GreatHearts®

9th Grade Biology:

Inheritance Patterns and Human Genetics

March 23-27

Time Allotment: 40 minutes per day

Student Name: _____

Period: _____

Teacher Name: *Ms. Carstens*

Packet Overview

Date	Objective(s)	Page Number
Monday, March 23	1. Differentiate between sex chromosomes and autosomes. 2. Identify how X- and Y-linked genes affect inheritance.	2
Tuesday, March 24	1. Describe how X- and Y-linked genes affect the inheritance of traits. 2. Explain the effect of crossing-over on the inheritance of genes in linkage groups.	7
Wednesday, March 25	1. Differentiate between chromosome mutations and gene mutations. 2. Identify types of mutations.	12
Thursday, March 26	1. Identify the significance of using pedigrees to determine genetic traits and disorders. 2. Identify inheritance patterns and factors affecting them.	18
Friday, March 27	1. Identify and describe genetic disorders. 2. Identify and describe types of genetic disorder treatment.	26

Additional Notes:

⇒ Textbook pages for readings are scanned and found in this packet. See each day's instructions for the page numbers of the reading for that day.

⇒ A minor assessment is found on the last two pages of this packet, pp. 32-33. On Friday, after completing the assignment for the day, you will complete this minor assessment reviewing the unit. You may use your notes from the week. It should take approximately 12-15 minutes.

Academic Honesty

I certify that I completed this assignment independently in accordance with the GHNO Academy Honor Code.

Student signature:

_____

I certify that my student completed this assignment independently in accordance with the GHNO Academy Honor Code.

Parent signature:

_____

I. Monday, March 23

Unit – Ch 12: Inheritance Patterns and Human Genetics
Lesson 1: Chromosomes and Inheritance (Part 1)

Unit Overview:
In our next unit of biology, we will study chromosomes and their unique role in inherited traits as well as inheritance patterns in human genetics. We will explore research that led to the discovery of sex determination, sex-linked genes and traits, and linked genes. We will also differentiate between various types of mutations, identify conditions that can lead to these mutations, and explore preventative measures and therapies associated with the treatment of diseases and disorders that result from mutations.

As we begin this unit, it is important to remember what we have learned from previous lessons about DNA. First, recall that DNA is a nucleic acid formed by monomers called nucleotides, each consisting of a nitrogenous base (adenine, thymine, cytosine, guanine), a sugar called deoxyribose, and a phosphate group. These monomers are structured in long, patterned strands and are matched with a complementary, antiparallel strand, creating a double-helix structure.

We also know that chromosomes are structures composed primarily of an organism's DNA found in the cytoplasm of a prokaryotic cell and in the nucleus of a eukaryotic cell. Human beings contain 23 pairs of chromosomes (46 total), with one chromosome in each pair provided by the mother and the other provided by father. Having pairs of chromosomes results in organisms having two or more alternative forms of a gene, called alleles, for each inherited trait. Dominant trait alleles mask recessive trait alleles when present in a pair together. In fact, recessive traits only appear when both alleles are recessive. These allele pairs are passed to the offspring when a male gamete (sperm) and a female gamete (egg), produced in meiosis, are joined together during fertilization.

Finally, we understand that when meiosis and fertilization occur correctly and DNA is replicated, transcribed and translated accurately, an organism's chromosomes provide information for gene expression and the production of proteins that help carry out an organism's life functions. However, mistakes in any of these processes can lead to complications such as mutations. Mutations can be to the benefit of an organism, of no significance to an organism or can place an organism at a disadvantage, potentially harming it.

Objectives: Be able to do this by the end of this lesson.
1. Contrast sex chromosomes and autosomes.
2. Identify how an individual's sex is determined.

Introduction to Chapter 12 - Lesson 1
In this first lesson, you will read about the research of a scientist named Thomas Hunt Morgan. His experiments surrounding the common fruit fly (*Drosophila melanogaster* led to discoveries and observations of the X- and Y-chromosomes as well as X- and Y-linked traits. In addition to Morgan's research, you will read about how an organism's sex is determined.

Read and annotate pages 235-236 from your text (*found on pgs. 4-5 in this packet*). After reading and annotating, complete the guided outline and questions on the following page.

GUIDED OUTLINE: Chromosomes and Inheritance (pp. 235-236)

❖ Chromosomes and Inheritance

➢ Chromosomes

- Early Work
  - Thomas Hunt _____ experimented with _____ _____ in the early 1900s.
  - Each fly had _____ pairs of chromosomes. Three of the pairs were _____; one pair differed in _____ and _____
  - X- and Y-chromosomes are called _____ _____

    How are X- and Y-chromosomes different?

    _____

    _____
- Sex Chromosomes and Autosomes
  - Sex chromosomes – contain genes that determine the _____ of an individual
  - Autosomes – remaining chromosomes that _____

    _____
  - *Interpreting Figure 12.2, p. 236.* How are human female and male karyotypes similar? How are they different?

    _____

    _____

    _____
- Sex Determination
  - In mammals, how does the individual become male? How does the individual become female?

    _____

    _____

    _____

    _____

    Using the Punnett Square below, cross a male (XY) and a female (XX) to determine the ratio of male to female offspring.

