Chapter 14: Mendel and the Gene Idea

14.1 Explain Mendel's two laws of inheritance.

14.2 Describe the laws of probability and explain how they can be used to solve complex genetics problems.

14.3 Use examples to show how genetic inheritance patterns can be affected by complete dominance, incomplete dominance, codominance, multiple alleles, pleiotropy, epistasis, and polygenic inheritance.

14.4 Explain what a pedigree is and use examples to show how human traits follow Mendelian patterns of inheritance.

If you have completed a first-year high school biology course, some of this chapter will serve as a review for the basic concepts of Mendelian genetics. For other students, this may be your first exposure to genetics. In either case, this is a chapter that should be carefully mastered. Spending some time with this chapter, especially working genetics problems, will give you a solid foundation for the extensive genetics in the chapters to come.

Study Tip: There is no shortcut to understanding genetics. If you do numerous problems, it will soon make sense. We recommend that you print out p. 292 "Tips for Genetics Problems" and follow its suggestions. This part of your biology course is much like being in a math class, so plan to put pencil to paper and do *lots* of problems. Figure 14.1 in your chapter can be understood in light of what you learned in the last chapter on meiosis.

a. Why does meiosis result in four haploid gametes?

b. The offspring of purple and white-flowering plants all produce purple flowers, but still are able to form gametes for white-flowering offspring. What principle does this demonstrate?

Concept 14.1 Mendel used the scientific approach to identify two laws of inheritance

LO 14.1: Explain Mendel's two laws of inheritance.

1. What is the difference between a *character* and a *trait*? Explain using an example.

2. One of the keys to success for Mendel was his selection of pea plants. Not only were there many varieties, they also had short generation time and large numbers of offspring were produced. How did Mendel control the crosses so he was always certain of their parentage?

3. Define the following terms.

   P generation

   F_1 generation

   F_2 generation

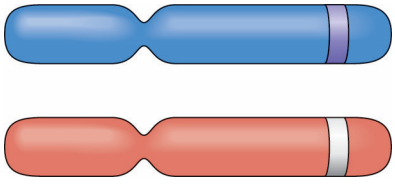
4. Explain how Mendel's simple cross of purple and white flowers did the following:

   a. refuted the "blending" hypothesis:

   b. determined dominant and recessive characteristics:

   c. demonstrated the merit of experiments that covered multiple generations:

5. Refer to Figure 14.4 in your text to label the *allele* for both purple and white flower color, a *homologous pair*, and the *locus* of the flower color gene.



6. The entire Figure 14.4 in your text explains a very important concept that many students have not considered: *Why is a particular trait recessive?* Note the difference shown between the DNA nucleotide sequences for the two different color alleles. Explain, at the molecular level, why purple or white flowers result.

7. In sexually reproducing organisms, why are there exactly two chromosomes in each homologous pair?

8. Mendel's model consists of four concepts. Describe each concept in the appropriate space below.

Mendel's Four Concepts	Description of Concept
First concept	
Second concept	
Third concept	
Fourth concept (law of segregation)	

9. In the third concept of Mendel's model, he explains that the _____ determines the organism's appearance if the two alleles at a locus differ while the _____ has no noticeable effect on the organism's appearance.

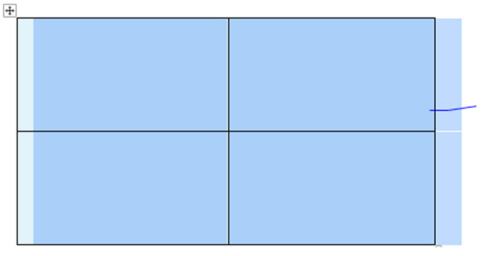
*We will use shorthand to indicate dominant and recessive alleles in working genetics problems. So many of our students make mistakes on problems because of their handwriting. Our advice is to use uppercase block print for dominant alleles (*A*) and lowercase script (a) for recessive alleles.*

Always begin each problem by writing out a "key" to the symbols you use. For a cross using purple and white flowered peas, this would be:

P = purple flowers	*p* = white flowers

10. Figure 14.5 in your text explains how to construct a Punnett square for a cross between true-breeding plants with purple or white flowers. You are going to reproduce this figure but use symbols instead of pictures.

Make a key for this problem. Use these symbols to show the cross for the P generation, the offspring in the F_1 generation, and both the cross and the offspring in the F_2 generation. Indicate the alleles for each individual as well as the gametes it produces and complete the Punnett square.

Key:	
P Generation	
F_1 Generation	
F_2 Generation	

11. Genetics problems have their own vocabulary, and you will have difficulties in solving them unless you master the following terms. *Define* the terms and give an example of each *using the following symbols:*

P = purple flowers	*p* = white flowers

homozygous

heterozygous

genotype

phenotype

parental cross

monohybrid cross

dihybrid cross

12. Refer to the problem you did in question 10 to answer the following questions:

    a. What is the F_2 phenotypic ratio?

    b. What is the F_2 genotypic ratio?

    c. Which generation is heterozygous?

    d. Which generation has both heterozygous and homozygous offspring?

