CHAPTER 7

Genetics and Heredity

FOUNDATIONS OF GENETICS

Genetics is the branch of biology that deals with patterns of **inheritance**, or heredity. *Heredity* is the biological process by which parents pass on genetic information to their offspring through their gametes. The science of genetics originated with the work of an Austrian monk, *Gregor Mendel*, who performed a series of experiments on pea plants between 1856 and 1868.

Principles of Mendelian Genetics

In his breeding experiments, Mendel (who, like everyone else at that time, had no knowledge of genes or chromosomes) made careful observations of the inheritance patterns of specific contrasting traits found in pea plants. Through a mathematical analysis of the traits found in the large numbers of offspring from his experimental crosses, Mendel developed his principles of *dominance, segregation,* and *independent assortment.* Mendel also concluded that the traits he observed were controlled by pairs of inherited "factors," with one member of each pair coming from each parent organism. Thus, in organisms that reproduce sexually, half of the offspring's genetic material is contributed by the female parent and half by the male parent. As a result, the offspring has traits from both parents, and is never identical to either one of them.

Gene–Chromosome Theory

The importance of Mendel's work was not recognized until the early 1900s, when the development of better microscopes enabled biologists to observe chromosome behavior during meiotic cell division. Biologists then linked the separation of homologous chromosome pairs during meiosis and their **recombination** at fertilization with the inheritance of Mendel's factors. Breeding experiments carried out by T. H. Morgan with the fruit fly, *Drosophila,* provided supporting evidence for Mendel's principles of inheritance.

Mendel's inherited, or **hereditary**, factors—now known as **genes**—are arranged in a linear fashion on the chromosomes. Each gene has a definite position, or *locus* (plural, *loci*), on the chromosome. The two alternate genes that control each trait are called *alleles,* and they are located in the same position on homologous chromosomes. This *gene–chromosome theory* explains the hereditary patterns observed by Mendel.

Gene Expression

Every organism has at least two alleles that govern every trait. As mentioned, these two genes are passed on—one from the mother and one from the father—to the offspring. The genes encode information that is expressed as the traits of the organism, a phenomenon called **gene expression.** A single gene (that is, one set of alleles) may control one or several traits. Alternatively, some traits are determined by more than one gene (that is, by more than one set of alleles).

Although all the body cells in an organism contain the same genetic instructions, the cells may differ considerably from one another in structure and function. The reason is that, in any given cell, only some of the genes are expressed, while all other genes are inactivated. For example, in liver cells, it is mainly the genes that pertain to liver functions that are active, while the other genes are inactive. The same is true of all other cells in a body. You can think of the genes on a cell's chromosomes as recipes in a cookbook: the book may contain hundreds of recipes, but if you are making a chocolate cake, you will read only the instructions for making that item. Likewise, the cell reads only the instructions for making its specific products.

Genes that are "on" are expressed, while those that are "off" are not expressed. There are many mechanisms that can switch genes on and off, including intracellular chemicals, enzymes, regulatory proteins, and the cell's environment. In addition, a particular gene may alternately be expressed or inactivated, depending on the cell's needs at the time.

SOME MAJOR CONCEPTS IN GENETICS

Dominance

In his experiments, Mendel crossed plants that were pure for contrasting traits. For example, he crossed pure tall plants with pure short plants. All the offspring of such crosses showed only one of the two contrasting traits. In the cross of tall plants and short plants, all the offspring were tall. In this type of inheritance, the allele that is expressed in the offspring is said to be *dominant*; the allele that is present but not expressed is said to be *recessive*. This pattern illustrates Mendel's principle of dominance.

By convention, the dominant allele is represented by a capital letter, while the recessive allele is represented by the lowercase form of the same letter. For example, the allele for tallness, which is dominant, is shown as *T*, while the allele for shortness, which is recessive, is shown as *t*.

