Chapter 11  Introduction to Genetics

Summary

11–1 The Work of Gregor Mendel

The scientific study of heredity is called genetics. Gregor Mendel used purebred pea plants in a series of experiments to understand inheritance.

Pea flowers have both male and female parts. Normally, pollen from the male part of the pea flower fertilizes the female egg cells of the same flower. This is called self-pollination. Seeds that come from self-pollination inherit all their characteristics from just one parent.

To carry out his experiments, Mendel had to prevent self-pollination. He did this by cutting away the pollen-bearing male parts and then dusting pollen from another plant on the flower. This process is called cross-pollination. The seeds that come from cross-pollination are the offspring of two different parents.

Mendel decided to study just a few traits, or characteristics, of the pea plants. He studied seven traits: seed shape, seed color, seed coat color, pod shape, pod color, flower position, and plant height.

First, Mendel crossed two plants with different characters, or forms, for the same trait. For example, one plant was tall and the other was short. Mendel used the seeds produced by this cross to grow plants. These plants were hybrids. Hybrids are the offspring of crosses between parents with different traits.

To Mendel’s surprise, the hybrid plants looked like only one of the parents. He concluded that each trait was controlled by one gene that occurred in two different forms. The different forms of a gene are called alleles. Mendel formed the theory of dominance. He concluded that some alleles are dominant, while others are recessive. Whenever a living thing inherits a dominant allele, that trait is visible. The effects of a recessive allele are not seen if the dominant allele is present.

Mendel wanted to know what happened to the recessive allele. He allowed his hybrid plants to self-pollinate. Some of the plants that were produced showed the recessive trait. The alleles responsible for the recessive characters had not disappeared. Before, the dominant allele had masked the recessive allele, so it was not visible. Mendel concluded that the alleles for the same trait can be separated. He called this segregation. Alleles segregate when gametes are formed. Each gamete carries only one copy of each gene.

11–2 Probability and Punnett Squares

Mendel used the principles of probability to explain his results. Probability is the likelihood that a particular event will occur. Probability can be used to predict the outcome of genetic crosses because alleles segregate randomly. The gene combinations that might result from a genetic cross can be determined by drawing a Punnett square.

In a Punnett square, alleles are represented by letters. A capital letter represents the dominant allele, and a lowercase letter represents the recessive allele. Organisms that have two identical alleles for a particular trait are called homozygous. Homozygous organisms are true-breeding for a particular trait. Organisms that have two different alleles for a particular trait are called heterozygous. Heterozygous organisms are hybrid for a particular trait.

The physical traits of an organism make up its phenotype (for example, height). The genetic makeup of an organism is its genotype (for example, TT or Tt).
One important rule of probability is that probabilities predict the average outcome of a large number of events. They cannot predict what will happen in a single event. The more organisms examined, the closer the numbers will get to the expected values.

11–3 Exploring Mendelian Genetics

Mendel wondered whether genes that determine one trait have anything to do with genes that determine another trait. He wanted to know, for example, whether the gene that determines seed shape affects the gene for seed color. To answer this question, he did an experiment. He crossed plants and recorded two traits—seed shape and seed color.

Mendel found that the gene controlling seed shape did not affect the gene controlling seed color. Mendel concluded that genes can segregate independently, or undergo independent assortment, during gamete formation.

11–4 Meiosis

According to Mendel, living things inherit a single copy of each gene from each of their parents. When gametes are formed, these two copies are separated.

Gametes are made during meiosis. In a complex process, the number of chromosomes in each cell is cut in half. The chromosomes are different from one another and from the parent cell.

There are two stages in meiosis. During the first stage, the DNA in special cells in the reproductive organs is copied. The cells then divide. Two cells are formed. These cells are different from each other and different from the parent cell. In the second stage of meiosis, the cells divide again. This time, their DNA is not copied first. Four daughter cells are produced. Each cell contains half the number of chromosomes of the original parent cell.

In male animals, the gametes produced by meiosis are called sperm. Some plants also have sperm cells. In females, meiosis produces one large reproductive cell and three smaller cells. In animals, the larger reproductive cell is called an egg. In some plants, it is called an egg cell. The three smaller cells produced during meiosis are called polar bodies. They do not participate in reproduction.

