Chapter 15: The Chromosomal Basis of Inheritance

15.1 Discuss Mendel's laws of inheritance in relation to the behavior of chromosomes during meiosis and fertilization, and explain Morgan's experiments with red- and white-eyed fruit flies.

15.2 Describe the inheritance patterns displayed by sex-linked genes and the effects of X inactivation in female mammals.

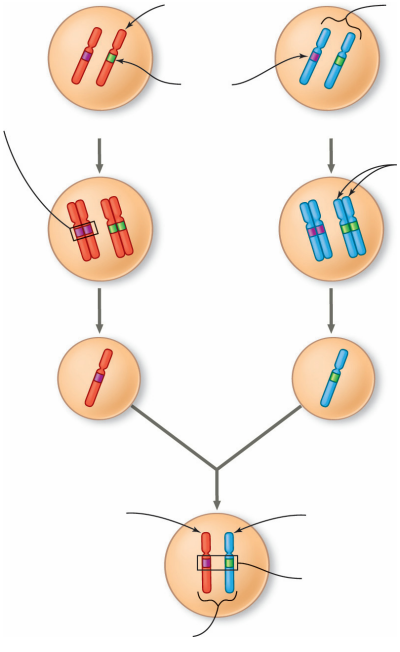
15.3 Explain how linkage of genes and crossing over affect inheritance.

15.4 List different types of alterations to chromosome number and structure, and explain how they can cause genetic disorders.

15.5 Identify the basis for non-Mendelian inheritance patterns shown by (1) organelle genes and (2) imprinted genes.

Your study of genetics continues with this chapter. There are many problems that illustrate the fine points of inheritance, including sex-linkage, linkage, and pedigrees. Practice doing the problems and associated exercises, and you will be rewarded with better understanding of the concepts.

Study Tip: What is the relationship between genes and chromosomes? Figure 15.1 says it all with respect to genes, chromosomes, meiosis, and fertilization. It is essential for you to be confident you understand how all of these relate. Your study of Chapters 13 and 14 has given you sufficient background to complete all the labels for this diagram. Test yourself by trying to add all the labels before you look at it in your text. You know this!



Just as with Chapter 14, this topic is much like being in a math class, so plan to put pencil to paper to illustrate situations and do lots of problems. This is what we mean by "Active Learning". As is generally the case with learning, "reading" the chapter is not enough to fully understand the material.

Homologous chromosomes may have alleles that are the same or they may have alleles that are different. Explain how this can be possible. Figure 15.1 will help with your answer.

Concept 15.1 Mendelian inheritance has its physical basis in the behavior of chromosomes

LO 15.1: Discuss Mendel's laws of inheritance in relation to the behavior of chromosomes during meiosis and fertilization, and explain Morgan's experiments with red- and white-eyed fruit flies.

1. How does the *chromosome theory of inheritance* provide a physical explanation for Mendelian inheritance?

2. The first evidence that a specific gene was associated on a specific chromosome came from the work of *Thomas Hunt Morgan* in the early 1900s. He selected *Drosophila melanogaster* as his experimental organism. List at least three reasons why the fruit fly is an excellent subject for genetics studies.

3. What is meant by the *wild type*?

   What is the alternative to the wild type phenotype?

4. The notation developed for wild type and mutant traits follows some accepted conventions. Notate the following genotypes for a female fruit fly:

   a. a fly homozygous for red eyes _____

   b. a fly heterozygous for red eyes _____

   c. a fly homozygous for white eyes _____

5. When Thomas Hunt Morgan mated his first white-eyed male fly with a red-eyed female fly, he came to the startling conclusion that the trait for eye color was located on the chromosome that determines sex. What was the phenotypic result of this cross in the F_1 and F_2 generations? Show this cross.

6. Note that Morgan made a major breakthrough in thinking about inheritance. He showed that genes are located on specific chromosomes. Work through Inquiry Figure 15.3 in your text to follow his results and conclusions.

   a. What would Mendelian genetics predict for the results of the F_1 cross described in question 5?