13. In pea plants, *T* indicates the allele for tall plants, and *t* is the allele for dwarf plants. If you have a tall plant, demonstrate with a *testcross* how it could be determined if the plant is homozygous tall or heterozygous tall.

14. As you start to work word problems in genetics, two things are critical: the parent's genotype must be correct, and the gametes must be formed correctly. Using Figure 14.8 in your text as your guide, explain how the gametes are derived for the following cross. (You should have four different gametes. This is Mendel's law of independent assortment in action.)

$$YyRr \times YyRr$$

15. Complete the cross given in question 14 by placing the gametes for each parent below, along the top and sides of the table. Fill in the possible combinations in the *Punnett square*. Finally, provide the phenotypic ratio of the offspring.

Phenotypes and Phenotypic Ratio:

16. Explain Mendel's *law of independent assortment*.

17. Independent assortment of chromosomes occurs in meiosis. Refer to Figure 13.11, p. 265, in your text. Explain the event in meiosis when independent assortment occurs.

Before leaving this concept, complete the three problems in Concept Check 14.1 on p. 276 of your textbook. The problems are worked and explained in the Answer section on p. A-13 at the back of the book.

Concept 14.2 Probability laws govern Mendelian inheritance

LO 14.2: Describe the laws of probability and explain how they can be used to solve complex genetics problems.

18. An event that is certain to occur has a probability of_____ , and an event that is certain not to occur has a probability of _____.

19. In probability, what is an *independent event*?

20. State the *multiplication rule* and give an original example.

21. State the *addition rule* and give an original example.

22. What is the probability that a couple will have a girl, a boy, a girl, and a boy in this specific order? Show your calculation.

Complex genetics problems would require a great deal of time if solved using Punnett squares. For example, a trihybrid cross would need a 64-square box to solve! Using the multiplication and addition rules allows you to "shortcut." The section that begins on p. 277 of your text "Solving Complex Genetics Problems with the Rules of Probability" will take you through the steps and thinking you need to use this technique.

Before leaving this concept, complete the three problems in Concept Check 14.2 on p. 278 of your textbook. The problems are worked and explained in the Answer section on p. A-14 at the back of the book. Remember, you will master this material much as you succeed in learning mathematics—practice and more practice!

Concept 14.3 Inheritance patterns are often more complex than predicted by simple Mendelian genetics

LO 14.3: Use examples to show how genetic inheritance patterns can be affected by complete dominance, incomplete dominance, codominance, multiple alleles, pleiotropy, epistasis, and polygenic inheritance.

23. Alleles can show different degrees of dominance. Explain how *incomplete dominance* is different from *complete dominance* and give an example of incomplete dominance.

24. Compare and contrast *codominance* with *incomplete dominance*.

25. Dominant alleles are not necessarily more common than recessive alleles in the gene pool. Explain why this is true.

26. Explain what is meant when a gene is said to have *multiple alleles*. Blood groups are an excellent human example of this.

27. Blood groups are so important medically that you should be able to solve genetics problems based on blood types. The first step in accomplishing that is to understand the genotypes of each blood type. Before working any problems, complete this ABO blood type chart. Begin with a key.

KEY:

Genotype	Red Blood Cell Appearance	Phenotype (blood group)

28. Question 2 in *Concept Check 14.3* is a blood type problem. Complete it here, and show your work.

29. What is *pleiotropy*? Explain how this is significant in cystic fibrosis and sickle-cell disease.

30. Explain *epistasis*.

31. Explain why the dihybrid cross, detailed in Figure 14.12 in your text, has four yellow Labrador retrievers instead of the three that would have been predicted by Mendel's work.

32. *Quantitative variation* usually indicates *polygenic inheritance*. What is a good example of this?

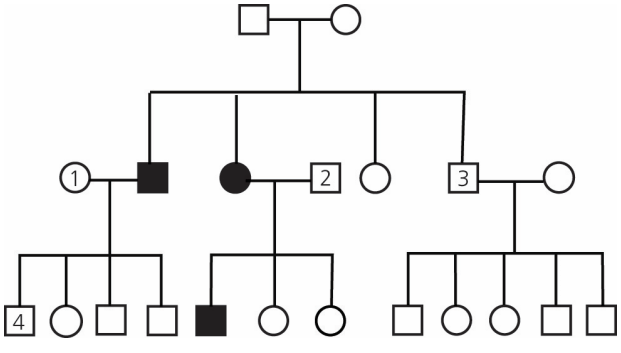
33. Explain the potential influence of the environment on phenotypic expression and cite an example.

Concept 14.4 Many human traits follow Mendelian patterns of inheritance

LO 14.4: Explain what a pedigree is and use examples to show how human traits follow Mendelian patterns of inheritance.

