to sunlight. When parts of a leaf are covered with dark paper, chlorophyll production stops in the area that is covered. Only the exposed part produces chlorophyll, is green, and performs photosynthesis.

Stress and nutrition can affect gene expression. For example, someone who has a tall genotype may not develop a tall phenotype if his or her growth is stunted by malnutrition.

PLANT AND ANIMAL BREEDING

Using the principles of genetics, plant and animal breeders have been able to produce, improve, and maintain new varieties of plants and animals. Methods of **selective breeding** used by such people include artificial selection, inbreeding, and hybridization.

In *artificial selection*, individuals with the most desirable traits (for example, sheep with thick, soft wool) are crossed or allowed to mate in the hopes that their offspring will show the desired traits.

The offspring of selected organisms may be mated with one another to produce more individuals with the desirable traits. This technique, called *inbreeding*, involves the mating of closely related organisms. (Of course, the risk of inbreeding is that harmful recessive genes are more likely to be inherited and cause disorders in the offspring.)

Two varieties of a species may have different desirable traits. In a technique called *hybridization*, breeders cross two such varieties in the hope of producing hybrid offspring that show the desirable traits of both varieties. For example, if one variety of rose has very large petals and another variety has a very sweet scent, their hybrid might show both desirable traits.

QUESTIONS

PART A

56. If bean plant seedlings are germinated in the dark, the seedlings will lack green color. The best explanation for this condition is that (1) bean plants are heterotrophic organisms (2) bean seedlings lack nitrogen compounds in their cotyledons (3) the absence of an environmental factor limits the expression of a genotype (4) bean plants cannot break down carbon dioxide to produce oxygen in the dark
57. In many humans, exposing the skin to sunlight over prolonged periods of time results in

Human Genetic Disorders

Some diseases caused by genetic abnormalities are sickle-cell anemia, Tay-Sachs disease, and phenylketonuria. These disorders are caused by gene mutations.

Sickle-cell anemia is a blood disorder found most commonly in individuals of African descent. The disorder is caused by a gene mutation that results in the production of abnormal hemoglobin molecules and red blood cells. The abnormal hemoglobin and sickle-shaped cells do not carry oxygen efficiently, resulting in anemia. The sickle-shaped red cells also tend to obstruct blood vessels, causing severe pain. Sickle-cell anemia occurs in individuals homozygous for the trait. Both homozygous and heterozygous individuals can be detected by blood tests.

Tay-Sachs disease is a recessive genetic disorder in which nerve tissue in the brain deteriorates because of an accumulation of fatty material. The disorder is a result of the body's inability to synthesize a particular enzyme. Tay-Sachs disease, which is fatal, occurs most commonly among Jewish people of Central European descent.

Phenylketonuria (PKU) is a disorder in which the body cannot synthesize an enzyme necessary for the normal metabolism of the amino acid phenylalanine. The disease, which occurs in homozygous recessive individuals, is characterized by the development of mental retardation. Analysis of the urine of newborn infants can detect PKU. If PKU is detected, mental retardation can be prevented by maintaining a diet free of phenylalanine.

Detection of Genetic Disorders

Some human genetic disorders can be detected either before or after birth by the use of one or more of the following techniques.

Advances in genetic research have resulted in the development of simple blood and urine tests that can determine if an individual has certain genetic disorders. Carriers of sickle-cell anemia and Tay-Sachs disease can be identified by these screening techniques.

Karyotyping is a technique in which a greatly enlarged photograph of the chromosomes of a cell is prepared. The homologous pairs of chromosomes are matched together, and the chromosomes are examined to see if there are any abnormalities in number or structure.

Amniocentesis is a technique in which a small sample of amniotic fluid is withdrawn from the amniotic sac of a pregnant woman. The fluid contains fetal cells, which can be used for karyotyping or for chemical analysis. Amniocentesis

is used in the identification of sickle-cell anemia, Tay-Sachs disease, and Down syndrome in fetuses.

Genetic Counseling

The various techniques described above are used by *genetics counselors* to inform concerned parents about the possible occurrence of genetic defects in their children. For couples whose families show the presence of a particular genetic disorder, a pedigree chart may be developed to predict the probability of their children's having the disorder. Amniocentesis, followed by karyotyping and chemical tests, may be performed once pregnancy is established.

QUESTIONS

PART A

68. An inherited metabolic disorder known as phenylketonuria (PKU) is characterized by severe mental retardation. This condition results from the inability to synthesize a single (1) enzyme (2) hormone (3) vitamin (4) carbohydrate
69. Which statement best describes amniocentesis? (1) Blood cells of an adult are checked for anemia. (2) Saliva of a child is analyzed for the amino acids. (3) Urine of a newborn baby is analyzed for the amino acid phenylalanine. (4) Fluid surrounding a fetus is removed for chemical and genetic analysis.
70. Which is a genetic disorder in which abnormal hemoglobin leads to fragile red blood cells and obstructed blood vessels? (1) phenylketonuria (2) sickle-cell anemia (3) leukemia (4) Down syndrome
71. Human disorders such as PKU and sickle-cell anemia, which are defects in the synthesis of individual proteins, are most likely the result of (1) gene mutations (2) nondisjunction (3) crossing-over (4) polyploidy
72. Which technique can be used to examine the chromosomes of a fetus for possible genetic defects? (1) pedigree analysis (2) analysis of fetal urine (3) karyotyping (4) blood cell tests

PART B-2

73. Give three reasons why a direct study of the inheritance of human traits is difficult to carry out.
74. Briefly describe the two ways that information about patterns of human heredity is usually obtained.