_____ Female : _____ Male

	_____	_____
_____		
_____		

CHROMOSOMES AND INHERITANCE

Francis Collins and his lab group discovered the gene responsible for cystic fibrosis (CF). Cystic fibrosis often is a fatal genetic disorder. Thick, sticky mucus builds up and blocks ducts in the pancreas and intestines and causes difficulty in breathing. In this chapter, you will learn how diseases, such as CF, and characteristics, such as eye color, are inherited and expressed.

CHROMOSOMES

Jeff Pinard, a student in Collins's lab, studied more about how CF is inherited. Pinard, shown in Figure 12-1, has CF. Pinard and the rest of Collins's group were able to study the CF gene in part because of work carried out by geneticists in the early 1900s.

Early Work

In the early 1900s, researcher Thomas Hunt Morgan began experimenting with the small fruit fly *Drosophila melanogaster*. Morgan observed that the flies have four pairs of chromosomes. He also observed that three of the pairs were identical in both females and males, but one pair differed in size and shape. In females, the fourth pair had two identical chromosomes, now called *X chromosomes*. In males, the fourth pair had one X chromosome, but also a shorter chromosome, now called a *Y chromosome*. Today, geneticists call the X and Y chromosomes *sex chromosomes*.

FIGURE 12-1
Student Jeff Pinard uses molecular techniques to learn about the genetic variation that causes his symptoms of cystic fibrosis (CF).

SECTION 1

OBJECTIVES

- **Distinguish** between sex chromosomes and autosomes.
- **Explain** the role of sex chromosomes in sex determination.
- **Describe** how an X- or Y-linked gene affects the inheritance of traits.
- **Explain** the effect of crossing-over on the inheritance of genes in linkage groups.
- **Distinguish** between chromosome mutations and gene mutations.

VOCABULARY

sex chromosome
autosome
sex-linked trait
linked gene
chromosome map
map unit
germ-cell mutation
somatic-cell mutation
lethal mutation
deletion
inversion
translocation
nondisjunction
point mutation
substitution
frameshift mutation
insertion mutation

(a) Female karyotype

(b) Male karyotype

FIGURE 12-2

Human female and male karyotypes have in common 22 chromosome pairs, called *autosomes*. The karyotype of females (a) differs from that of males (b) in only the 23rd pair, which consists of the sex chromosomes. The 23rd chromosome pair has two X chromosomes in females and one X chromosome and one Y chromosome in males.

Sex Chromosomes and Autosomes

The **sex chromosomes** contain genes that determine the sex (gender) of an individual. The remaining chromosomes that are not directly involved in determining the sex of an individual are called **autosomes.** As in fruit flies, human males have an X and a Y chromosome, and human females have two X chromosomes. Figure 12-2a shows an example of the 23 pairs of human chromosomes from a female; Figure 12-2b shows an example of chromosomes from a male. In certain organisms, such as chickens and moths, males have two identical sex chromosomes, and females have two different sex chromosomes. Most plants and some fish lack sex chromosomes entirely.

Word Roots and Origins

autosome

from the Greek *autos,* meaning "self," and *soma,* meaning "a body"

Sex Determination

Like other homologous chromosomes, sex chromosomes pair during meiosis I. As meiosis proceeds, the paired chromosomes separate and move to different cells. As a result, a sperm cell has an equal chance of receiving an X chromosome or a Y chromosome. Each egg, however, receives only an X chromosome. This system of sex determination results in a one-to-one ratio of males to females. Each egg and sperm cell also receives a single copy of each autosome.

In mammals, when an egg that carries an X chromosome is fertilized by a sperm carrying a Y chromosome, the offspring has an XY pair and is male. Likewise, when an egg is fertilized by a sperm cell carrying an X chromosome, the offspring has an XX pair and is female. In a male mammal, the Y chromosome contains a gene called *SRY* for **S**ex-determining **R**egion **Y.** This gene codes for a protein that causes the gonads of an embryo to develop as testes. Because female embryos do not have the *SRY* gene, the gonads develop as ovaries.

Closing: Check your understanding of the lesson by answering the following question in 4-6 sentences.

1. How are human chromosomes similar to that of other species? How are they different? (Provide at least two similarities and at least one difference.)

_____

_____

_____

_____

_____

_____

_____

_____

_____

_____

II. Tuesday, March 24

Unit – Ch 12: Inheritance Patterns and Human Genetics
Lesson 2: Chromosomes and Inheritance (Part 2)

Lesson 2 Socratic Guiding Question: Keep this question in mind as you study!
What impact did Morgan's research have on our understanding of inheritance and human genetics?

Objectives: Be able to do this by the end of this lesson.

1. Describe how X- and Y-linked genes affect the inheritance of traits.
2. Explain the effect of crossing-over on the inheritance of genes in linkage groups.

Introduction to Lesson 2

From the unit introduction, recall that sex chromosomes are formed during meiosis. In previous lessons, you also learned that during fertilization in humans, gametes (male and female sex cells) combine to provide the offspring with 23 pairs of chromosomes. In your lesson yesterday, we discovered that 22 pairs of chromosomes in humans are identical, but one pair differs between males and females. In today's lesson, you will be exploring genes and traits associated with this difference. You will also continue to delve into Morgan's research and observations regarding what he called "linked genes." Our final discovery informs you of a tool called a chromosome map, developed by researchers, and how it impacts the study of genes.