Meiosis is very different from mitosis. Mitosis makes two cells that are exactly alike. The cells are also exactly like the parent cell. Meiosis, however, produces four cells. Each of the cells has only half the number of chromosomes of the parent cell. The cells are genetically different from one another.

11–5 Linkage and Gene Maps

Some genes are almost always inherited together. These genes belong to the same linkage group. A chromosome is a group of linked genes. It is actually the chromosomes that assort independently during gamete formation, not single genes.

The location of genes can be mapped to a chromosome. The rate of crossover events is used to find the distance between genes on a chromosome. The farther apart two genes are, the more likely they will be separated by a crossover event.
Section 11–1 The Work of Gregor Mendel
(pages 263–266)

Key Concepts
• What is the principle of dominance?
• What happens during segregation?

Gregor Mendel’s Peas (pages 263–264)
1. The scientific study of heredity is called ___________________________.

2. Circle the letter of each sentence that is true about Gregor Mendel’s peas.
   a. The male parts of pea flowers produce eggs.
   b. When pollen fertilizes an egg cell, a seed for a new plant is formed.
   c. Pea plants normally reproduce by self-pollination.
   d. Seeds that are produced by self-pollination inherit their characteristics from two different plants.

3. What does it mean when pea plants are described as being true-breeding?

4. To perform his experiments, how did Mendel prevent pea flowers from self-pollinating and control their cross-pollination?

Genes and Dominance (pages 264–265)

Match the term with its definition.

<table>
<thead>
<tr>
<th>Terms</th>
<th>Definitions</th>
</tr>
</thead>
<tbody>
<tr>
<td>5. genes</td>
<td>a. Specific characteristics that vary from one individual to another</td>
</tr>
<tr>
<td>6. hybrids</td>
<td>b. The offspring of crosses between parents with different traits</td>
</tr>
<tr>
<td>7. traits</td>
<td>c. Chemical factors that determine traits</td>
</tr>
<tr>
<td>8. alleles</td>
<td>d. The different forms of a gene</td>
</tr>
</tbody>
</table>

9. State the principle of dominance. ___________________________________________

10. Is the following sentence true or false? An organism with a recessive allele for a particular form of a trait will always exhibit that form. ______________________

11. Circle the letters of the traits controlled by dominant alleles in Mendel’s pea plants.
    a. tall           b. short       c. yellow       d. green
12. How did Mendel find out whether the recessive alleles were still present in the F1 plants?

13. About one fourth of the F2 plants from Mendel’s F1 crosses showed the trait controlled by the _____________ allele.

14. Circle the letter of each sentence that is true about Mendel’s explanation of the results from his F1 cross.
   a. Mendel assumed that a dominant allele had masked the corresponding recessive allele in the F1 generation.
   b. The trait controlled by the recessive allele never showed up in any F2 plants.
   c. The allele for shortness was always inherited with the allele for tallness.
   d. At some point, the allele for shortness was segregated, or separated, from the allele for tallness.

15. What are gametes? __________________________________________________________________________

16. Complete the following diagram to show how alleles segregate during the formation of gametes.

17. In the diagram above, the dominant allele is represented by ______ and the recessive allele is represented by ______.
Section 11–2 Probability and Punnett Squares  
(pages 267–269)

Key Concepts
• How do geneticists use the principles of probability?
• How do geneticists use Punnett squares?

Genetics and Probability  (page 267)
1. The likelihood that a particular event will occur is called ________________.
2. Circle the letter of the probability that a single coin flip will come up heads.
   a. 100 percent  b. 75 percent  c. 50 percent  d. 25 percent
3. Is the following sentence true or false? The past outcomes of coin flips greatly affect the outcomes of future coin flips. ________________
4. Why can the principles of probability be used to predict the outcomes of genetic crosses? ________________

Punnett Squares  (page 268)
5. How do geneticists use Punnett squares? ________________

6. Complete the Punnett square to show the possible gene combinations for the F₁ offspring.

PUNNETT SQUARE FOR Tt × Tt

<table>
<thead>
<tr>
<th></th>
<th>T</th>
<th>t</th>
</tr>
</thead>
<tbody>
<tr>
<td>T</td>
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<tr>
<td>t</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Match the terms with the definitions.