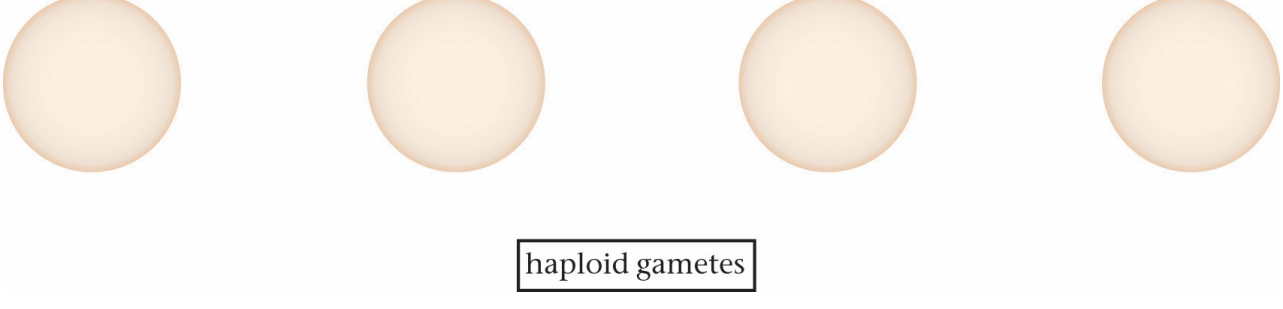
   b. What unusual result of phenotypes was seen in the F_2 generation of that cross?

   c. What does this suggest?

7. Figure 15.4 in your text is very important, as it ties together Mendel's laws, which you studied in Chapter 14, and the chromosome theory of inheritance. Notice that each resultant gamete has one long chromosome and one short chromosome. Which Mendelian law explains this?

8. Each resultant gamete has one of *each* homologous chromosome pairs. Which Mendelian law explains this?

9. Use the F_1 generation gamete formation from Figure 15.4 in your text to illustrate the *law of segregation* and the *law of independent assortment* by showing the four possible haploid gametes that could result in meiosis. Use two different colored markers to show this on the figure below.



10. Going back to chromosome arrangement in meiosis, when do the two events you illustrated in question 9 occur?

11. To demonstrate that you understand this concept, consider a cell with two pairs of chromosomes. Sketch the four different ways these chromosomes might be arranged during metaphase I of meiosis.

Concept 15.2 Sex-linked genes exhibit unique patterns of inheritance

LO 15.2: Describe the inheritance patterns displayed by sex-linked genes and the effects of X inactivation in female mammals.

12. Humans and other mammals have two types of sex chromosomes, designated **X** and **Y**. Describe how they differ from each other and make a small sketch to illustrate this.

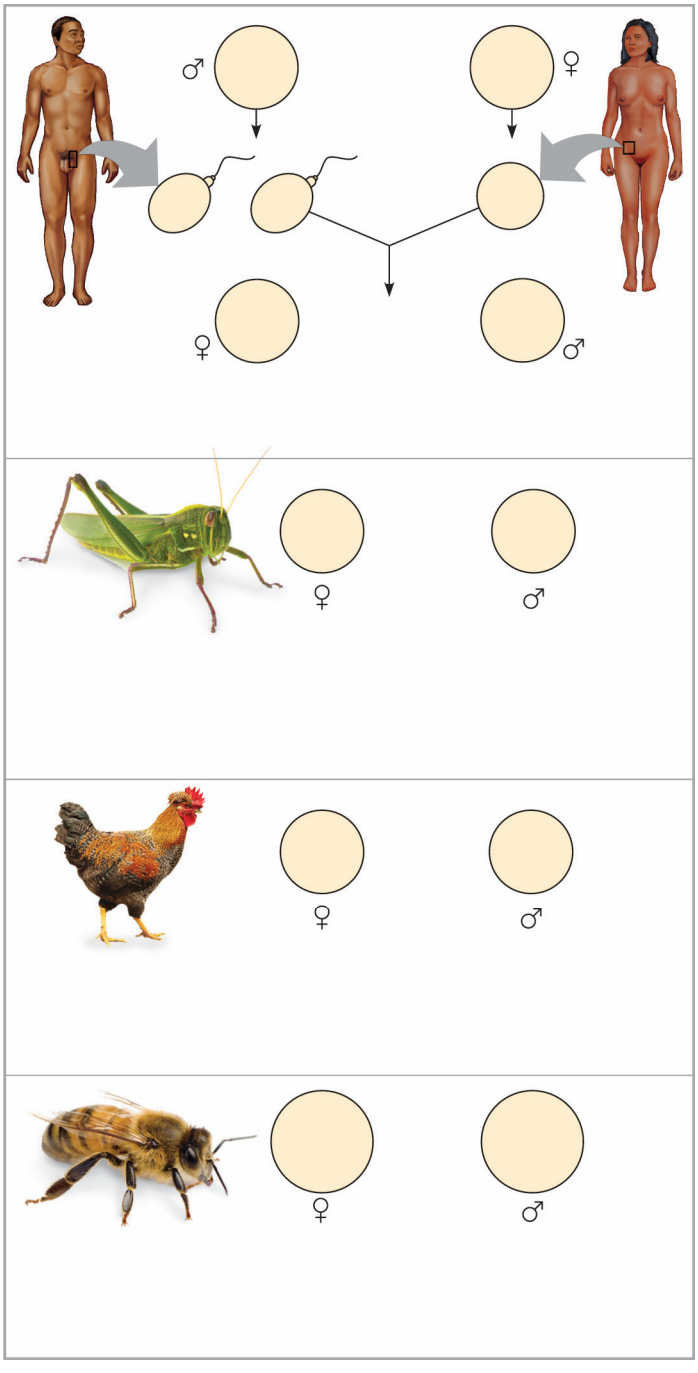
13. There are several variations in the way sex is determined in different species. Complete the following figure to name and explain four different methods of sex determination.