Read and annotate pages 237-238 from your text (*found on pgs. 9-10 in this packet*). After reading and annotating, complete the guided outline and questions below.

GUIDED OUTLINE: Chromosomes and Inheritance (pp. 236-237)

- Effects of Gene Location
  - Describe the experiment and data that led Morgan to further his research on sex-linked genes and traits. (See Fig. 12-3 for assistance)

    _____
    _____
    _____
    _____
    _____
  - Sex-Linked Genes and Traits
    - Based on the results from the research data above, Morgan hypothesized that the gene for eye color in fruit flies is carried on the _____, while the Y chromosome _____ an allele for the eye-color gene
    - X-linked chromosomes are found on the _____
    - Y-linked chromosomes are found on the _____
    - Explain why there are more X-linked traits.

_____

_____

- Explain why a male who carries a recessive trait on the X chromosome will exhibit a sex-linked trait.

  _____

  _____

- Linked Genes
  - Morgan called pairs of genes that tend to be inherited together _____ _____; he called a set of these a _____ _____
  - Morgan hypothesized that genes are linked because _____ _____
  - Because mutations are rare, Morgan inferred that the process of _____ during meiosis must be responsible for the linked genes.
- Chromosome Mapping
  - What is a chromosome map? Why is it useful for scientists?

    _____

    _____

    _____

    _____
  - The crossing-over for two traits is _____ to the _____ between them on a chromosome

<u>Complete the following: Sex-Linked Punnett Square Practice</u>

1. A woman without hemophilia marries a man with hemophilia. They have a daughter with hemophilia. What is the genotype of the mother and father?

2. A colorblind man marries a female who is a carrier for colorblindness. What is the probability that they will have a child who is colorblind?

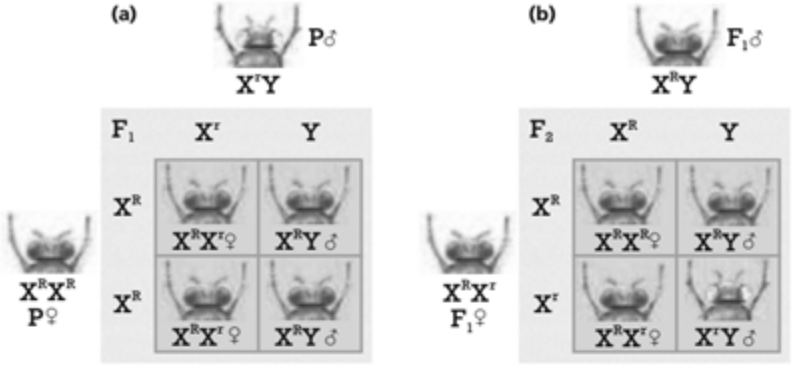


FIGURE 12-3

Eye color is a sex-linked trait in fruit flies, as these two Punnett squares show. (a) A male (♂) with white eyes mated to a female (♀) with red eyes yields all red-eyed F_1 offspring. (b) An F_1 cross results in an F_2 generation in which all females are red-eyed, half of the males are red-eyed, and half of the males are white-eyed.

EFFECTS OF GENE LOCATION

When Morgan was doing his research with fruit flies, one of the lab members noticed that a single male fruit fly had white eyes instead of the red eyes that are normally found in the flies. Morgan crossed this white-eyed male with a normal red-eyed female, and found that all the F_1 offspring had red eyes, as shown in Figure 12-3a. This demonstrated that the red-eye trait is dominant to the white-eye trait. Morgan next crossed F_1 males with F_1 females, as shown in Figure 12-3b. The resulting F_2 generation showed the expected ratio of three red-eyed flies to one white-eyed fly. Unexpectedly, however, all of the white-eyed flies were male.

Sex-Linked Genes and Traits

Based on this surprising observation, Morgan hypothesized that the gene for eye color is carried on the X chromosome and that the Y chromosome lacks an allele for the eye-color gene. An X chromosome carries a gene for eye color, either X^R (red-eye allele) or X^r (white-eye allele). In a cross of an X^RX^R female (red-eyed) with an X^rY male (white-eyed), all of the F_1 females will be X^RX^r (red-eyed), and all of the F_1 males will be X^RY (red-eyed).

In the F_2 generation, half of the females will be X^RX^R, and the other half will be X^RX^r. Because all have the dominant allele *R*, all will be red-eyed. In the F_2 males, however, half will be X^RY (red-eyed), but the other half will be X^rY (white-eyed).

The results of these experiments showed Morgan not only that genes reside on chromosomes but also that the red eye-color gene resides on the X chromosome. Morgan called genes located on the X chromosome *X-linked genes.* He called genes on the Y chromosome, such as *SRY* in humans, *Y-linked genes.* The term **sex-linked trait** refers to a trait that is coded for by an allele on a sex chromosome. The X chromosome is much larger than the Y chromosome, so there are more X-linked than Y-linked traits. Most X-linked alleles have no homologous counterpart on the Y chromosome. Because males have only one X chromosome, a male who carries a recessive allele on the X chromosome will exhibit the sex-linked trait.