Terms
_____ 7. genotype
_____ 8. homozygous
_____ 9. phenotype
_____ 10. heterozygous

Definitions
a. Organisms that have two identical alleles for a particular trait (TT or tt)
b. Organisms that have two different alleles for the same trait (Tt)
c. Physical characteristic of an organism (tall)
d. Genetic makeup of an organism (Tt)

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11. Is the following sentence true or false? Homozygous organisms are true-breeding for a particular trait. ________________

12. Is the following sentence true or false? Plants with the same phenotype always have the same genotype. ________________

**Probability and Segregation (page 269)**

13. Circle the letter of each sentence that is true about probability and segregation.
   a. In an F₁ cross between two hybrid tall pea plants (Tt), ½ of the F₂ plants will have two alleles for tallness (TT).
   b. The F₂ ratio of tall plants to short plants produced in a cross between two hybrid tall pea plants (Tt) is 3 tall plants for every 1 short plant.
   c. Mendel observed that about ¾ of the F₂ offspring showed the dominant trait.
   d. Segregation occurs according to Mendel’s model.

14. In Mendel’s model of segregation, what was the ratio of tall plants to short plants in the F₂ generation? ________________

**Probabilities Predict Averages (page 269)**

15. Is the following sentence true or false? Probabilities predict the precise outcome of an individual event. ________________

16. How can you be sure of getting the expected 50 : 50 ratio from flipping a coin? ________________

17. The ________________, the number of offspring from a genetic cross, the closer the resulting numbers will get to expected values.

18. Is the following sentence true or false? The ratios of an F₁ generation are more likely to match Mendelian predicted ratios if the F₁ generation contains hundreds or thousands of individuals. ________________

**Reading Skill Practice**

Taking notes helps the reader focus on the main ideas and the vocabulary of the reading. Take notes while rereading Section 11–2. Note the main ideas and the highlighted, boldface terms in the order in which they are presented. You may copy the ideas word for word or summarize them using your own words. Do your work on a separate sheet of paper.
Section 11–3 Exploring Mendelian Genetics
(pages 270–274)

Key Concepts
• What is the principle of independent assortment?
• What inheritance patterns exist aside from simple dominance?

Independent Assortment (pages 270–271)
1. In a two-factor cross, Mendel followed ________________ different genes as they passed from one generation to the next.

2. Write the genotypes of the true-breeding plants that Mendel used in his two-factor cross.

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>Genotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>a. round yellow peas</td>
<td>________________</td>
</tr>
<tr>
<td>b. wrinkled green peas</td>
<td>________________</td>
</tr>
</tbody>
</table>

3. Circle the letter that best describes the F1 offspring of Mendel’s two-factor cross.
   a. Homozygous dominant with round yellow peas
   b. Homozygous recessive with wrinkled green peas
   c. Heterozygous dominant with round yellow peas
   d. Heterozygous recessive with wrinkled green peas

4. Is the following sentence true or false? The genotypes of the F1 offspring indicated to Mendel that genes assort independently.
   ________________

5. How did Mendel produce the F2 offspring? ________________________________

6. Circle the letter of the phenotypes that Mendel would expect to see if genes segregated independently.
   a. round and yellow
   b. wrinkled and green
   c. round and green
   d. wrinkled and yellow

7. What did Mendel observe in the F2 offspring that showed him that the alleles for seed shape segregate independently of those for seed color? ________________________________

8. What were the phenotypes of the F2 generation that Mendel observed? ________________
9. What was the ratio of Mendel’s F₂ generation for the two-factor cross?  

10. Complete the Punnett square below to show the predicted results of Mendel’s two-factor cross.

**MENDEL’S TWO-FACTOR CROSS**  
*RrYy × RrYy*

<table>
<thead>
<tr>
<th></th>
<th><strong>RY</strong></th>
<th><strong>Ry</strong></th>
<th><strong>rY</strong></th>
<th><strong>ry</strong></th>
</tr>
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<tbody>
<tr>
<td><strong>RY</strong></td>
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<tr>
<td><strong>Ry</strong></td>
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<tr>
<td><strong>ry</strong></td>
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</tr>
</tbody>
</table>

11. State Mendel’s principle of independent assortment.

**A Summary of Mendel’s Principles**  
(page 272)

12. Circle the letter of each sentence that is true about Mendel’s principles.

   a. The inheritance of biological characteristics is determined by genes that are passed from parents to their offspring.
   b. Two or more forms of the gene for a single trait can never exist.
   c. The copies of genes are segregated from each other when gametes are formed.
   d. The alleles for different genes usually segregate independently of one another.

