

Chapter 8

Human Genetics and Biotechnology Worksheets



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- **Lesson 8.1: Human Chromosomes and Genes**
- **Lesson 8.2: Human Inheritance**
- **Lesson 8.3: Biotechnology**

8.1 Human Chromosomes and Genes

Lesson 8.1: True or False

Name _____ Class _____ Date _____

Write true if the statement is true or false if the statement is false.

- _____ 1. The human genome consists of about 3 million base pairs.
- _____ 2. By 2003, scientists had sequenced all of the base pairs of a sample of human DNA.
- _____ 3. In humans, chromosome 22 is the largest chromosome, and chromosome 1 is the smallest chromosome.
- _____ 4. None of the genes on the Y-chromosome is essential to survival.
- _____ 5. The role of the majority of the 3 billion base pairs in the human genome is not known.
- _____ 6. Humans have 22 pairs of autosomes.
- _____ 7. Genes that are located on the different chromosomes are linked genes.
- _____ 8. Linkage is related to crossing-over during meiosis.
- _____ 9. Females have two X chromosomes, and males have an X and a Y chromosome.
- _____ 10. Genes that assort independently during meiosis will always be in different gametes.
- _____ 11. The hemophilia A gene is on the X chromosome.
- _____ 12. Only the X chromosome contains genes that determine sex.
- _____ 13. The female is the “default” sex of the human species.
- _____ 14. Most sex-linked genes are on the Y chromosome.
- _____ 15. Most human cells have 23 chromosomes.

Lesson 8.1: Critical Reading

Name _____ Class _____ Date _____

Read these passages from the text and answer the questions that follow.

Chromosomes and Genes

Each species has a characteristic number of chromosomes. The human species is characterized by 23 pairs of chromosomes, as shown in the FlexBook.

Autosomes

Of the 23 pairs of human chromosomes, 22 pairs are autosomes. **Autosomes** are chromosomes that contain genes for characteristics that are unrelated to sex. These chromosomes are the same in males and females. The great majority of human genes are located on autosomes.

Sex Chromosomes

The remaining pair of human chromosomes consists of the **sex chromosomes**, X and Y. Females have two X chromosomes, and males have one X and one Y chromosome. In females, one of the X chromosomes in each cell is inactivated and known as a Barr body. This ensures that females, like males, have only one functioning copy of the X chromosome in each cell.

The X chromosome is much larger than the Y chromosome. The X chromosome has about 2,000 genes, whereas the Y chromosome has fewer than 100, none of which are essential to survival. Virtually all of the X chromosome genes are unrelated to sex. Only the Y chromosome contains genes that determine sex. A single Y chromosome gene, called SRY (which stands for sex-determining region Y gene), triggers an embryo to develop into a male. Without a Y chromosome, an individual develops into a female, so you can think of female as the default sex of the human species. Can you think of a reason why the Y chromosome is so much smaller than the X chromosome?

Human Genes

Humans have an estimated 20,000 to 22,000 genes. This may sound like a lot, but it really isn't. Far simpler species have almost as many genes as humans. However, human cells use splicing and other processes to make multiple proteins from the instructions encoded in a single gene. Of the 3 billion base pairs in the human genome, only about 25 percent make up genes and their regulatory elements. The functions of many of the other base pairs are still unclear.

The majority of human genes have two or more possible alleles. Differences in alleles account for the considerable genetic variation among people. In fact, most human genetic variation is the result of differences in individual DNA bases within alleles.

Questions

1. What are autosomes? How many do humans have?

2. Compare the X and Y chromosome.

3. In terms of sex chromosomes, what is the genotype of a female? a male?

4. How do humans use their genes to produce more than 22,000 proteins?

5. What is the importance of alleles in humans?

Lesson 8.1: Multiple Choice

Name _____ Class _____ Date _____

Circle the letter of the correct choice.