If, in an organism, the two genes of a pair of alleles are the same, for example, *TT* or *tt*, the organism is said to be *homozygous*, or pure, for that trait. The genetic makeup of the organism, which is its *genotype*, is either homozygous dominant (*TT*) or homozygous recessive (*tt*). If the two genes of a pair of alleles are different, for example, *Tt*, the organism is said to be *heterozygous*, or *hybrid*, for that trait.

The physical appearance of an organism that results from its genetic makeup is called its *phenotype*. For example, a pea plant that is heterozygous for height has the genotype *Tt* and the phenotype of being tall. When an organism that is homozygous for the dominant trait is crossed with an organism that is homozygous for the recessive trait ($TT \times tt$), the phenotype of the offspring is like that of the dominant parent. Thus, the heterozygous offspring (*Tt*) is tall.

In studies involving genetic crosses, the organisms that are used to begin the studies are called the *parent generation*. The offspring produced by crossing members of the parent generation are called the *first filial*, or F_1, *generation*. The offspring of a cross between members of the F_1 generation make up the *second filial*, or F_2, *generation*.

QUESTIONS

PART A

1. When a strain of fruit flies homozygous for light body color is crossed with a strain of fruit flies homozygous for dark body color, all the offspring have light body color. This illustrates Mendel's principle of (1) segregation (2) dominance (3) incomplete dominance (4) independent assortment
2. Two genes located in corresponding positions on a pair of homologous chromosomes and associated with the same characteristic are known as (1) gametes (2) zygotes (3) chromatids (4) alleles
3. For a given trait, the two genes of an allelic pair are not alike. An individual possessing this gene combination is said to be (1) homozygous for that trait (2) heterozygous for that trait (3) recessive for that trait (4) pure for that trait
4. In pea plants, flowers located along the stem (*axial*) are dominant to flowers located at the end of the stem (*terminal*). Let *A* represent the allele for axial flowers and *a* represent the allele for terminal flowers. When plants with axial flowers are crossed with plants having terminal flowers, all of the offspring have axial flowers. The genotypes of the parent plants are most likely (1) $aa \times aa$ (2) $Aa \times Aa$ (3) $aa \times Aa$ (4) $AA \times aa$
5. Curly hair in humans, white fur in guinea pigs, and needlelike spines in cacti all partly describe each organism's (1) alleles (2) autosomes (3) chromosomes (4) phenotype
6. The appearance of a recessive trait in offspring of animals most probably indicates that (1) both parents carried at least one recessive gene for that trait (2) one parent was homozygous dominant and the other parent was homozygous recessive for that trait (3) neither parent carried a recessive gene for that trait (4) one parent was homozygous dominant and the other parent was hybrid for that trait
7. Which statement describes how two organisms may show the same trait yet have different genotypes for that phenotype? (1) One is homozygous dominant and the other is heterozygous. (2) Both are heterozygous for the dominant tra (3) One is homozygous dominant and th

other is homozygous recessive. (4) Both are homozygous for the dominant trait.

8. In cabbage butterflies, white color (*W*) is dominant and yellow color (*w*) is recessive. If a pure white cabbage butterfly mates with a yellow cabbage butterfly, all the resulting (F_1) butterflies are heterozygous white. Which cross represents the genotypes of the parent generation? (1) *Ww* × *ww* (2) *WW* × *Ww* (3) *WW* × *ww* (4) *Ww* × *Ww*
9. Most of the hereditary information that determines the traits of an organism is located in (1) only those cells of an individual produced by meiosis (2) the nuclei of body cells of an individual (3) certain genes in the vacuoles of body cells (4) the numerous ribosomes in certain cells
10. The characteristics of a developing fetus are most influenced by (1) gene combinations and their expression in the embryo (2) hormone production by the father (3) circulating levels of white blood cells in the placenta (4) milk production in the mother

PART B-2

11. Explain how two organisms can have the same phenotype but different genotypes.
12. To illustrate your answer to question 11, pick a trait and use a letter to represent it. Write the genotypes of the parents and F_1 generations for each organism.