13. When two or more forms of the gene for a single trait exist, some forms of the gene may be _______________ and others may be _______________.

**Beyond Dominant and Recessive Alleles**  
(pages 272–273)

14. Is the following sentence true or false? All genes show simple patterns of dominant and recessive alleles. _______________
15. Complete the table of the different patterns of inheritance.

### PATTERNS OF INHERITANCE

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>One allele is not completely dominant over another. The heterozygous phenotype is somewhere in between the two homozygous phenotypes.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Both alleles contribute to the phenotype of the organism.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Genes have more than two alleles.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Two or more genes control a trait.</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Applying Mendel’s Principles** *(page 274)*

16. List three criteria Thomas Hunt Morgan was looking for in a model organism for genetic studies.
   
   a.  
   b.  
   c.  

17. Is the following sentence true or false? Mendel’s principles apply not just to pea plants but to other organisms as well. 

**Genetics and the Environment** *(page 274)*

18. Characteristics are determined by interaction between genes and the
Section 11–4 Meiosis  (pages 275–278)

Key Concepts
• What happens during the process of meiosis?
• How is meiosis different from mitosis?

Introduction  (page 275)
1. List the two things that Mendel’s principles of genetics required in order to be true.
   a. ________________________________________________________________
   ________________________________________________________________
   b. ________________________________________________________________
   ________________________________________________________________

Chromosome Number  (page 275)
2. What does it mean when two sets of chromosomes are homologous? _____________
   ________________________________________________________________
   ________________________________________________________________

3. Circle the letter of each way to describe a diploid cell.
   a. 2N
   b. Contains two sets of homologous chromosomes
   c. Contains a single set of homologous chromosomes
   d. A gamete

4. Circle the letter of the number of chromosomes in a haploid Drosophila cell.
   a. 8   b. 4   c. 2   d. 0

Phases of Meiosis  (pages 275–277)
5. Draw the chromosomes in the diagrams below to show the correct phase of meiosis.

   ![Prophase I](image1.png)  ![Metaphase I](image2.png)  ![Anaphase II](image3.png)
6. Identify which phase of meiosis is shown in the diagrams below.

7. Why is meiosis described as a process of reduction division?

8. What are the two distinct divisions of meiosis?
   a. 
   b. 

9. Is the following sentence true or false? The diploid cell that enters meiosis becomes 4 haploid cells at the end of meiosis.

10. How does a tetrad form in prophase I of meiosis?

11. Circle the number of chromatids in a tetrad.
   a. 8  b. 6  c. 4  d. 2

12. What results from the process of crossing-over during prophase I?
13. Circle the letter of each sentence that is true about meiosis.
   a. During meiosis I, homologous chromosomes separate.
   b. The two daughter cells produced by meiosis I still have the two complete sets of
      chromosomes, as does a diploid cell.
   c. During anaphase II, the paired chromatids separate.
   d. After meiosis II, the four daughter cells contain the diploid number of chromosomes.

**Gamete Formation (page 278)**

Match the products of meiosis with the descriptions.

<table>
<thead>
<tr>
<th>Product of Meiosis</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>14. eggs</td>
<td>a. Haploid gametes produced in males</td>
</tr>
<tr>
<td>15. sperm</td>
<td>b. Haploid gametes produced in females</td>
</tr>
<tr>
<td>16. polar bodies</td>
<td>c. Cells produced in females that do not</td>
</tr>
<tr>
<td></td>
<td>participate in reproduction</td>
</tr>
</tbody>
</table>

**Comparing Mitosis and Meiosis (page 278)**

17. Circle the letter of each sentence that is true about mitosis and meiosis.
   a. Mitosis produces four genetically different haploid cells.
   b. Meiosis produces two genetically identical diploid cells.
   c. Mitosis begins with a diploid cell.
   d. Meiosis begins with a diploid cell.