- All of the DNA of the human species makes up the human
 - genes.
 - genome.
 - chromosomes.
 - DNA.
- Humans have _____ bases divided among _____ chromosomes.
 - 3 million, 23
 - 3 million, 23 pairs of
 - 3 billion, 23
 - 3 billion, 23 pairs of
- Differences between the X and Y chromosomes include which of the following? (1) The X chromosome has many more genes than the Y chromosome. (2) Virtually all of the X chromosome genes are unrelated to sex, whereas the Y chromosome contains genes that determine sex. (3) Both males and females have only one functioning copy of the X chromosome in each cell.
 - 1 only
 - 2 only
 - 1 and 2
 - 1, 2, and 3
- The goal of the Human Genome Project was to
 - sequence all 3 billion base pairs of human DNA.
 - sequence all human DNA and identify all 22,000 proteins.
 - develop linkage maps of all 22 autosomes of chromosomes.
 - all of the above
- Linked genes
 - are on homologous chromosomes.
 - are on the same chromosome.
 - are on sister chromatids.
 - are on non-homologous chromosomes.
- Most of the human genome is made of
 - genes.
 - regulatory regions.
 - intergenic regions.
 - chromosomes.
- Which of the following statements is correct?
 - The higher the frequency of crossing-over, the closer together on the same chromosome the genes are presumed to be.
 - The lower the frequency of crossing-over, the closer together on the same chromosome genes are presumed to be.
 - The lower the frequency of crossing-over, the farther apart on the same chromosome the genes are presumed to be.

- (d) With a high frequency of crossing-over, genes are presumed to be on different chromosomes.
8. A normal human male has
- (a) 22 autosomes, and one X chromosome and one Y chromosome.
 - (b) 22 pairs of autosomes, and one X chromosome and one Y chromosome.
 - (c) 23 autosomes, and one X chromosome and one Y chromosome.
 - (d) 23 pairs of autosomes, and one X chromosome and one Y chromosome.

Lesson 8.1: Vocabulary I

Name _____ Class _____ Date _____

Match the vocabulary word with the proper definition.

Definitions

- _____ 1. 20,000 to 22,000 in humans
- _____ 2. an international project to sequence the entire human genome
- _____ 3. determine the sex of the person
- _____ 4. genes that are located on the same chromosome
- _____ 5. all of the DNA of the human species
- _____ 6. genes on the X-chromosome
- _____ 7. chromosomes that contain genes for characteristics that are unrelated to sex
- _____ 8. female
- _____ 9. male
- _____ 10. 23 pairs in humans
- _____ 11. shows the locations of genes on a chromosome
- _____ 12. genes located on the sex chromosomes

Terms

- a. autosome
- b. chromosomes
- c. gene
- d. human genome
- e. Human Genome Project
- f. linkage map
- g. linked genes
- h. sex chromosomes
- i. sex-linked gene
- j. X-linked gene
- k. XX
- l. XY

Lesson 8.1: Vocabulary II

Name _____ Class _____ Date _____

Fill in the blank with the appropriate term.

1. The human _____ is all of the DNA of a human.
2. The X and Y chromosomes are known as the _____ chromosomes.
3. Genes that are located on the same chromosome are _____ genes.
4. Differences in alleles account for the considerable _____ variation among people.
5. Most sex-linked genes are on the _____ chromosome.
6. Chromosomes 1 to 22 are known as _____.
7. Chromosome _____ is the largest chromosome.
8. A linkage _____ shows the locations of genes on a chromosome.
9. The number of human genes is about _____ to _____.
10. Genes on non-homologous chromosomes are not linked.
11. Linkage explains why certain characteristics are frequently _____ together.
12. Humans have 23 pairs of _____.

Lesson 8.1: Critical Writing

Name _____ Class _____ Date _____

Thoroughly answer the question below. Use appropriate academic vocabulary and clear and complete sentences.

Being very specific, what makes you different from everyone else?

8.2 Human Inheritance

Lesson 8.2: True or False

Name _____ Class _____ Date _____

Write true if the statement is true or false if the statement is false.

