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CHAPTER 12 Human Genetics

Summary of Key Concepts

Concept 12.1 The nucleus contains an information-rich genome. (pp. 248–249)

An organism's *genome* is its complete set of DNA. The DNA that makes up your genome is found in chromosomes in the nucleus of most of the cells in your body.

Each of the 46 chromosomes in your cells contains one long DNA molecule. The DNA is able to fit in the nucleus because of the way it is packed. First, the DNA molecule wraps tightly around small proteins called *histones*. Next, the DNA and the histones wrap into a tight coil. This coil twists again into a thick structure called a "supercoil." Each DNA molecule is a long sequence of nucleotides, each containing one of four nitrogenous bases: A, T, G, or C. Different individuals have different nucleotide sequences.

In 2000, scientists working on the Human Genome Project provided a rough draft of the human genome's entire sequence. Scientists are now working to determine which parts of the genome code for proteins and what the functions of those proteins are. The information from the Human Genome Project can be used in many ways. Evolutionary relationships between species can be examined. Medical researchers can use the data to identify genes associated with diseases and to develop treatments for genetic disorders.

1. Describe how DNA is packed in the nucleus of cells. _____

_____

_____

2. Describe two ways scientists can use the data from the Human Genome Project. _____

_____

_____

Concept 12.2 Accidents affecting chromosomes can cause disorders. (pp. 250–254)

Occasionally an error in meiosis produces a gamete with an abnormal number of chromosomes. *Trisomy 21* is a condition in which a person has three number 21 chromosomes. Trisomy 21 results in a set of symptoms called *Down syndrome*. Errors in chromosome number such as trisomy 21 usually happen because of nondisjunction. In *nondisjunction* a pair of homologous chromosomes or sister chromatids fail to separate during meiosis. This produces a gamete with two copies of that chromosome. (Remember, gametes usually contain only one copy of each chromosome.)

Other disorders are caused by changes to chromosomes. A *duplication* occurs when a part of a chromosome is repeated. A *deletion* occurs when a part of a chromosome is lost. An *inversion* occurs when a part of a chromosome is reversed. A *translocation* occurs when a portion of a chromosome attaches to a non-homologous chromosome.

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Other changes are caused by single genes changing their position on the chromosome or moving to another chromosome entirely. Barbara McClintock discovered these "jumping genes," called *transposons*, while studying the genetics of corn in the 1940s. Sometimes transposons are inserted into the middle of an existing gene, disrupting the function of that gene.

3. How does nondisjunction cause trisomy 21? _____

_____

_____

4. List four types of changes that cause damage to chromosomes. _____

_____

5. Define the term *transposon,* and explain the effect a transposon can have on other genes. _____

_____

_____

Concept 12.3 Mendel's principles apply to humans. (pp. 255–259)

Scientists studying the inheritance of traits in humans often use a diagram called a pedigree. A *pedigree* traces the occurrence of a trait in a family.

Most human genetic disorders are inherited as recessive traits. Some of these disorders, such as albinism, are not life-threatening. Others, such as Tay-Sachs disease and cystic fibrosis, are very serious disorders. An individual with one copy of the allele for a recessive disorder who does not show symptoms is called a *carrier.* Mendel's principles can be applied to find out the probability of a carrier having a child with that disorder. Some human disorders are inherited as dominant traits. Dominant alleles for deadly disorders are less common than recessive alleles for deadly disorders. Individuals with a dominant allele for a deadly disorder usually do not live to pass the allele on to offspring.

Some human disorders are sex-linked, meaning the allele for the trait is located on a sex chromosome. Red-green color blindness is an example of a sex-linked human disorder. The allele for color-blindness is found on the X chromosome, so a male who inherits one allele for this recessive trait will be colorblind. A female must inherit two copies of the recessive allele in order to be colorblind.

A *genetic counselor* is an individual trained to collect and interpret family histories of genetic disorders. If two people who are considering having children have family histories of a genetic disorder, a genetic counselor can help them determine their risk of having a child with the disorder.

6. What is a carrier? _____

_____

7. Why are males more likely than females to be colorblind? _____

_____

_____

_____

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Concept 12.4 Genetic changes contribute to cancer. (pp. 260–261)
Two classes of genes code for proteins that regulate the cell cycle. One class of genes codes for proteins called *growth factors*, which start the process of cell division. The other class of genes, *tumor-suppressor genes*, produce proteins that stop cell division when a cell's DNA is damaged.

Cancer usually does not result from one mutation to one gene. It takes several mutations for cancer to develop. Mutations can cause a gene to become a cancer-causing gene, called an *oncogene*. Most mutations that result in cancer occur in body cells and are not passed to offspring. Sometimes a mutation occurs in cells that give rise to gametes, and these mutations are passed from generation to generation. One example of a cancer gene that is inherited is *BRCA1,* a mutated form of a tumor-suppressor gene that increases the risk of breast cancer in women who inherit it.

8. Describe the functions of growth factors and tumor-suppressor genes.

_____

_____

9. What is an oncogene? _____

_____

Reading Skills Practice

Defining terms Define the Key Terms in Concept 12.2 using your own words. Be sure each definition is a complete sentence.

Vocabulary Review and Reinforcement

In 1–8, write the letter of the correct definition on the line next to each term.

_____ **1.** growth factors
_____ **2.** genome
_____ **3.** tumor-suppressor genes
_____ **4.** carrier
_____ **5.** transposon
_____ **6.** trisomy 21
_____ **7.** oncogene
_____ **8.** histones

a. condition caused by having three number 21 chromosomes
b. small proteins involved in DNA packing
c. "jumping gene"
d. individual with one copy of the allele for a recessive disorder
e. cancer-causing gene
f. genes that code for proteins that stop cell division
g. complete set of genetic material in an organism
h. proteins that initiate cell division

In 9–16, fill in the blanks with the appropriate terms from the chapter.

9. A(n) _____ collects and analyzes data about inheritance patterns and explains the results and their significance.

10. People with trisomy 21 have a general set of symptoms called _____.

11. A(n) _____ occurs when a part of a chromosome is repeated.

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12. A family tree that records and traces the occurrence of a trait in a family is a(n) _____.

13. An event called _____ is usually caused by a pair of homologous chromosomes failing to separate during meiosis.

14. A(n) _____ is a change in a chromosome involving reversal of a fragment of the chromosome.

15. A(n) _____ occurs when a fragment of one chromosome attaches to a nonhomologous chromosome.

WordWise

Find and circle eight Key Terms from the chapter in the puzzle below. Words may appear horizontally, vertically, or diagonally. Then write a definition for each term on a separate sheet of paper.

w	d	e	l	e	t	i	o	n	e	e	n
r	r	e	j	w	t	y	n	k	f	p	o
y	p	o	l	p	y	p	c	s	g	t	s
c	q	w	h	i	s	t	o	n	e	g	o
l	a	q	o	y	u	e	g	k	n	m	p
d	u	r	e	n	i	a	e	b	o	y	s
s	a	d	r	r	c	e	n	q	m	t	n
k	o	c	r	i	a	r	e	w	e	r	a
a	y	i	n	v	e	r	s	i	o	n	r
z	p	e	d	i	g	r	e	e	s	o	t