PART C

13. Why do the offspring of sexually reproducing organisms resemble both parents? Why are they not identical to either one of the parents?
14. Explain why the body cells of an organism can differ in structure and function, even though they all contain the same genetic information.

Segregation and Recombination

When gametes are formed during meiosis, the two chromosomes of each homologous pair separate, or *segregate*, randomly. Each gamete contains only one allele for each trait. After the gametes fuse during fertilization, the resulting (zygote) cell contains pairs of homologous chromosomes, but new combinations of alleles may be present. This process is described by Mendel's principle of segregation.

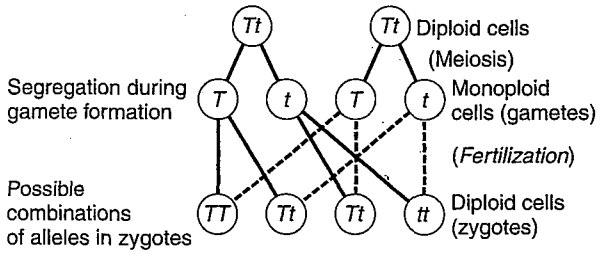


Figure 7-1. Segregation and recombination of alleles.

Figure 7-1 illustrates segregation and recombination in a cross between two individuals that are heterozygous for tallness. In a large number of such crosses, with a large number of offspring, two types of numerical ratios can be observed. In terms of genotype, the ratio is 1 homozygous dominant (*TT*) : 2 heterozygous (*Tt*) : 1 homozygous recessive (*tt*). In terms of phenotype, the ratio is 3 tall : 1 short. These genotype and phenotype ratios are typical for all crosses between organisms that are hybrid for one trait.

The Testcross

To determine the genotype of an organism that shows the dominant phenotype, a testcross is performed. In a *testcross*, the organism in question is crossed with a homozygous recessive organism (Figure 7-2). If the test organism is homozygous dominant, all the offspring will be heterozygous and show the dominant phenotype. If any

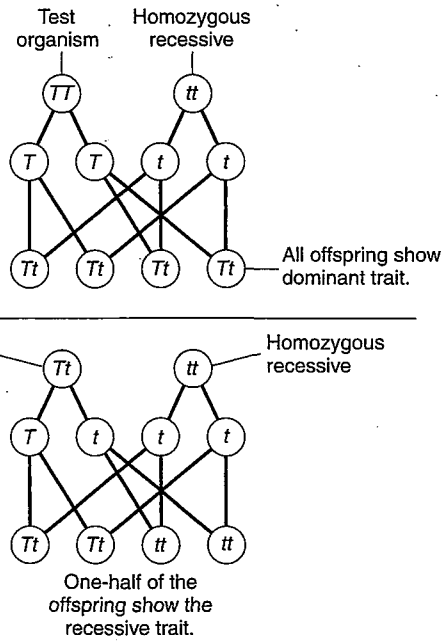


Figure 7-2. Use of a testcross to determine an organism's genotype.

offspring show the recessive phenotype, the individual being tested would have to be heterozygous.

Punnett Square

The possible offspring of a genetic cross are often shown with a diagram called a *Punnett square.* We can use a Punnett square to show the possible offspring of a cross between a heterozygous tall pea plant (*Tt*) and a homozygous short pea plant (*tt*).

The first step in using a Punnett square is to determine the possible genotypes of the gametes of each parent. In this example, the heterozygous tall plant (*Tt*) produces two types of gametes: half will contain the dominant gene for height, *T*, and half will contain the recessive gene, *t*. The gametes of the homozygous short plant (*tt*) will each contain the recessive gene for height, *t*.

As shown in Figure 7-3, the letters that represent the trait carried by the gametes of one parent are written next to the boxes on the left side of the square; the letters for the gametes of the other parent are written above the boxes on top of the square. The letters are combined to show offspring genotypes as follows: letters on top of the square are written in the boxes below them, and letters on the side are written in the boxes to the right of them. The dominant gene, when present, is written first. The pairs of letters in the four boxes represent the possible combinations of genes in the offspring of the cross. Of the possible offspring of this cross, half would be heterozygous tall (*Tt*) and half would be homozygous (recessive) short (*tt*).