**Reading Skill Practice**

You can often increase your understanding of what you’ve read by making comparisons. A compare-and-contrast table helps you to do this. On a separate sheet of paper, make a table to compare the processes of mitosis and meiosis. For more information about compare-and-contrast tables, see Organizing Information in Appendix A in your textbook.
Section 11–5 Linkage and Gene Maps
(pages 279–280)

Key Concept
• What structures actually assort independently?

Gene Linkage (page 279)
1. Is the following sentence true or false? Thomas Hunt Morgan discovered that some genes violated the principle of independent assortment. ________________
2. Morgan grouped the Drosophila genes that were inherited together into four ________________ groups.
3. List the two conclusions that Morgan made about genes and chromosomes.
   a. ________________________________
   ________________________________
   b. ________________________________
   ________________________________
4. Why didn’t Mendel observe gene linkage? ________________________________
   ________________________________
   ________________________________
   ________________________________
   ________________________________

Gene Maps (pages 279–280)
5. Explain why two genes found on the same chromosome are not always linked forever.
   ________________________________
   ________________________________
   ________________________________
   ________________________________
6. The new combinations of alleles produced by crossover events help to generate genetic ________________.
7. Is the following sentence true or false? Genes that are closer together are more likely to be separated by a crossover event in meiosis. ________________
8. What is a gene map? ________________________________
   ________________________________
   ________________________________
   ________________________________
9. How is a gene map constructed? ________________________________
   ________________________________
   ________________________________
   ________________________________
Chapter 11  Introduction to Genetics

Vocabulary Review

Labeling Diagrams  Use the words listed below to label the Punnett square. Some words may be used twice.

- heterozygous parent
- dominant allele
- recessive allele
- homozygous offspring
- heterozygous offspring

Matching  In the space provided, write the letter of the definition that best matches each term.

- 7. phenotype  a. likelihood that something will happen
- 8. gamete  b. shows the relative locations of genes on a chromosome
- 9. genetics  c. physical characteristics of an organism
- 10. probability  d. containing one set of chromosomes
- 11. haploid  e. sex cell
- 12. gene map  f. chemical factor that determines traits
- 13. gene  g. specific characteristic
- 14. multiple alleles  h. scientific study of heredity
- 15. trait  i. gene with more than two alleles

Completion  Fill in the blanks with terms from Chapter 11.

16. The process in which two genes segregate independently is called ____________________________.
17. Plants that, if left to self-pollinate, produce offspring identical to themselves are called ____________________________.
18. The offspring of crosses between parents with different traits are called ____________________________.
19. The process during sexual reproduction in which male and female sex cells join is called ____________________________.
20. The process of reduction division in which the number of chromosomes per cell is cut in half is called ____________________________.
Chapter 14  The Human Genome

Summary

14–1 Human Heredity
Biologists can analyze human chromosomes by looking at a karyotype. A karyotype is a picture of the chromosomes from a cell arranged in homologous pairs.

Humans have 46 chromosomes. Two of these chromosomes, X and Y, are the sex chromosomes. Females have two X chromosomes (XX). Males have one X and one Y chromosome (XY). The other 44 chromosomes are called autosomes.

Human genes are inherited according to the same principles of genetics described by Mendel. To study the inheritance of human traits, biologists use a pedigree chart. A pedigree shows the relationships within a family. The inheritance of a certain trait in a family can be traced using a pedigree. From this, biologists can infer the genotypes of family members.

It is difficult to associate an observed human trait with a specific gene. Many human traits are polygenic, meaning that they are controlled by many genes. The environment also influences many traits.

Some of the first human genes to be identified were those that control blood type. Red blood cells can carry two different antigens, called A and B. Antigens are molecules that can be recognized by the immune system. The presence or absence of the A and B antigens produces four possible blood types: A, B, AB, and O. The ABO blood types are determined by a single gene with three alleles.

In addition to the ABO antigens, there is another antigen on red blood cells called the Rh antigen. People who have the Rh antigen are Rh positive. People without it are Rh negative. A single gene with two alleles determines the Rh blood group.

