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Chapter 14

The Human Genome

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

This section explains what scientists know about human chromosomes, as well as the inheritance of certain human traits and disorders. It also describes how scientists study the inheritance of human traits.

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. Autosomes are all the chromosomes, except the sex chromosomes.

d. Biologists would write 46XY to indicate a human female.

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

Male (XY) × Female (XX)

	X	X
X		
Y		

4. Why is there the chance that half of the zygotes will be 46XX and half will be 46XY? _____

_____

_____

5. Is the following sentence true or false? Human chromosomes contain both protein and a single, double-stranded DNA molecule.

_____

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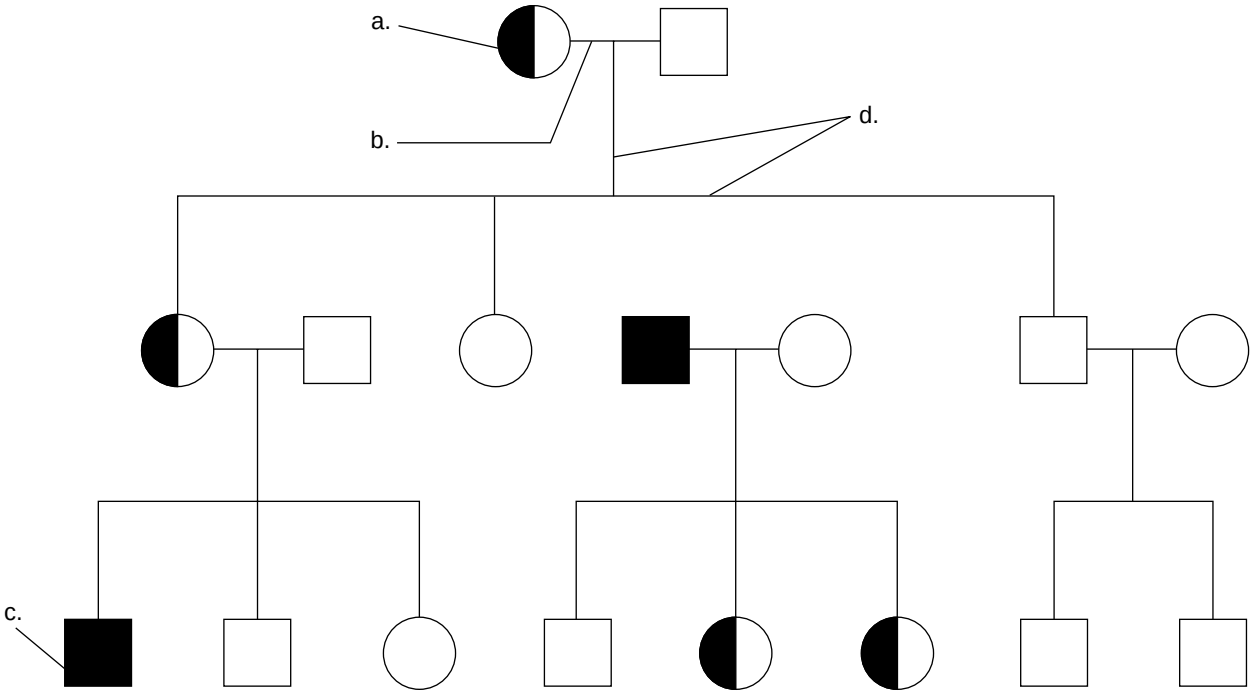
Human Traits (pages 342–343)

6. 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



_____ **7.** A person that has the trait

_____ **8.** A male

_____ **9.** A carrier of the trait

_____ **10.** Represents a marriage

_____ **11.** A female

_____ **12.** Connects parents to their children

13. 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)

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

_____

15. 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.

16. 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.

	Description	Genetic Disorder
_____	**17.** Nervous system breakdown caused by an autosomal recessive allele	**a.** Phenylketonuria (PKU)
_____	**18.** A form of dwarfism caused by an autosomal dominant allele	**b.** Tay-Sachs disease
_____	**19.** A buildup of phenylalanine caused by an autosomal recessive allele	**c.** Achondroplasia
_____	**20.** A progressive loss of muscle control and mental function caused by an autosomal dominant allele	**d.** Huntington's disease

From Gene to Molecule (pages 346–348)

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

_____

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

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

_____

24. What makes an allele dominant, recessive, or codominant? _____

_____

Section 14–2 Human Chromosomes (pages 349–353)

This section describes the structure of human chromosomes. It also describes genetic disorders that are sex-linked, as well as disorders caused by nondisjunction.

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 on the same chromosome tend to be inherited together.

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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 compare-and-contrast table for sex-linked genes.

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.

$X^CX^c \times X^CY$

	X^C	Y
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 saw a white cat with orange and black spots, could it be 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 genotype XO has inherited only one X chromosome and is sterile.

b. Females with the genotype 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.