Linkage

Mendel's observation of the independent inheritance of different traits was the basis for his principle of independent assortment. When the events of meiosis were discovered, it became clear that traits are inherited independently of

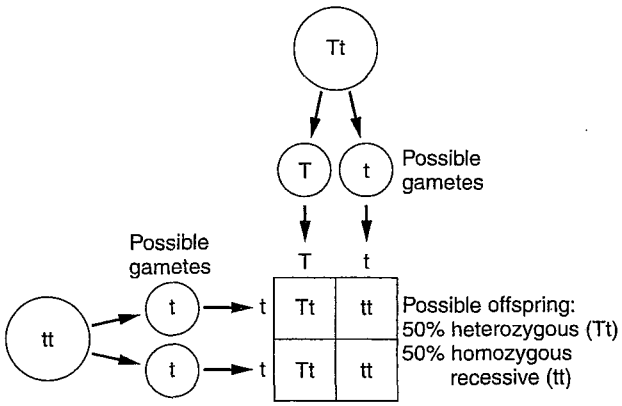


Figure 7-3. Use of a Punnett square to determine possible genotypes of offspring.

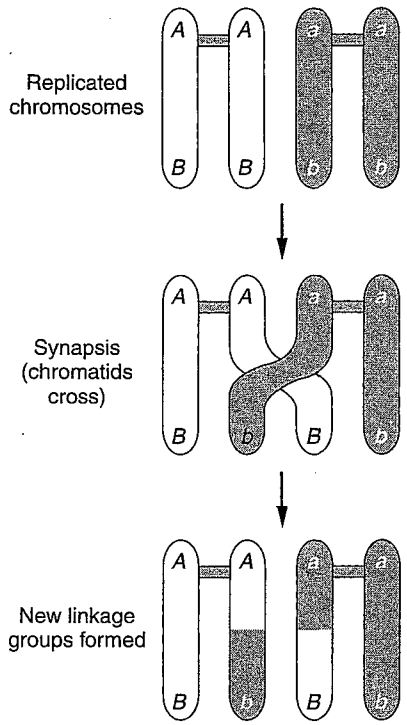


Figure 7-4. Crossing-over of chromatids.

one another only when their genes are on nonhomologous chromosomes. However, when the genes for two different traits are located on the same pair of homologous chromosomes, they tend to be inherited together. Such genes are said to be *linked.* The patterns of inheritance and phenotype ratios for linked traits are different from those of nonlinked traits (the kind observed by Mendel).

Crossing-Over

During *synapsis* in the first meiotic division, the chromatids of a pair of homologous chromosomes often twist around each other, break, exchange segments, and rejoin (Figure 7-4). This exchange of segments, called *crossing-over,* results in a rearrangement of linked genes and produces variations in offspring. Crossing-over is an important source of genetic variation in sexual reproduction.

QUESTIONS

PART A

15. Polydactyly is a characteristic in which a person has six fingers per hand. Polydactyly is dominant over the trait for five fingers. If a man who is heterozygous for this trait marries a woman with the normal number of fingers,

what are the chances that their child would be polydactyl? (1) 0% (2) 50% (3) 75% (4) 100%

16. A cross between two pea plants that are hybrid for a single trait produces 60 offspring. Approximately how many of the offspring would be expected to exhibit the recessive trait? (1) 15 (2) 45 (3) 30 (4) 60

17. Which principle states that during meiosis chromosomes are distributed to gametes in a random fashion? (1) dominance (2) linkage (3) segregation (4) mutation

18. In guinea pigs, black coat color is dominant over white coat color. The offspring of a mating between two heterozygous black guinea pigs would probably show a phenotype ratio of (1) two black to two white (2) one black to three white (3) three black to one white (4) four black to zero white