There are several human genetic disorders, including phenylketonuria (PKU), Huntington disease, and sickle cell disease. PKU is caused by a recessive allele. It is expressed only in individuals who have inherited a recessive allele from each parent. Huntington disease is caused by a dominant allele. It is expressed in any person who has that allele. Sickle cell disease is caused by a codominant allele.

Scientists are beginning to understand which changes in the DNA sequence cause certain genetic disorders. Cystic fibrosis is caused by the deletion of three bases in the middle of the sequence for a protein. This deletion inactivates the protein, which causes the symptoms of this disorder. Only one DNA base is changed in the allele that causes sickle cell disease. This base change produces a blood protein that is less soluble than normal.

14–2 Human Chromosomes
The two smallest human chromosomes, chromosomes 21 and 22, were the first chromosomes to have their DNA sequences identified. Both have many genes important for health. Both have regions of DNA that do not code for proteins.

Genes located on the X and Y chromosomes, the sex chromosomes, are said to be sex-linked. They are inherited in a different pattern than genes located on autosomes. For example, all alleles linked to the X chromosome, including those responsible for colorblindness, hemophilia, and Duchenne muscular dystrophy, are expressed in males even if they are recessive alleles. However, in order for these recessive alleles to be expressed in females, there must be two copies of them.
Females have two X chromosomes. Males have only one. To account for this difference, one X chromosome in females is randomly turned off. The turned-off chromosome forms a dense region in the nucleus known as a Barr body. Barr bodies are not found in males because their single X chromosome must be active.

The most common error during meiosis is nondisjunction. Nondisjunction is the failure of chromosomes to separate properly during meiosis. It causes abnormal numbers of chromosomes to find their way into gametes. This may result in a disorder of chromosome number. An example of autosomal nondisjunction is Down syndrome, in which there is an extra copy of chromosome 21. Nondisjunction can also occur in sex chromosomes. In Turner’s syndrome, a female has only one X chromosome. In Klinefelter’s syndrome, there are extra X chromosomes.

14–3 Human Molecular Genetics

Biologists can use techniques in molecular biology to read, analyze, and even change the DNA code of human genes. Genetic tests are available to test parents for the presence of recessive alleles for genetic disorders.

In a process called DNA fingerprinting, individuals can be identified by analyzing sections of DNA that have little or no known function. These sections of DNA vary widely from one person to the next.

In 1990, scientists around the world began the Human Genome Project. The goal was to identify the DNA sequence for the entire DNA in a human cell. In 2000, the human genome was sequenced. Now the project goal is to analyze these sequences. One way scientists are analyzing the DNA is by looking for genes. To do this, they look for promoter sequences. These are sequences that bind RNA polymerase.

Information about the human genome can be used to cure genetic disorders by gene therapy. In one method of gene therapy, a virus is used to deliver the normal gene into cells to correct the genetic defects. The virus is changed so that it cannot cause disease. The normal gene is attached to the DNA of the virus. The inserted gene can make proteins that correct the genetic defect.

There are risks and problems with gene therapy. Having the power to manipulate human DNA doesn’t necessarily make it right. People in a society are responsible for making sure that the tools made available by science are used wisely.
Section 14–1 Human Heredity (pages 341–348)

Key Concepts
• How is sex determined?
• How do small changes in DNA cause genetic disorders?

Human Chromosomes (pages 341–342)
1. How do biologists make a karyotype?

2. Circle the letter of each sentence that is true about human chromosomes.
   a. The X and Y chromosomes are known as sex chromosomes because they determine an individual’s sex.
   b. Males have two X chromosomes.
   c. All the chromosomes except the sex chromosomes are autosomes.
   d. Biologists would write 46,XY to indicate a human female.

3. Complete the Punnett square below to show how the sex chromosomes segregate during meiosis.

4. Why is there the chance that half of the zygotes will be female and half will be male?
**Human Traits** (pages 342–343)

5. What does a pedigree chart show?

6. A person who expresses the trait

7. A male

8. A person who does not express the trait

9. Represents a marriage

10. A female

11. Connects parents to their children

12. Give two reasons why it is impossible to associate some of the most obvious human traits with single genes.

   a. 

   b. 