- _____ 1. Characteristics that are encoded in DNA are called genetic diseases.
- _____ 2. Widow's peak and hitchhiker's thumb are multiple allele traits.
- _____ 3. Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits.
- _____ 4. Most human traits have more complex modes of inheritance than simple Mendelian inheritance.
- _____ 5. Because it is a polygenic trait, human height can be represented by a bell-shaped graph.
- _____ 6. Pleiotropy is when a more than one gene affects a single trait.
- _____ 7. Most genetic disorders are controlled by dominant alleles.
- _____ 8. Triple X syndrome results in XYXX males.
- _____ 9. A karyotype is a picture of a cell's chromosomes.
- _____ 10. A recessive X-linked allele is always expressed in males.
- _____ 11. The alleles for ABO blood type are the A, B, AB and O alleles.
- _____ 12. Amniocentesis can be used to see if the mother has any genetic abnormalities.
- _____ 13. Down syndrome is also known as trisomy 21.
- _____ 14. A mutant recessive allele is not expressed in people who inherit just one copy of it.
- _____ 15. Epistasis is when one gene affects the expression of another gene.

Lesson 8.2: Critical Reading

Name _____ Class _____ Date _____

Read these passages from the text and answer the questions that follow.

Genetic Disorders

Many genetic disorders are caused by mutations in one or a few genes. Other genetic disorders are caused by abnormal numbers of chromosomes.

Genetic Disorders Caused by Mutations

Table 8.1 lists several genetic disorders caused by mutations. Some of the disorders are caused by mutations in autosomal genes, others by mutations in X-linked genes. Which disorder would you expect to be more common in males than females?

Table 8.1: Genetic Disorders Caused by Mutations

Genetic Disorder	Direct Effect of Mutation	Signs and Symptoms of the Disorder	Mode of Inheritance
Marfan syndrome	defective protein in connective tissue	heart and bone defects and unusually long, slender limbs and fingers	autosomal dominant
Sickle cell anemia	abnormal hemoglobin protein in red blood cells	sickle-shaped red blood cells that clog tiny blood vessels, causing pain and damaging organs and joints	autosomal recessive
Vitamin D-resistant rickets	lack of a substance needed for bones to absorb minerals	soft bones that easily become deformed, leading to bowed legs and other skeletal deformities	X-linked dominant
Hemophilia A	reduced activity of a protein needed for blood clotting	internal and external bleeding that occurs easily and is difficult to control	X-linked recessive

Few genetic disorders are controlled by dominant alleles. A mutant dominant allele is expressed in every individual who inherits even one copy of it. If it causes a serious disorder, affected people may die young and fail to reproduce. Therefore, the mutant dominant allele is likely to die out of the population. A mutant recessive allele, such as the allele that causes sickle cell anemia, is not expressed in people who inherit just one copy of it. These people are called carriers. They do not have the disorder themselves, but they carry the mutant allele and can pass it to their offspring. Thus, the allele is likely to pass on to the next generation rather than die out.

Chromosomal Disorders

Mistakes may occur during meiosis that result in **nondisjunction**. This is the failure of replicated chromosomes to separate during meiosis II. Some of the resulting gametes will be missing a chromosome, while others will have an extra copy of the chromosome. If such gametes are fertilized and form zygotes, they usually do not survive. If they do survive, the individuals are likely to have serious genetic disorders.

Table 8.2 lists several genetic disorders that are caused by abnormal numbers of chromosomes.

Table 8.2: **Genetic Disorders Caused by Abnormal Numbers of Chromosomes**

Genetic Disorder	Genotype	Phenotypic Effects
Down syndrome	extra copy (complete or partial) of chromosome 21	developmental delays, distinctive facial appearance, and other abnormalities
Turner's syndrome	one X chromosome but no other sex chromosome (XO)	female with short height and infertility (inability to reproduce)
Triple X syndrome	three X chromosomes (XXX)	female with mild developmental delays and menstrual irregularities
Klinefelter's syndrome	one Y chromosome and two or more X chromosomes (XXY, XXXY)	male with problems in sexual development and reduced levels of the male hormone testosterone

Having the wrong number of chromosomes causes the genetic disorders described in **Table 8.2**. Most chromosomal disorders involve the X chromosome. Look back at the X and Y chromosomes and you will see why. The X and Y chromosomes are very different in size, so nondisjunction of the sex chromosomes occurs relatively often.