a. _____

b. _____

c. _____

d. _____



14. What is the *SRY* gene? Where is it found, and what does it do?

15. What is the definition of a *sex-linked gene*?

In humans, the term *sex-linked gene* refers to genes located on the portion of the X chromosome that has no homologous portion on the Y chromosome. The genes are actually X-linked genes and the ones we will refer to now.

16. Name and describe three human sex-linked disorders.

    1.

    2.

    3.

17. Figure 15.7 in your text gives three sample problems. Make a key to show the notation that you will use to indicate:

    _____ A male with color-blindness

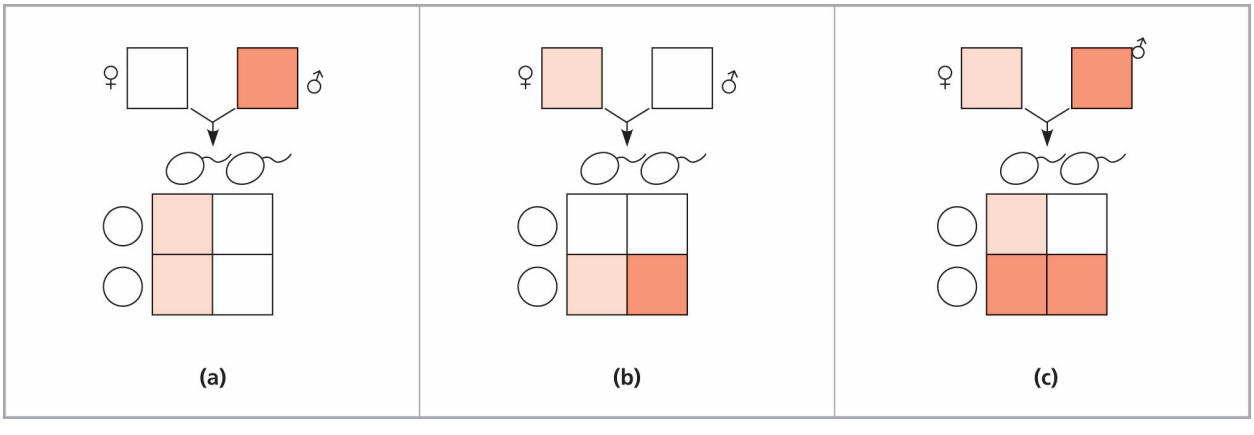
    _____ A female carrier of color-blindness

    _____ A male with normal vision

    _____ A female who is a not a carrier and has normal vision

    _____ A female with color-blindness

18. Work through problems 15.7a, 15.7b, and 15.7c in the Punnett squares provided below. Initially try the problems without help from the text figure, and then check your work.



19. Neither Tim nor Shonda has Duchenne muscular dystrophy, but their firstborn son does. Make a Punnett square on the next page to show the parental genotypes as well as all possible outcomes for their children. Then use your results to answer the questions that follow. (*This is question 2 from Concept Check 15.2, and the answer is in Appendix A of your textbook.*)

a._____ What is the probability that a second child will have the disease?

b._____ What is the probability if the second child is a boy?

c. _____ What is the probability if the second child is a girl?

20. What is a *Barr body*? Why do human females show a Barr body in their cells?

21. X inactivation maintains the proper gene dosage. How is the X chromosome inactivated?

22. Why can you say that all calico cats are females?

Concept 15.3 Linked genes tend to be inherited together because they are located near each other on the same chromosome

LO 15.3: Explain how linkage of genes and crossing over affect inheritance.