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**Human Genes** *(pages 344–346)*

13. Why is it difficult to study the genetics of humans?

14. Circle the letter of each sentence that is true about human blood group genes.
   a. The Rh blood group is determined by a single gene.
   b. The negative allele (Rh⁻) is the dominant allele.
   c. All of the alleles for the ABO blood group gene are codominant.
   d. Individuals with type O blood are homozygous for the i allele (ii) and produce no antigen on the surface of red blood cells.

15. Is the following sentence true or false? Many human genes have become known through the study of genetic disorders.

**Match the genetic disorder with its description.**

<table>
<thead>
<tr>
<th>Genetic Disorder</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>16. Phenylketonuria (PKU)</td>
<td>a. Nervous system breakdown caused by an autosomal recessive allele</td>
</tr>
<tr>
<td>17. Tay-Sachs disease</td>
<td>b. A form of dwarfism caused by an autosomal dominant allele</td>
</tr>
<tr>
<td>18. Achondroplasia</td>
<td>c. A buildup of phenylalanine caused by an autosomal recessive allele</td>
</tr>
<tr>
<td>19. Huntington disease</td>
<td>d. A progressive loss of muscle control and mental function caused by an autosomal dominant allele</td>
</tr>
</tbody>
</table>

**From Gene to Molecule** *(pages 346–348)*

20. What is the normal function of the protein that is affected in cystic fibrosis?

21. A change in just one DNA base for the gene that codes for the protein causes sickle-shaped red blood cells.

22. What is the advantage of being heterozygous for the sickle cell allele?

23. What makes an allele dominant, recessive, or codominant?
Section 14–2 Human Chromosomes  (pages 349–353)

Key Concepts
- Why are sex-linked disorders more common in males than in females?
- What is nondisjunction, and what problems does it cause?

Human Genes and Chromosomes  (page 349)
1. Circle the letter of each sentence that is true about human genes and chromosomes.
   a. Chromosomes 21 and 22 are the largest human chromosomes.
   b. Chromosome 22 contains long stretches of repetitive DNA that do not code for proteins.
   c. Biologists know everything about how the arrangements of genes on chromosomes affect gene expression.
   d. Human genes located close together on the same chromosome tend to be inherited together.

Sex-Linked Genes  (pages 350–351)
2. What are sex-linked genes? ____________________________________________________________________

3. Is the following sentence true or false? The Y chromosome does not contain any genes at all. __________

4. Complete the table describing sex-linked disorders.

<table>
<thead>
<tr>
<th>SEX-LINKED DISORDERS IN HUMANS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Disorder</strong></td>
</tr>
<tr>
<td>----------------</td>
</tr>
<tr>
<td>Colorblindness</td>
</tr>
<tr>
<td></td>
</tr>
<tr>
<td></td>
</tr>
</tbody>
</table>

5. Is the following sentence true or false? All X-linked alleles are expressed in males, even if they are recessive. ________________
6. Complete the Punnett square to show how colorblindness is inherited.

![Punnett square](image)

**X-Chromosome Inactivation (page 352)**

7. How does the cell “adjust” to the extra X chromosome in female cells? __________

8. What is a Barr body? ________________

9. Is the following sentence true or false? Barr bodies are found only in males. __________

10. If you see a white cat with orange and black spots, is it most likely a male or a female? Explain. __________

**Chromosomal Disorders (pages 352–353)**

11. What occurs during nondisjunction? ____________________________

12. Is the following sentence true or false? If nondisjunction occurs, gametes may have abnormal numbers of chromosomes. __________

13. The condition in which an individual has three copies of a chromosome is known as ____________________, which means “three bodies.”
14. Is the following sentence true or false? Down syndrome occurs when an individual has two copies of chromosome 21. ________________

15. Circle the letter of the characteristic of Down syndrome.
   a. dwarfism          c. colorblindness
   b. mental retardation d. muscle loss

16. Why does an extra copy of one chromosome cause so much trouble? ________________

17. Circle the letter of each sentence that is true about sex chromosome disorders.
   a. A female with the karyotype 45,X has inherited only one X chromosome and is sterile.
   b. Females with the karyotype 47,XXY have Klinefelter’s syndrome.
   c. Babies have been born without an X chromosome.
   d. The Y chromosome contains a sex-determining region that is necessary for male sexual development.