19. The offspring of a mating between two heterozygous black guinea pigs would probably show a genotype ratio of (1) 1 *BB* : 2 *Bb* : 1 *bb* (2) 3 *Bb* : 1 *bb* (3) 2 *BB* : 2 *bb* (4) 2 *BB* : 1 *Bb* : 1 *bb*

20. If a breeder wanted to discover whether a black guinea pig was homozygous (*BB*) or heterozygous (*Bb*) for coat color, the animal in question would have to be crossed with an individual that has the genotype (1) *BB* (2) *bb* (3) *Bb* (4) *BbBb*

21. Mendel's principle of independent assortment applies to traits whose genes are found on (1) homologous chromosomes (2) sex chromosomes (3) the same chromosome (4) nonhomologous chromosomes

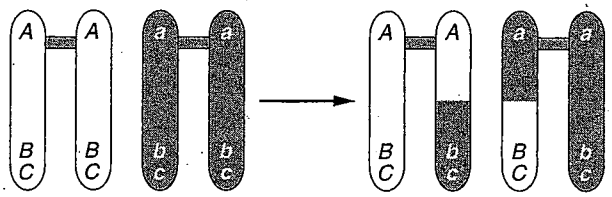
22. The process in which the chromatids of pairs of homologous chromosomes exchange segments is called (1) linkage (2) crossing-over (3) independent assortment (4) intermediate inheritance

23. In horses, black coat color is dominant over chestnut coat color. Two black horses produce both a black-coated and a chestnut-coated offspring. If coat color is controlled by a single pair of genes, it can be assumed that (1) in horses, genes for coat color frequently mutate (2) one of the parent horses is homozygous dominant and the other is heterozygous for coat color (3) both parent horses are homozygous for coat color (4) both parent horses are heterozygous for coat color

PART B-2

24. Based on your answer to question 23, explain how two black horses could produce a chestnut-colored offspring.

Base your answers to questions 25 through 27 on the diagram below, which represents a pair of homologous chromosomes at the beginning of meiosis. The letters A, B, C, a, b, *and* c *represent pairs of alleles located on the chromosomes.*



25. Compare the sets of chromosomes on the left with those on the right. Explain what has happened.

26. What process (not shown) is responsible for the observed results?

27. How does this process lead to variations among offspring?

PART C

28. When is a testcross used? Explain how it works.

29. Explain the following statement: Traits are inherited independently of one another only if their genes are on non-homologous chromosomes. You may use diagrams to support your explanation.

Sex Determination

The diploid cells of many organisms contain two types of chromosomes: *autosomes* and *sex chromosomes*. There is generally one pair of sex chromosomes, and all the other chromosomes are autosomes. In human body cells there are 22 pairs of autosomes and one pair of sex chromosomes. The sex chromosomes are called the *X* and *Y* chromosomes. Females have two *X* chromosomes, and males have one *X* and one *Y* chromosome.

During meiotic cell division, the sex chromosomes, like all other chromosome pairs, are separated (Figure 7-5, page 102). The resulting gametes contain only one sex chromosome. Since females have two *X* chromosomes, each female gamete receives an *X* chromosome. Since the genotype of males is *XY*, sperm cells may receive either an *X* or a *Y* chromosome. The sex of the offspring is determined at fertilization and depends on whether the egg is fertilized by a sperm with an *X* or a sperm with a *Y* chromosome. If the sperm has an *X* chromosome, the resulting zygote will be female (*XX*). If the sperm has a *Y* chromosome, the resulting zygote will be male (*XY*).

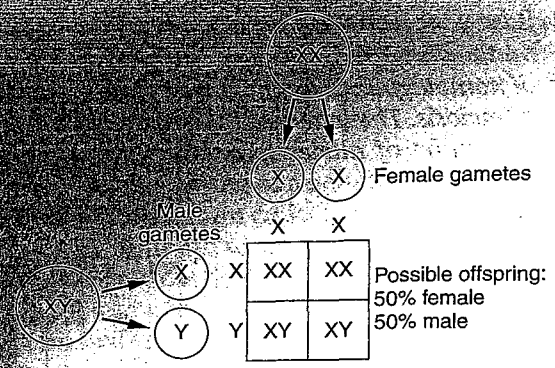


Figure 7-5. Sex determination of offspring.