Quick Lab

Modeling Linkage

Materials Two kinds of candy, toothpicks, pencil, paper

Procedure Use two kinds of candy, each kind of which has two colors, to represent genes for two traits. Long noses are dominant over short noses. Large ears are dominant over small ears. One color of candy will represent the dominant allele, and a different color candy will represent the recessive allele. Use these materials to determine the outcome of a cross between two individuals, each heterozygous for both traits. Your teacher will tell you if the genes are linked or not.

1. Draw a Punnett square. Use the appropriate alleles to make gametes for each individual. Then place the allele combinations in each square representing the possible zygotes from that cross.
2. If your genes are linked, you must use toothpicks to link the genes together before you arrange gametes on your Punnett square.

Analysis What is the phenotypic ratio in the offspring when the genes are not linked? What is the phenotypic ratio when the genes are linked? Explain the difference.

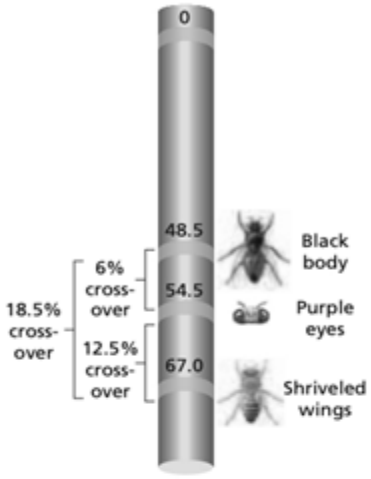


FIGURE 12-4

The cross-over frequency between the gene for black body and the gene for purple eyes is 6 percent, so these two genes are 6 map units apart. The cross-over frequency between the purple-eye gene and the shriveled-wing gene is 12.5 percent, which is equivalent to 12.5 map units. The black-body gene and the shriveled-wing gene are 18.5 map units apart.

Linked Genes

Morgan and other geneticists hypothesized that if genes are inherited together, the reason is that they occur on the same chromosome. For example, Morgan studied two fly genes—one for body color and one for wing length—located on the same autosome. Gray body, *G*, was dominant to black body, *g*, and long wings, *L*, were dominant to short wings, *l*. Morgan crossed gray-bodied, long-winged (*GGLL*) flies with black-bodied, short-winged (*ggll*) flies. All the F_1 offspring had the genotype *GgLl* and were gray with long wings.

Morgan then crossed members of the F_1 generation with one another (*GgLl* × *GgLl*) to produce an F_2 generation. The flies in the F_2 generation occurred in a phenotypic ratio of three gray, long-winged flies to one black, short-winged fly. If the alleles of the two genes had been located on different chromosomes, they would have assorted independently and produced an F_2 generation with a phenotypic ratio of 9:3:3:1 as in Mendel's peas. Morgan called pairs of genes that tend to be inherited together **linked genes,** and he called a set of linked genes a *linkage group*.

Morgan hypothesized that genes are linked because they are found on the same chromosome. An unexpected observation helped confirm this hypothesis. His F_2 crosses produced a few offspring unlike either parent, with gray bodies and short wings (*Ggll*) or black bodies and long wings (*ggLl*). Morgan realized mutations are too rare to explain all the exceptions he saw. Morgan thus inferred that the natural rearrangement process during crossing-over must be responsible. Recall that *crossing-over* is the exchange of pieces of DNA between homologous chromosomes. Crossing-over during the first division of meiosis does not create new genes or delete old ones. Instead, it rearranges allele combinations.

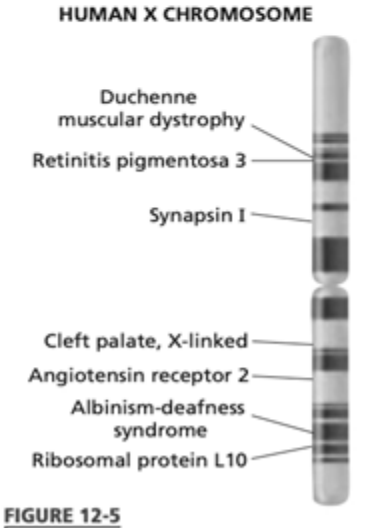


FIGURE 12-5

The locations of a few genes on the X chromosome are shown here.

Chromosome Mapping

The farther apart two genes are located on a chromosome, the more likely a cross-over will occur. The greater the percentage of F_2 offspring showing recombinant traits, the farther apart the genes for those traits must lie on a chromosome.

Researchers conduct breeding experiments and use the resulting data to prepare a chromosome map. A **chromosome map** is a diagram that shows the linear order of genes on a chromosome. Alfred H. Sturtevant, one of Morgan's students, made the first chromosome map for flies, as shown in Figure 12-4. To prepare his map, Sturtevant compared the frequency of crossing-over for several genes. The percentage of crossing-over for two traits is proportional to the distance between them on a chromosome. Sturtevant defined one **map unit** as a frequency of crossing-over of 1 percent.

Today, researchers have new techniques to map genes. A simplified map of the human X chromosome, made by using these new techniques, is shown in Figure 12-5.

Closing: Check your understanding of the lesson by answering the following question in 4-6 sentences.