**Reading Skill Practice**

Writing an outline is a useful way to organize the important facts in a section. Write an outline of Section 14–2. Use the section headings as the headings in your outline. Include only the important facts and main ideas in your outline. Be sure to include the vocabulary terms. Do your work on a separate sheet of paper.
Section 14–3 Human Molecular Genetics
(pages 355–360)

Key Concepts

• What is the goal of the Human Genome Project?
• What is gene therapy?

Human DNA Analysis (pages 355–357)

1. Biologists search the volumes of the human genome using __________________________.

2. Why might prospective parents decide to have genetic testing? __________________________

3. Circle the letter of each sentence that is true about genetic testing.
   a. It is impossible to test parents to find out if they are carriers for cystic fibrosis or Tay-Sachs disease.
   b. Labeled DNA probes can be used to detect specific sequences found in disease-causing alleles.
   c. Some genetic tests use changes in restriction enzyme cutting sites to identify disease-causing alleles.
   d. DNA testing makes it possible to develop more effective therapy and treatment for individuals affected by genetic disease.

4. What is DNA fingerprinting? __________________________

5. Complete the flowchart to show the steps in DNA fingerprinting.

   Small sample of DNA is cut with a(an) __________________________ enzyme.

   The fragments are separated by size using __________________________.

   Fragments with highly variable regions are detected with a(an) __________________________, revealing a series of DNA bands of various sizes.

   The pattern of bands produced is the __________________________, which can be distinguished statistically from the pattern of any other individual in the world.
6. Circle the letter of each source for a DNA sample from an individual.
   a. blood c. clothing
   b. sperm d. hair with tissue at the base

7. Is the following sentence true or false? DNA evidence is not reliable enough to be used to convict criminals. _________________

**The Human Genome Project (pages 357–358)**

8. What is the Human Genome Project? ____________________________

9. Circle the letter of each sentence that is true about the Human Genome Project.
   a. The human genome is the first genome entirely sequenced.
   b. The human genome is about the same size as the genome of *E. coli*.
   c. Researchers completed the genomes of yeast and fruit flies during the same time they sequenced the human genome.
   d. A working copy of the human genome was completed in June 2000.

10. What were the three major steps in the process of sequencing the human genome?
    a. ____________________________________________
    b. ____________________________________________
    c. ____________________________________________

11. What is an open reading frame, and what is it used for? ____________________________

12. The mRNA coding regions of most genes are interrupted by ____________________________

13. List three other parts of the gene that researchers look for.
    a. ____________________________________________
    b. ____________________________________________
    c. ____________________________________________

14. Why are biotechnology companies interested in genetic information? ____________________________
15. Is the following sentence true or false? Human genome data are top secret and can be accessed only by certain people. ________________

**Gene Therapy (pages 359–360)**

16. What is gene therapy? ____________________________________________________________

17. Circle the letter of each sentence that is true about gene therapy.
   a. When the normal copy of the gene is inserted, the body can make the correct protein, which eliminates the disorder.
   b. So far, no one has been successfully cured of a genetic disorder using gene therapy.
   c. Viruses are often used to carry the normal genes into cells.
   d. Viruses used in gene therapy often cause disease in the patients.

18. Is the following sentence true or false? All gene therapy experiments have been successful. ________________

**Ethical Issues in Human Genetics (page 360)**

19. What other changes could be made to the human genome by manipulating human cells? ____________________________________________________________

20. What is the responsibility of society in biology? ________________________________

21. Is the following true or false? Scientists should be expected to make all ethical decisions regarding advances in human genetics. ________________
Chapter 14     The Human Genome

Vocabulary Review

Labeling Diagrams     Use the words listed below to label the diagram.

- autosome
- sex chromosome
- karyotype

Matching     In the space provided, write the letter of the definition that best matches each term.

4. karyotype     a. chart that shows the relationships within a family
5. sex chromosomes     b. failure of homologous chromosomes to separate in meiosis
6. autosomes     c. picture of chromosomes arranged in pairs
7. pedigree     d. test used to identify individuals by analyzing sections of DNA
8. sex-linked gene     e. chromosomes that determine an individual’s sex
9. nondisjunction     f. gene located on the X or Y chromosome
10. DNA fingerprinting     g. chromosomes that do not determine sex