Questions

1. What two main ways cause genetic disorders?
2. What are the signs and symptoms of sickle cell anemia?
3. Why are few genetic disorders controlled by dominant alleles?
4. Explain what causes chromosomal disorders.
5. Describe the cause and symptoms of Down syndrome.

Lesson 8.2: Multiple Choice

Name _____ Class _____ Date _____

Circle the letter of the correct choice.

- Which of the following statements concerning genetic disorders is correct? (1) Many genetic disorders are caused by mutations in one or a few genes. (2) Some genetic disorders are caused by abnormal numbers of chromosomes. (3) Most genetic disorders are caused by dominant alleles. (4) Chromosomal disorders result from nondisjunction during mitosis.
 - 1 only
 - 1 and 2
 - 1, 2, and 3
 - all four statements are correct
- Single-gene X-linked traits have a different pattern of inheritance than single-gene autosomal traits because
 - females have just one X chromosome.
 - females have two X chromosomes.
 - males have two X chromosomes.
 - males have just one X chromosome.
- ABO blood type is a multiple allele trait. Which of the following are possible ABO blood type phenotypes? (1) A, (2) B, (3) AB, (4) O, (5) AO, (6) BO
 - 1 and 2 only
 - 3 and 4 only
 - 5 and 6 only
 - 1, 2, 3, and 4
- Hemophilia A is due to
 - a defective protein in connective tissue.
 - abnormal hemoglobin protein in red blood cells.
 - reduced activity of a protein needed for blood clotting.
 - lack of a substance needed for bones to absorb minerals.
- Down syndrome is due to
 - nondisjunction of chromosome 21.
 - nondisjunction of the X chromosome.
 - one Y chromosome and two or more X chromosomes.
 - nondisjunction of the Y chromosome.
- Which of the following statements is true?
 - A recessive X-linked allele is always expressed in males.
 - Males will have two alleles for any X-linked trait.
 - Males must inherit two copies of a recessive X-linked allele to express the recessive trait.
 - X-linked recessive traits are less common in males than females.
- In a polygenic trait, such as adult height, most people
 - will have a phenotype close to the average.
 - will have the dominant phenotype.
 - will have a heterozygous genotype.
 - will be 5 feet 8 inches tall.

8. A mother has red-green color blindness. Her husband is not affected.
- (a) Half of their daughters will have red-green color blindness.
 - (b) All of their daughters will have red-green color blindness.
 - (c) All of their sons will have red-green color blindness.
 - (d) All of their children will have red-green color blindness.

Lesson 8.2: Vocabulary I

Name _____ Class _____ Date _____

Match the vocabulary word with the proper definition.

Definitions

- _____ 1. involves inserting normal genes into cells with mutant genes
- _____ 2. when one gene affects the expression of another gene
- _____ 3. the inheritance of traits controlled by a single gene with two alleles
- _____ 4. trait controlled by genes on the sex chromosomes
- _____ 5. when a single gene affects more than one trait
- _____ 6. an example of a multiple allele trait
- _____ 7. the failure of replicated chromosomes to separate during meiosis
- _____ 8. trait controlled by a gene on one of the 22 human autosomes
- _____ 9. characteristics (traits) encoded in DNA
- _____ 10. shows how a trait is passed from generation to generation within a family
- _____ 11. red-green color blindness
- _____ 12. traits controlled by a single gene with more than two alleles

Terms

- a. ABO blood type
- b. autosomal trait
- c. epistasis
- d. gene therapy
- e. genetic trait
- f. Mendelian inheritance
- g. multiple allele trait
- h. nondisjunction
- i. pedigree
- j. pleiotropy
- k. sex-linked trait
- l. X-linked trait

Lesson 8.2: Vocabulary II

Name _____ Class _____ Date _____

Fill in the blank with the appropriate term.

