

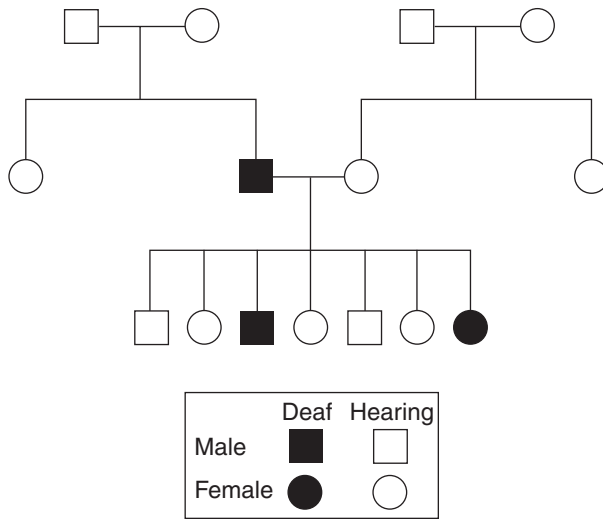
Chapter 14 The Human Genome

Chapter Vocabulary Review

Completion *On the lines provided, complete the following sentences with one of the following terms:*

autosomes karyotype pedigree nondisjunction sex chromosomes

1. A picture of chromosomes arranged in pairs is an example of a (an) _____.
2. The X and Y chromosomes are called the _____.
3. The 44 chromosomes that are not involved in sex determination are the known as the _____.
4. The chart below, showing how a trait is passed from one generation to the next in a family, is called a (an) _____.



5. Down syndrome is caused by _____.

Multiple Choice *On the lines provided, write the letter of the answer that best completes the sentence or answers the question.*

- _____ 6. Genes located on X or Y chromosomes are called
 - a. sex organs.
 - b. autosomes.
 - c. a pedigree.
 - d. sex-linked.
- _____ 7. One example of a sex-linked disorder is
 - a. sickle cell disease.
 - b. hemophilia.
 - c. Down syndrome.
 - d. Tay-Sachs disease.
- _____ 8. Nondisjunction results in a(an)
 - a. inactivated chromosome.
 - b. abnormal number of chromosomes.
 - c. normal number of genes.
 - d. normal number of chromosomes.

- _____ 9. What happens when nondisjunction occurs?
- a. Chromosomes fail to separate.
 - b. Chromosomes separate normally.
 - c. Sections of chromosomes are deleted.
 - d. Sections of chromosomes become inverted.
- _____ 10. How is DNA fingerprinting commonly used?
- a. to convict criminals or overturn convictions
 - b. to sequence all human DNA
 - c. to rapidly sequence DNA with computers
 - d. to identify and locate human genes
- _____ 11. DNA samples for human DNA fingerprinting can be obtained from
- a. sperm.
 - b. blood.
 - c. hair with attached tissue.
 - d. all of the above.

Short Answer *On the lines provided, answer the following questions.*

12. What type of picture would a biologist look at to determine whether a fetus has Down syndrome?

13. Why are the X and Y chromosomes called sex chromosomes?

14. How many autosomes are found in a human diploid cell?

15. What type of chart would help a genetic counselor track the passage of a trait through the generations of a family?

Chapter 14 The Human Genome

Section 14-1 Human Heredity (pages 341-348)



Key Concepts

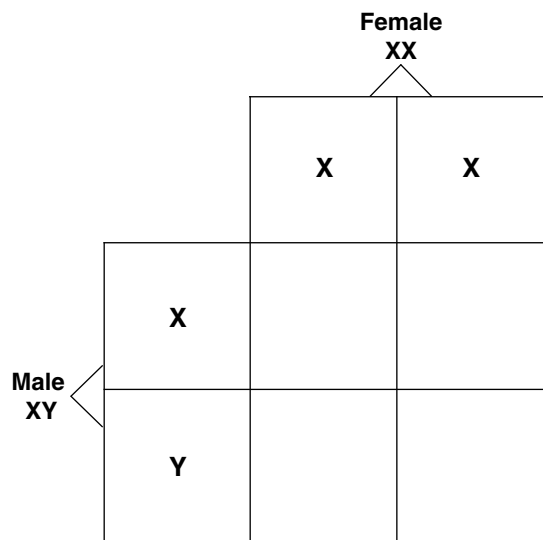
- How is sex determined?
- How do small changes in DNA cause genetic disorders?

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.