1. Why is Morgan's research significant to the study of inheritance patterns and genetics? What did he observe and discover regarding what we know of chromosomes and inheritance?

_____

_____

_____

_____

_____

_____

_____

_____

_____

_____

III. Wednesday, March 25

Unit – Ch 12: Inheritance Patterns and Human Genetics
Lesson 3: Chromosomes and Inheritance (Part 3)

Objectives: Be able to do this by the end of this lesson.

1. Differentiate between chromosome mutations and gene mutations.
2. Identify types of mutations.

Introduction to Lesson 3

In Chapters 10-11, we were introduced to mutations. Recall that a mutation is a change in the nucleotide-base sequence of a gene or DNA molecule. We also know that mutations can be to the benefit of an organism, of no significance to an organism or can place an organism at a disadvantage, potentially harming it.

In today's lesson, we will be exploring the differences between mutations that can occur in body cells as well as in reproductive cells. We will also be looking into specific mutations that can occur in chromosomes and in genes.

Read pages 239-240 from your text (*found on pgs. 14-15 in your packet*). As you read, highlight the various mutations and complete the following tasks found below and on the next page. Use complete sentences the answer the questions.

GUIDED NOTES: Mutations – pgs. 239-240

- There are **three categories of mutations**:

Category	Definition/Description
Germ-cell mutation	
	⇒ Mutation that occurs in an organism's body cells ⇒ CAN affect the organism itself, but CANNOT be inherited in offspring ⇒ Examples: human skin cancer, leukemia
Lethal mutations	

  - How do some beneficial mutations give an organism and evolutionary advantage?

    _____

    _____

    _____

- Within the three categories, mutations can occur as _____ mutations or as _____ mutations, or point mutations.

- ***Chromosome mutations*** involve changes in the _____ of a chromosome or the _____ or _____ of a chromosome.
  - **Types of chromosome mutations** (See Figure 12-6 for visuals):

Chromosome Mutation	Description
Deletion	
	⇒ A chromosomal segment breaks off, flips around backward, and reattaches
Translocation	
Nondisjunction	

  - Down syndrome is a result of a chromosome mutation. What type of chromosome mutation causes Down syndrome and how Down syndrome occurs in an individual?

_____
_____
_____
_____

- A ***point mutation***, or gene mutation, is the _____, _____, or _____ of a single nucleotide within a _____ gene or other segment of DNA on a chromosome.
  - **Types of point mutations** (See Figure 12-7 for visuals):

Point Mutation	Description/Result of…
Substitution	
Frameshift mutation	
Insertion mutation	

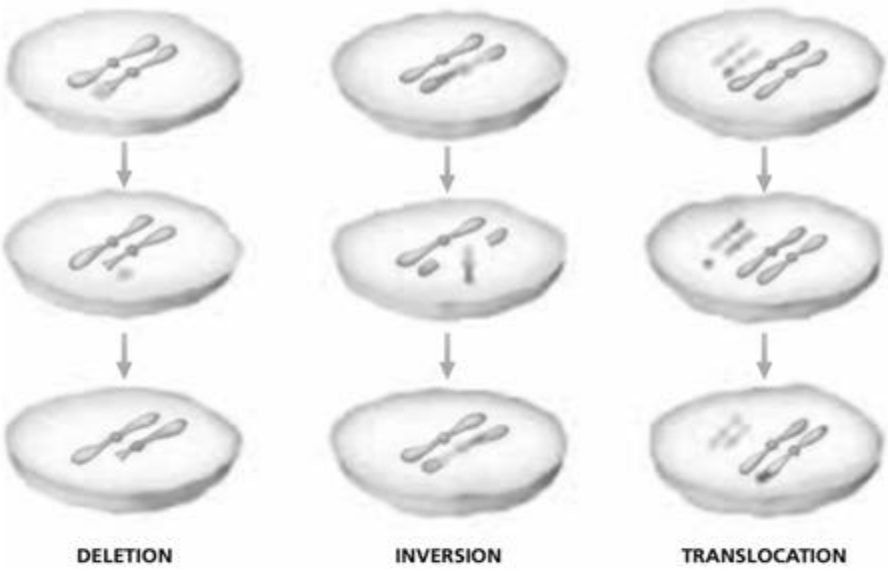


FIGURE 12-6
In some types of mutations, chromosomes break. In a deletion, a piece of a chromosome is lost. In an inversion, a piece flips and reattaches. In a translocation, a broken piece attaches to a nonhomologous chromosome.

MUTATIONS

Cystic fibrosis results from a mutation. A *mutation* is a change in the nucleotide-base sequence of a gene or DNA molecule. **Germ-cell mutations** occur in an organism's gametes. Germ-cell mutations do not affect the organism itself, but they can be passed on to offspring. **Somatic-cell** (soh-MAT-ik SEL) **mutations** take place in an organism's body cells and can therefore affect the organism. For example, certain types of human skin cancer and leukemia result from somatic-cell mutations. Somatic-cell mutations cannot be inherited.

Lethal mutations cause death, often before birth. Some mutations, however, result in phenotypes that are beneficial to the individual. Organisms with beneficial mutations have a better chance of surviving and reproducing, and therefore have an evolutionary advantage. Mutations provide the variation upon which natural selection acts. Mutations can involve an entire chromosome or a single DNA nucleotide.