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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)

This section explains how genetic engineering techniques are being used to study the genes and chromosomes in the human genome. It also describes how this information is used for 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. Circle the letter of each source for a DNA sample from an individual.

a. blood

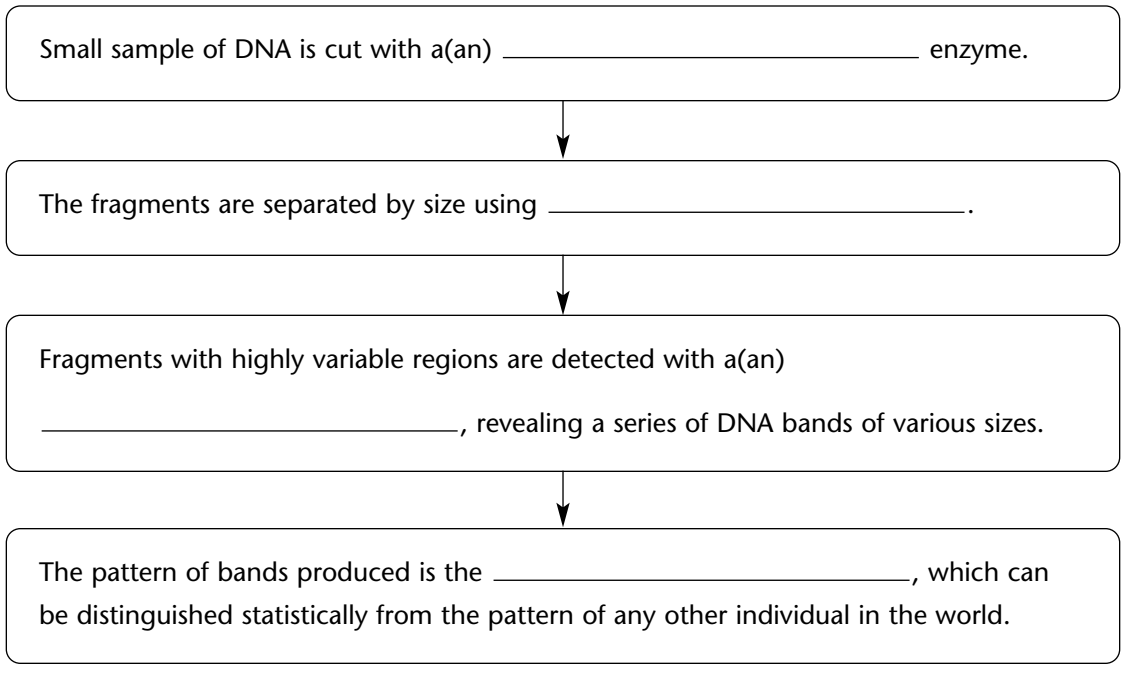
b. sperm

c. clothing

d. hair with tissue at the base

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

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



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. The sequence 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 the central question about the human genome that biologists will study next? _____

_____

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12. What is an "open reading frame" and what is it used for? _____

_____

_____

13. The mRNA coding regions of most genes are interrupted by _____, which have special DNA sequences marking their boundaries.

14. List three other parts of the gene that researchers look for.

a. _____

b. _____

c. _____

15. Why are biotechnology companies interested in genetic information? _____

_____

16. Is the following sentence true or false? Human genome data is top secret and can be accessed only by certain people.

_____

Gene Therapy (pages 359–360)

17. What is gene therapy? _____

_____

_____

18. 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.

19. Have all gene therapy experiments been successful? Explain. _____

_____

_____

_____

Ethical Issues in Human Genetics (page 360)

20. What other changes could be made to the human genome by manipulating human cells? _____

_____

_____

21. What is the ultimate goal of biology? _____

_____

22. What is the responsibility of society in biology? _____

_____

_____

_____

23. Is the following true or false? Scientists should be expected to make all ethical decisions regarding advances in human genetics.

_____

WordWise

Use the clues to fill in the blanks with vocabulary terms from Chapter 14. Then, put the numbered letters in the correct spaces to find the hidden message.

Clues	Vocabulary Terms
Occurs when homologous chromosomes fail to separate during meiosis	___ ___ ___(1) ___(2) ___ ___ ___ ___(3) ___ ___(4) ___ ___ ___ ___(5)
Describes a trait that is controlled by many genes	___ ___(6) ___ ___ ___(7) ___ ___ ___ ___(8)
In humans, Y is a sex ____.	___(9) ___(10) ___ ___ ___ ___ ___(11) ___ ___ ___(12)
Technique that uses DNA to identify individuals	___(13) ___(14) ___(15) ___ ___ ___(16) ___ ___ ___ ___(17) ___ ___ ___(18) ___ ___ ___ ___ ___
Chart that shows the relationships within a family	___ ___ ___(19) ___ ___(20) ___ ___ ___(21)
A picture of chromosomes arranged in pairs	___ ___(22) ___(23) ___ ___ ___(24) ___ ___ ___(25)
A gene located on the X or Y chromosome is a _____ gene.	___(26) ___ ___ - ___ ___(27) ___(28) ___ ___(29) ___
Chromosomes that are not sex chromosomes	___(30) ___ ___ ___ ___(31) ___ ___ ___(32) ___(33)

Hidden Message:

___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___
4 10 30 28 20 32 33 16 5 13 14 15

___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___ ___
8 22 3 26 21 7 25 1 12 24 18 9

___ ___ ___ ___ ___ ___ ___ ___ ___ .
19 27 31 6 17 2 29 23 11