1. Characteristics that are encoded in DNA are called genetic _____.
2. Sex-linked traits are usually associated with genes on the _____ chromosome.
3. Traits controlled by a single gene with more than two alleles are called multiple _____-traits.
4. Mendelian inheritance refers to the inheritance of traits controlled by a single _____ with two _____.
5. ABO blood type is a _____ allele trait.
6. Sickle cell anemia results in an abnormal _____ protein in red blood cells.
7. Hemophilia A results in reduced activity of a protein needed for blood _____.
8. _____ is caused by an extra copy of chromosome 21.
9. An example of a recessive _____ trait would be red-green color blindness.
10. Human height is an example of a _____ trait.
11. Many genetic disorders are caused by _____ in one or a few genes.
12. Nondisjunction occurs during _____.

Lesson 8.2: Critical Writing

Name _____ Class _____ Date _____

Thoroughly answer the question below. Use appropriate academic vocabulary and clear and complete sentences.

Explain two complex modes of human inheritance. Give examples.

8.3 Biotechnology

Lesson 8.3: True or False

Name _____ Class _____ Date _____

Write true if the statement is true or false if the statement is false.

- _____ 1. Two common biotechnology techniques are gene cloning and genetic engineering.
- _____ 2. Gene cloning is the process of isolating and making copies of a chromosome.
- _____ 3. Biotechnology has raised ethical questions.
- _____ 4. When genes are cloned, DNA polymerase is used to join two pieces of DNA together.
- _____ 5. Recombinant DNA is made from joining DNA from different sources.
- _____ 6. Insulin was the first human protein to be produced by gene cloning.
- _____ 7. The purpose of biotechnology is to create organisms that are useful to humans.
- _____ 8. The polymerase chain reaction makes many copies of a gene or other DNA segment.
- _____ 9. Gene cloning involves three steps: isolation, transformation, and selection.
- _____ 10. The three steps of PCR are denaturing, annealing, and elongation.
- _____ 11. The enzyme Taq Polymerase can work at high temperatures.
- _____ 12. Transgenic crops have been created that make some food taste better.
- _____ 13. Denaturing DNA occurs at room temperature.
- _____ 14. Medicine and agriculture are two major fields that use biotechnology.

Lesson 8.3: Critical Reading

Name _____ Class _____ Date _____

Read these passages from the text and answer the questions that follow.

Gene Cloning

Gene cloning is the process of isolating and making copies of a gene. This is useful for many purposes. For example, gene cloning might be used to isolate and make copies of a normal gene for gene therapy. Gene cloning involves four steps: isolation, ligation, transformation, and selection.

1. In isolation, an enzyme is used to break DNA at a specific base sequence. This is done to isolate a gene.
2. During ligation, the enzyme DNA ligase combines the isolated gene with plasmid DNA from bacteria. (Plasmid DNA is circular DNA that is not part of a chromosome and can replicate independently.) The DNA that results is called **recombinant DNA**.
3. In transformation, the recombinant DNA is inserted into a living cell, usually a bacterial cell. Changing an organism in this way is also called **genetic engineering**.
4. Selection involves growing transformed bacteria to make sure they have the recombinant DNA. This is a necessary step because transformation is not always successful. Only bacteria that contain the recombinant DNA are selected for further use.

Polymerase Chain Reaction

The **polymerase chain reaction (PCR)** makes many copies of a gene or other DNA segment. This might be done in order to make large quantities of a gene for genetic testing. PCR involves three steps: denaturing, annealing, and extension. They are repeated many times in a cycle to make large quantities of the gene.

1. Denaturing involves heating DNA to break the bonds holding together the two DNA strands. This yields two single strands of DNA.
2. Annealing involves cooling the single strands of DNA and mixing them with short DNA segments called primers. Primers have base sequences that are complementary to segments of the single DNA strands. As a result, bonds form between the DNA strands and primers.
3. Extension occurs when an enzyme (Taq polymerase or Taq DNA polymerase) adds nucleotides to the primers. This produces new DNA molecules, each incorporating one of the original DNA strands.

Questions

1. What is gene cloning?

2. What is PCR? Why is PCR done?

3. What are the three steps of PCR?

4. What are the four steps of gene cloning?
5. What is recombinant DNA?
6. Why is it important to select for transformed bacteria?