Chromosome Mutations

Chromosome mutations involve changes in the structure of a chromosome or the loss or gain of a chromosome. Three types of chromosome mutations are shown in Figure 12-6. A **deletion** is the loss of a piece of a chromosome due to breakage. In an **inversion,** a chromosomal segment breaks off, flips around backward, and reattaches. In a **translocation,** a piece of one chromosome breaks off and reattaches to a nonhomologous chromosome. In **nondisjunction** (NAHN-dis-JUNGK-shuhn), a chromosome fails to separate from its homologue during meiosis. One gamete receives an extra copy of a chromosome, and another gamete receives no copies. An example of nondisjunction that results in Down syndrome is shown in Figure 12-7.

FIGURE 12-7
Some chromosome mutations are the loss or gain of entire chromosomes. The mutation that gives a person three copies of chromosome 21 results in Down Syndrome.

FOR USE DURING AT-HOME INSTRUCTION SPRING 2020 ONLY

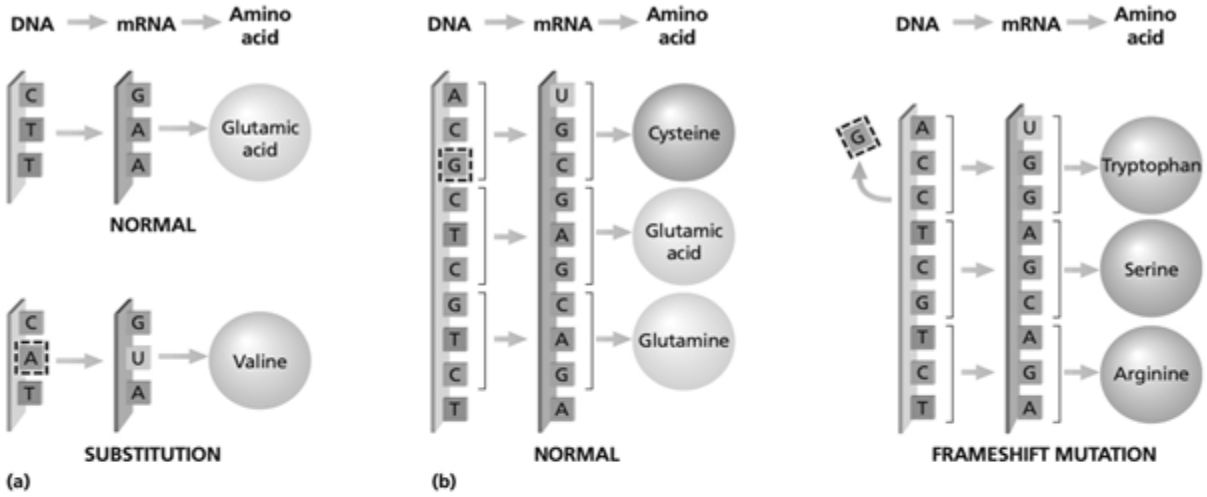


FIGURE 12-8
(a) In a substitution mutation, one nucleotide replaces another, forming a new codon that may signal the insertion of the wrong amino acid. (b) Deleting a nucleotide causes all subsequent codons to be incorrectly read, resulting in a frameshift mutation. Adding a nucleotide shifts the codon grouping too, and causes misreading.

Gene Mutations

The substitution, addition, or removal of a single nucleotide is a **point mutation,** which is a change that occurs within a single gene or other segment of DNA on a chromosome. In a **substitution,** one nucleotide replaces another, as shown in Figure 12-8a. If this substitution occurs in a codon, the amino acid can be changed. In a deletion mutation, one or more nucleotides in a gene are lost. This loss can cause incorrect grouping of the remaining codons, called a **frameshift mutation,** making all amino acids downstream change. A frameshift mutation is shown in Figure 12-8b. This mutation, in turn, can have a disastrous effect on the protein's function. In **insertion mutations,** one or more nucleotides are added to a gene, which can also result in a frameshift mutation.

Practice/Closing: Check your understanding of the lesson by completing the following:

1. Differentiate between a chromosome mutation and a point mutation. How are they alike? How are they different?

_____

_____

_____

_____

_____

_____

2. Use the Codon Amino Acid chart below to complete the practice mutation problems FIRST. Then check your work with the key found on the following page.