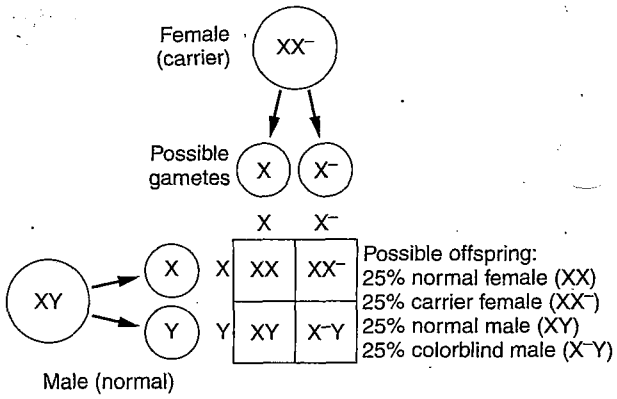


Figure 7-6. Inheritance of color blindness.

Sex-linked Traits

T. H. Morgan, in his experiments with fruit flies, found that some rare, abnormal recessive traits appear with greater frequency in males than in females. From his observations, Morgan concluded that the genes for these traits are present on the *X* chromosome and that there are no corresponding alleles for these traits on the *Y* chromosome. Genes found on the *X* chromosome are called *sex-linked genes*. Recessive sex-linked traits appear more frequently in males than in females because in females there is usually a normal, dominant allele on the other *X* chromosome, so that the phenotype is normal. In males, there is no second allele, so the presence of one recessive gene produces a recessive phenotype.

Both *hemophilia* and *color blindness* are sex-linked disorders; they occur more frequently in males than in females. Hemophilia is a condition in which the blood does not clot properly, while color blindness is an inability to distinguish certain colors. The genes for normal blood clotting and normal color vision are dominant; the genes for hemophilia and color blindness are recessive. For a female to show either of these disorders, she must have recessive genes (alleles) on both of her *X* chromosomes. Females with one normal, dominant gene and one recessive gene for these disorders are called *carriers*. They can pass the disorder to their offspring but do not themselves show symptoms of the disorder. Figure 7-6 shows the possible genotypes of children of a normal male and a female carrier of color blindness.

QUESTIONS

PART A

30. If a color-blind man marries a woman who is a carrier for color blindness, it is most probable that (1) all of their sons will have normal color vision (2) half of their sons will be color-blind (3) all of their sons will be color-blind (4) none of their children will have normal color vision

31. A color-blind man marries a woman with normal vision. Her mother was color-blind. They have one child. What is the chance that this child will be color-blind? (1) 0% (2) 25% (3) 50% (4) 100%

32. A color-blind woman marries a man who has normal color vision. What are their chances of having a color-blind daughter? (1) 0% (2) 25% (3) 75% (4) 100%

33. Which parental pair could produce a color-blind female? (1) homozygous normal-vision mother and color-blind father (2) color-blind mother and normal-vision father (3) heterozygous normal-vision mother and normal-vision father (4) heterozygous normal-vision mother and color-blind father

34. Which statement correctly describes the normal number and type of chromosomes present in human body cells of a particular sex? (1) Males have 22 pairs of autosomes and 1 pair of *XX* sex chromosomes. (2) Females have 23 pairs of autosomes. (3) Males have 22 pairs of autosomes and 1 pair of *XY* sex chromosomes. (4) Males have 23 pairs of autosomes.

35. Based on the pattern of inheritance known as sex linkage, if a male is a hemophiliac, how many genes for this trait are present on the sex chromosomes in each of his diploid cells? (1) 1 (2) 2 (3) 3 (4) 4

36. Traits controlled by genes on the *X* chromosome are said to be (1) sex-linked (2) mutagenic (3) incompletely dominant (4) homozygous