Lesson 8.3: Multiple Choice

Name _____ Class _____ Date _____

Circle the letter of the correct choice.

1. Recombinant DNA
 - (a) results from the ligation of an isolated gene and plasmid DNA.
 - (b) is inserted into a living cell in the transformation process.
 - (c) is screened for in the selection process.
 - (d) all of the above
2. The steps of gene cloning are, in order,
 - (a) isolation, transformation, ligation, and selection.
 - (b) isolation, ligation, transformation, and selection.
 - (c) ligation, transformation, isolation, and selection.
 - (d) selection, transformation, ligation, and isolation.
3. The steps of PCR are, in order,
 - (a) denaturing, annealing, and extension.
 - (b) denaturing, extension, and annealing.
 - (c) annealing, extension, and denaturation.
 - (d) extension, annealing, and denaturation.
4. Transgenic crops have been created that
 - (a) yield more food.
 - (b) resist insect pests.
 - (c) survive drought.
 - (d) all of the above
5. Ethical, legal, and social issues associated with biotechnology would include questions about
 - (a) the safety of genetically modified crops.
 - (b) the use of biotechnology in modifying a baby's genotype.
 - (c) the ownership of genetically modified organisms.
 - (d) all of the above
6. PCR allows scientists to
 - (a) rapidly make many copies of a gene or other DNA segment.
 - (b) clone a recombinant DNA in bacteria.
 - (c) ligate together two pieces of DNA from different sources.
 - (d) all of the above.
7. The first human protein produced using biotechnology was
 - (a) cytokine.
 - (b) insulin.
 - (c) DNA ligase.
 - (d) Taq polymerase.
8. Biotechnology methods are used in which of the following? (1) medicine, (2) agriculture, (3) law enforcement.
 - (a) 1 only
 - (b) 2 only
 - (c) 1 and 2

(d) 1, 2, and 3

Lesson 8.3: Vocabulary I

Name _____ Class _____ Date _____

Match the vocabulary word with the proper definition.

Definitions

- _____ 1. enzyme that joins two pieces of DNA
- _____ 2. first human protein to be produced by genetic engineering
- _____ 3. process that makes many copies of a gene or other DNA segment
- _____ 4. the process of isolating and making copies of a gene
- _____ 5. the process of placing recombinant DNA into a living cell
- _____ 6. circular DNA that is not part of a chromosome
- _____ 7. genetically modified plants
- _____ 8. changing an organism by transforming with recombinant DNA
- _____ 9. the use of technology to change the genetic makeup of living things for human purposes
- _____ 10. made by joining DNA from two different sources

Terms

- a. biotechnology
- b. DNA ligase
- c. gene cloning
- d. genetic engineering
- e. insulin
- f. plasmid
- g. polymerase chain reaction
- h. recombinant DNA
- i. transformation
- j. transgenic crop

Lesson 8.3: Vocabulary II

Name _____ Class _____ Date _____

Fill in the blank with the appropriate term.

1. Transgenic crops are genetically modified with new _____ that code for traits useful to humans.
2. The _____ makes many copies of a gene or other DNA segment.
3. DNA _____ can join together an isolated gene and plasmid DNA.
4. Gene _____ is the process of isolating and making copies of a gene.
5. Gene cloning involves four steps: isolation, ligation, _____, and selection.
6. Plasmid DNA is circular DNA that is not part of a _____.
7. Changing an organism by transforming with recombinant DNA is known as genetic _____.
8. The use of biotechnology has raised a number of _____, legal, and social issues.
9. The first step of the PCR process is _____.
10. _____ DNA is made by combining DNA from two different sources.
11. Biotechnology can be used to transform bacteria so they are able to make human _____.
12. Biotechnology is the use of _____ to change the genetic makeup of living things for human purposes.

Lesson 8.3: Critical Writing

Name _____ Class _____ Date _____

Thoroughly answer the question below. Use appropriate academic vocabulary and clear and complete sentences.

Describe PCR. Discuss the necessary steps and potential applications in detail.