TABLE 10-1 *Codons in mRNA*

First base	Second base U	Second base C	Second base A	Second base G	Third base
U	UUU Phenylalanine	UCU Serine	UAU Tyrosine	UGU Cysteine	U
	UUC Phenylalanine	UCC Serine	UAC Tyrosine	UGC Cysteine	C
	UUA Leucine	UCA Serine	UAA Stop	UGA Stop	A
	UUG Leucine	UCG Serine	UAG Stop	UGG Tryptophan	G
C	CUU Leucine	CCU Proline	CAU Histidine	CGU Arginine	U
	CUC Leucine	CCC Proline	CAC Histidine	CGC Arginine	C
	CUA Leucine	CCA Proline	CAA Glutamine	CGA Arginine	A
	CUG Leucine	CCG Proline	CAG Glutamine	CGG Arginine	G
A	AUU Isolecine	ACU Threonine	AAU Asparagine	AGU Serine	U
	AUC Isolecine	ACC Threonine	AAC Asparagine	AGC Serine	C
	AUA Isolecine	ACA Threonine	AAA Lysine	AGA Arginine	A
	AUG Methionine (Start)	ACG Threonine	AAG Lysine	AGG Arginine	G
G	GUU Valine	GCU Alanine	GAU Aspartic acid	GGU Glycine	U
	GUC Valine	GCC Alanine	GAC Aspartic acid	GGC Glycine	C
	GUA Valine	GCA Alanine	GAA Glutamic acid	GGA Glycine	A
	GUG Valine	GCG Alanine	GAG Glutamic acid	GGG Glycine	G

Mutations Practice

You have read about several types of mutations:

- ➔ **DELETION** (a base is lost/deleted)
- ➔ **INSERTION** (an extra base is added/inserted)
  ---Deletion and insertion may cause what's called a *FRAMESHIFT* mutation, meaning the reading "frame" of RNA changes, thus changing the amino acid sequence from that point forward
- ➔ **SUBSTITUTION** (one base is substituted for another)

Complete the following boxes below. Classify each as **Deletion, Insertion,** or **Substitution**

Original DNA Sequence: T A C A C C T T G G C G A C G A C T…

mRNA Sequence: _____

Amino Acid Sequence:

Mutated DNA Sequence #1 T A C A T C T T G G C G A C G A C T…

What's the **mRNA** sequence?_____(*Circle the change*)

amino acid sequence? _____

Will there likely be effects? _____ What type of mutation is this? _____

Mutated DNA Sequence #2 T A C G A C C T T G G C G A C G A C T…

What's the **mRNA** sequence?_____(*Circle the change*)

amino acid sequence? _____

Will there likely be effects? _____ What type of mutation is this? _____

Mutated DNA Sequence #3 T A C A C C T T A G C G A C G A C T…

What's the **mRNA** sequence?_____(*Circle the change*)

amino acid sequence? _____

Will there likely be effects? _____ What type of mutation is this? _____

MUTATIONS PRACTICE KEY:

2. Use the Codon Amino Acid chart below to complete the practice mutation problems.

TABLE 10-1 Codons in mRNA

First base	Second base U	Second base C	Second base A	Second base G	Third base
U	UUU Phenylalanine	UCU Serine	UAU Tyrosine	UGU Cysteine	U
	UUC Phenylalanine	UCC Serine	UAC Tyrosine	UGC Cysteine	C
	UUA Leucine	UCA Serine	UAA Stop	UGA Stop	A
	UUG Leucine	UCG Serine	UAG Stop	UGG Tryptophan	G
C	CUU Leucine	CCU Proline	CAU Histidine	CGU Arginine	U
	CUC Leucine	CCC Proline	CAC Histidine	CGC Arginine	C
	CUA Leucine	CCA Proline	CAA Glutamine	CGA Arginine	A
	CUG Leucine	CCG Proline	CAG Glutamine	CGG Arginine	G
A	AUU Isolecine	ACU Threonine	AAU Asparagine	AGU Serine	U
	AUC Isolecine	ACC Threonine	AAC Asparagine	AGC Serine	C
	AUA Isolecine	ACA Threonine	AAA Lysine	AGA Arginine	A
	AUG Methionine (Start)	ACG Threonine	AAG Lysine	AGG Arginine	G
G	GUU Valine	GCU Alanine	GAU Aspartic acid	GGU Glycine	U
	GUC Valine	GCC Alanine	GAC Aspartic acid	GGC Glycine	C
	GUA Valine	GCA Alanine	GAA Glutamic acid	GGA Glycine	A
	GUG Valine	GCG Alanine	GAG Glutamic acid	GGG Glycine	G

Mutations Practice

You have read about several types of mutations:

- ➔ **DELETION** (a base is lost/deleted)
- ➔ **INSERTION** (an extra base is added/inserted)
  ---Deletion and insertion may cause what's called a *FRAMESHIFT* mutation, meaning the reading "frame" of RNA changes, thus changing the amino acid sequence from that point forward
- ➔ **SUBSTITUTION** (one base is substituted for another)

Complete the following boxes below. Classify each as **Deletion, Insertion,** or **Substitution**

Original DNA Sequence: T A C A C C T T G G C G A C G A C T...
mRNA Sequence: AUG UGG AACC GCUGCUGA
Amino Acid Sequence: Methionine – Trytophan – Asparagine – Arginine – Cysteine – STOP

Mutated DNA Sequence #1 T A C A T C T T G G C G A C G A C T...
What's the mRNA sequence? AUGUAGAACCGCUGCUGA *(Circle the change)*
amino acid sequence? Methionine – STOP
Will there likely be effects? yes **What type of mutation is this?** substitution

Mutated DNA Sequence #2 T A C G A C C T T G G C G A C G A C T...
What's the mRNA sequence? AUGCUGGAA CCGCUGCUGA *(Circle the change)*
amino acid sequence? Methionine – Leucine – Glutamic Acid – Proline – Leucine – Leucine...
Will there likely be effects? yes **What type of mutation is this?** insertion

Mutated DNA Sequence #3 T A C A C C T T A G C G A C G A C T...
What's the mRNA sequence? AUGUGGAAUCGCUGCUGA *(Circle the change)*
amino acid sequence? Methionine – Tryptophan – Asparagine – Arginine – Cysteine – STOP
Will there likely be effects? no **What type of mutation is this?** substitution

IV. Thursday, March 26

Unit – Ch 12: Inheritance Patterns and Human Genetics
Lesson 4: Human Genetics (Part 1)

Lesson 4 Socratic Guiding Question: Keep this question in mind as you study!
What is the significance of studying pedigrees?