23. What are *linked genes*? Do linked genes sort independently?

24. If two genes are linked on the same chromosome, we call this combination the *parental combination*. These genes will be transmitted as a unit and will not sort independently. However, during meiosis, *crossing over* occurs between homologous chromosomes, and the linked genes can become "unlinked." In general, the farther two genes are from each other along the chromosome, the more often they will come "unlinked." *Genetic recombination* is the process during which linked genes become unlinked. What do geneticists call the offspring that show these new combinations?

25. Review meiosis. When does *crossing over* occur?

26. *Alfred H. Sturtevant*, a student of Thomas Hunt Morgan, used assumptions from observations of crossovers to map genes. What is a *linkage map*?

27. What is a *map unit*?

28. What would be the phenotypic ratio of a testcross between a wild-type dihybrid female fly with gray body and normal wings, and a homozygous recessive fly with black body, vestigial wings if the genes were *not* linked? Show the four possible phenotypes and the expected ratio below. (Inquiry Figure 15.9 on p.301 may be helpful.)

29. Your answer for question 28 serves as a point of comparison with the actual results. Because these genes *are* linked, the results are not what might have been predicted if the genes are on different chromosomes.

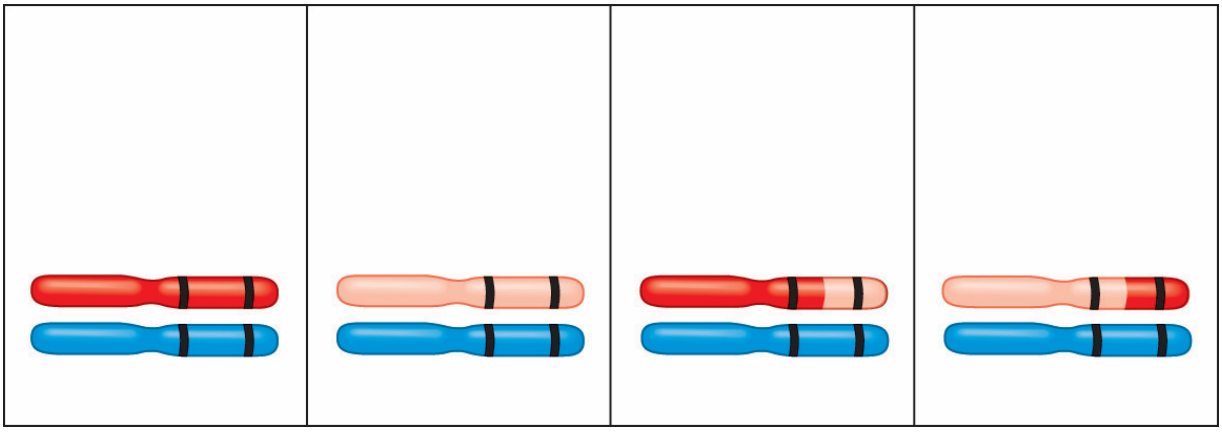
    Refer now to Figure 15.10 in your text.

    a. Is the parental female homozygous or heterozygous?

    b. Is the F_1 female homozygous or heterozygous?

    c. Why can the parental female produce only one type of gamete but the *phenotypically* identical F_1 female produce four types of gametes?

30. Show the phenotypes and number of each type of testcross offspring in the figure below. Indicate which offspring are the recombinants and which are the parental type. Finally, calculate the map distance between the two genes. Show all your work here and on the figure that follows.



31. Suppose alleles M and n are linked on one chromosome, and m and N are linked on the homologous chromosome. Individuals homozygous for M and n are mated with individuals homozygous for m and N. Their offspring are test crossed and the following results are recorded:

Mn/mN	232
mN/mN	240
MN/mn	15
mn/mn	13

How many units apart are these genes on the chromosome? (You will find the answer at the end of this section.)

32. When you are given the frequency of crossing over between genes, it is possible to map their location on a chromosome. (As a hint, place the two genes that have the greatest frequency of crossing over at either end and work from there.) What is the sequence of these genes on the chromosome if the frequency of crossing over between linked genes A and B is 35 percent; between B and C, 10 percent; between C and D, 15 percent; between C and A, 25 percent; between D and B, 25 percent? (Answer at the end of this chapter.)