34. Pedigree analysis is often used to determine mode of inheritance (dominant or recessive, for example). Be sure to read the "Tips for Pedigree Analysis" in Figure 14.15 in your text.

    Consider the following pedigree for the trait albinism (lack of skin pigmentation) in three generations of a family. (Solid symbols represent individuals with albinism.) Complete the unlabeled pedigree by indicating the genotypes for all involved.



    From your knowledge of Mendelian inheritance, answer the questions that follow.

    a. Is this trait caused by a dominant or recessive allele? What is the evidence for your response?

b. Determine the genotypes of the parents in the first generation. (Let ***A*** and ***a*** represent the alleles.)

Genotype of father _____; genotype of mother _____

What is the evidence for your response?

c. Determine the probable genotypes of the mates of the offspring with albinism in the second generation and grandson #4 in the third generation.

Genotypes: mate #1 _____; mate #2 _____; grandson #4 _____.

d. Can you determine the genotype of son #3 in the second generation? Why or why not?

35. Describe what you think is medically important to know about the behavior of recessive alleles.

36. For the following human inherited conditions, describe the condition and give the pattern of inheritance.

a. **albinism**

b. **Tay-Sachs disease**

c. **cystic fibrosis**

d. **sickle-cell disease**

e. **achondroplasia**

f. **Huntington's disease**

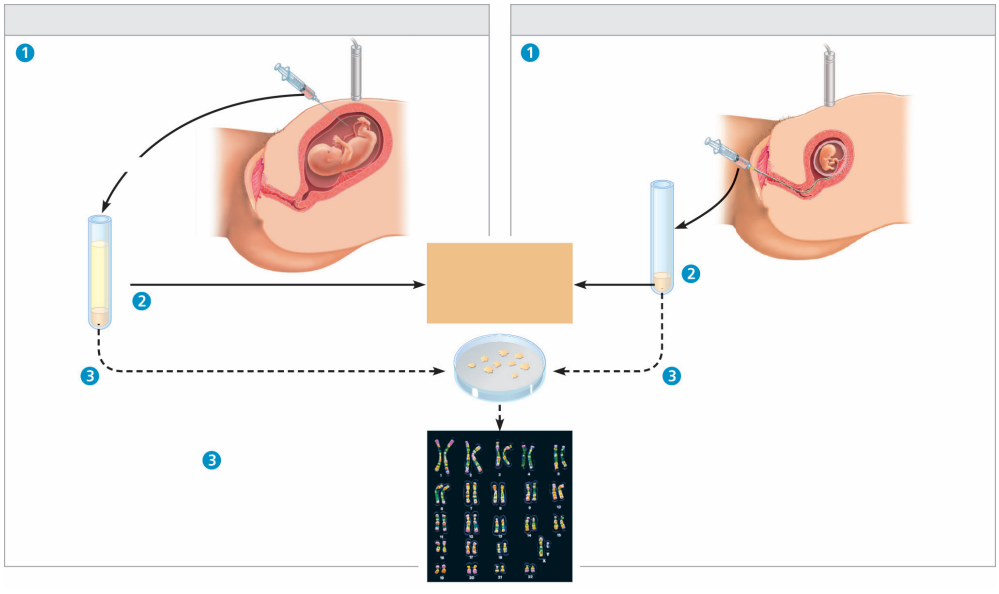
37. There are tests to identify carriers that allow couples with a family history of a disorder to make informed decisions about having children. Genetic counseling for these families is based on Mendelian genetics and probability rules. Work through the example of hypothetical couple Tyler and Lily on p. 288 of your text to understand the mathematics involved. Then, solve the following problems.

    Lucia and Jared each have a sibling with cystic fibrosis; neither Lucia or Jared nor any of their parents have the disease.

    a. Calculate the probability that a child born to this couple will have cystic fibrosis. Show your work and explain your answer.

    b. What would be the probability if a test revealed that Jared is a carrier, but Lucia is not? Show your work and explain your answer.

36. There are a number of tests that can be done to look for genetic disorders in a fetus. Imaging using *ultrasound* and analysis of fetal DNA that has escaped into the mother's blood are two noninvasive procedures. *Amniocentesis* and *chorionic villus sampling (CVS)* are two widely used methods for testing a fetus for genetic disorders. Use the following unlabeled diagram to explain the three main steps in amniocentesis and the three main steps of CVS.



Steps in amniocentesis	Steps in chorionic villus sampling

37. What are the strengths and weaknesses of each fetal test?

38. What are the symptoms of *phenylketonuria (PKU)*? How is newborn screening used to identify children with this disorder? Why is newborn screening important?

Test Your Understanding, p. 292

One of the ways to determine your understanding of Mendelian genetics is to work many genetics problems. Complete questions 1–14. Work neatly and show all work. As you know, you can check your solutions in Appendix A of your text.