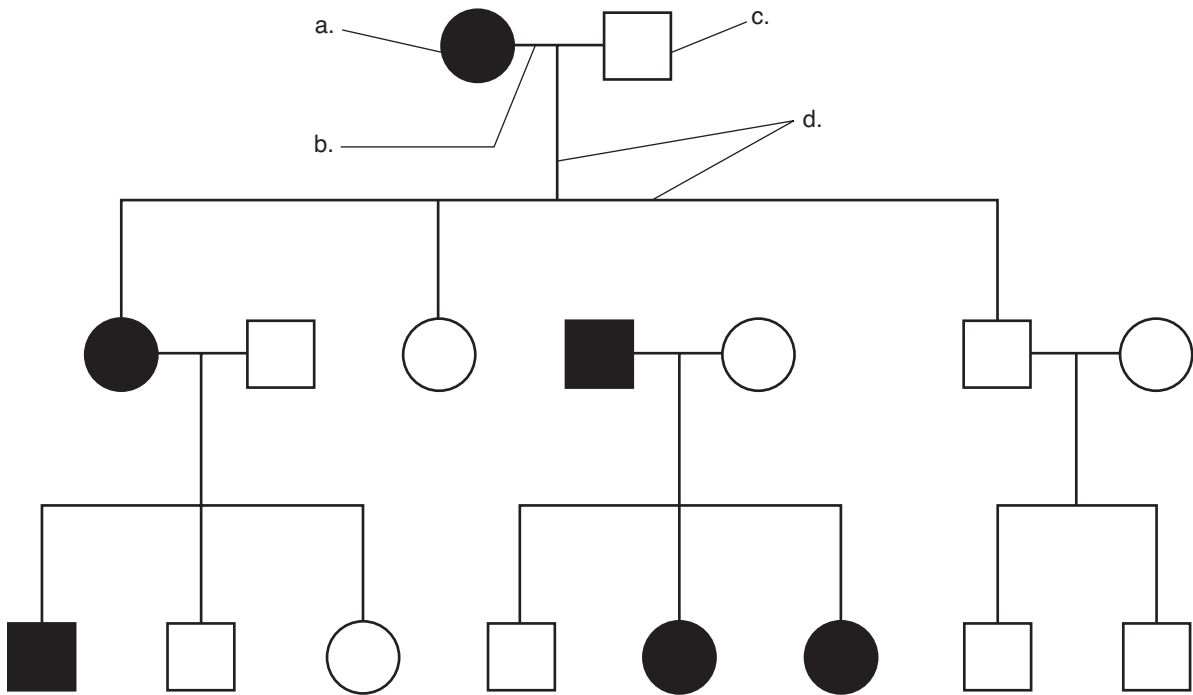
4. Why is there the chance that half of the zygotes will be female and half will be male?

Human Traits (pages 342–343)

5. What does a pedigree chart show? _____

Match the labels to the parts of the pedigree chart shown below. Some of the parts of the pedigree chart may be used more than once.

Pedigree Chart



- _____ 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. _____

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.

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

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.

SEX-LINKED DISORDERS IN HUMANS

Disorder	Description	Cause
Colorblindness		
		A recessive allele in either of two genes resulting in a missing protein required for normal blood clotting
		A defective version of the gene that codes for a muscle protein

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.

		X^cY	
		X^c	Y
X^cX^c	X^c		
	X^c		

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
 - b. mental retardation
 - c. colorblindness
 - 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

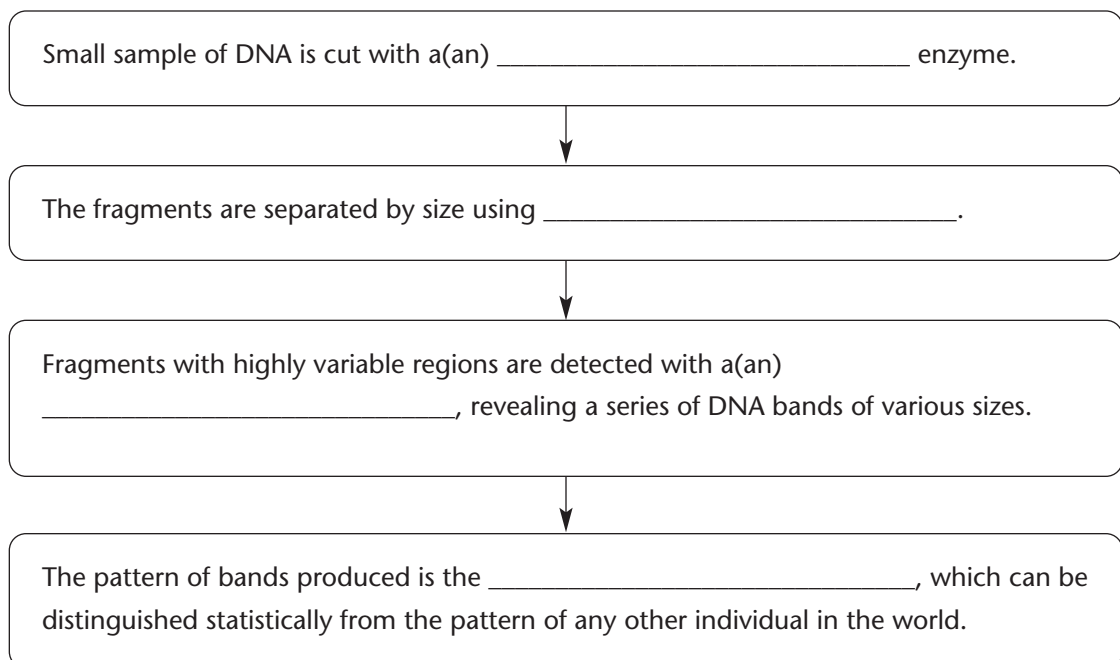
- What is the goal of the Human Genome Project?
- What is gene therapy?

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.



6. Circle the letter of each source for a DNA sample from an individual.
- a. blood
 - b. sperm
 - c. clothing
 - 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. _____