Study Tip

The Scientific Skills Exercise on p. 304 will help you understand and practice the use of the Chi-Square Test. If your instructor has introduced you to this statistical test, you should work through this problem.

Concept 15.4 Alterations of chromosome number or structure cause some genetic disorders

LO 15.4: List different types of alterations to chromosome number and structure, and explain how they can cause genetic disorders.

33. What occurs in *nondisjunction*?

34. Explain each of the following terms:

    aneuploidy

    monosomy

    trisomy

    polyploidy

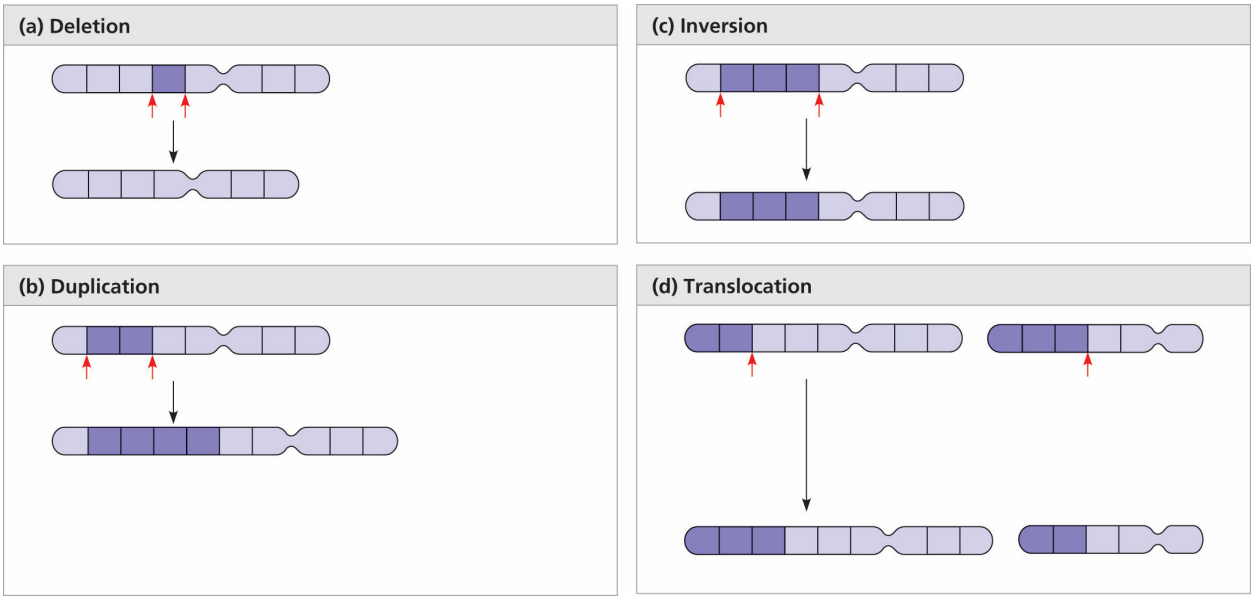
    Monosomy and trisomy are estimated to occur in _____ of human conceptions and are the main reason for pregnancy loss.

35. What causes *Down syndrome*? What are four characteristics of Down syndrome?

36. For each of the following human aneuploidies, give the sex of the individual as well as any physical manifestation of the syndrome.

	Sex	Physical Traits
XXY		
XXX		
XO		
XYY		

37. Chromosome structure can be altered in several ways. Place letters in the blocks to represent genes, and then explain what occurs in each type of alteration.



Concept 15.5 Some inheritance patterns are exceptions to standard Mendelian inheritance

LO 15.5: Identify the basis for non-Mendelian inheritance patterns shown by (1) organelle genes and (2) imprinted genes.

38. A number of genes will cause a variation in phenotype, depending on whether the gene came from the father or the mother. This variation occurs because of *genomic imprinting*. Explain genomic imprinting.

39. Although you inherited one chromosome of each pair from your mother and your father, you have inherited a group of genes from your mother only. What genes are these?

40. You should have identified mitochondrial DNA as the correct response to question 39 above. What other organelle has its own genes? These are *extranuclear genes*.

Answer to question 29: 5.6 map units. Answer to question 30: A D C B

Test Your Understanding, p. 313

Complete the following questions from the problems at the end of the chapter: questions 1, 2, 3, 6, 7.

Just as in Chapter 14, this section asks you to do a series of problems. One way to determine your understanding of genetics is to work many genetics problems, so don't skip this important step! Work neatly and show all work. As you know, you can check your solutions in your text.