Objectives: Be able to do this by the end of this lesson.
1. Identify the significance of using pedigrees to determine genetic traits and disorders.
2. Identify inheritance patterns and factors affecting them.

Introduction to Lesson 4
Recall from your study of the fundamentals of genetics that traits are inherited by offspring from parents. Each of these traits is a combination of two or more alleles to form the gene for that given trait. Alleles can be dominant or recessive, and genes can be homozygous (consisting of two of the same alleles for a trait, such as TT or tt) or heterozygous (consisting of two different alleles for a trait, such as Tt).
In this lesson, we will discover how the use pedigrees, or patterns of inherited traits within generations of families, help scientists better understand how traits are passed from one generation to the next, what genes control specific traits, and how these genes control traits.

Read and annotate pages 241-245 from your text (*found on pgs. 21-25 in this packet*). After reading and annotating, complete the guided outline and questions below.

GUIDED OUTLINE: Human Genetics (pp. 241-245)

- Human Genetics
  - Inheritance of Traits
    - Geneticists can study human genetic traits and trace genetic diseases/disorders through the generations by studying the phenotypes of family members in a **pedigree**
    - Pedigrees
      - Define **pedigree** – _____
        _____
      - Fill in the following key that is typical for a pedigree:
        - Squares – _____
        - Circles – _____
        - Filled symbol – _____
        - Empty symbol – _____
        - Horizontal line – _____
        - Vertical line – _____

      Why do you think pedigrees are helpful?
      _____
      _____

- Patterns of Inheritance
  - Complete the following statements:
    - If a trait is autosomal, _____ _____.
    - If a trait is sex-linked, _____ _____.
  - A **carrier** is an individual that has _____ copy of the _____ allele but does not have the _____.

➢ Genetic Traits and Disorders
- Genetic disorders are diseases or disabling conditions that have a _____ _____.
- Polygenic Inheritance
  - Most human characteristics are _____ characters, meaning they are influenced by _____ genes
  - List four examples of human characteristics that are influenced by polygenic characters: _____
- **Complex Characters** – characters that are _____ strongly both by the _____ and by _____.
  - Identify two human characteristics that are considered complex characters. Why are they considered complex characters? _____ _____ _____ _____
  - Biologists hope that by identifying environmental components that contribute to a disease, they can educate people in ways that minimize their risk of developing the disease.
- Multiple Alleles
  - Define the following:
    - **Multiple alleles** – _____ _____
    - **Codominance** – _____ _____
  - Blood types are controlled by multiple alleles in humans: I^A, I^B, and _____.

- **<u>Incomplete Dominance</u>** occurs when _____

  _____

- X-Linked Traits
  - Identify an X-linked trait in humans: _____

    Why are these traits typically seen more commonly in males?

    _____

    _____

    _____
- **<u>Sex-Influenced Traits</u>**
  - _____ and females can show different _____ even when they share the same genotype.
  - *Example*: Pattern baldness – due to higher levels of testosterone, males experience pattern baldness more than women
- Single-Allele Traits
  - A single allele of a gene controls single-allele traits
  - More than _____ human traits have been discovered to be governed by single _____ alleles.

Closing: Check your understanding of the lesson by completing the following question.

1. What is the significance of genetic study through the use of pedigrees? (3-5 sentences)

_____

_____

_____

_____

_____

_____

_____

_____

HUMAN GENETICS

This section investigates how geneticists analyze genetic data from families to track the inheritance of human genes. It also explores the genetic and environmental factors that influence human genetic traits and disorders, and discusses how geneticists detect and treat human genetic disorders.

INHERITANCE OF TRAITS

Geneticists can study human genetic traits and trace genetic diseases from one generation to the next by studying the phenotypes of family members in a pedigree.

Pedigrees

A **pedigree** is a diagram that shows how a trait is inherited over several generations. Figure 12-9 is a pedigree of a family that has several cases of cystic fibrosis. In a pedigree, squares stand for males and circles stand for females. A filled symbol means that the person has the trait or condition. An empty symbol means that the person does not have the trait or condition. A horizontal line joining a male and female indicates a mating. A vertical line indicates offspring arranged from left to right in order of their birth. Roman numerals label different generations.

SECTION 2

OBJECTIVES

- **Analyze** pedigrees to determine how genetic traits and genetic disorders are inherited.
- **Summarize** the different patterns of inheritance seen in genetic traits and genetic disorders.
- **Explain** the inheritance of ABO blood groups.
- **Compare** sex-linked traits with sex-influenced traits.
- **Explain** how geneticists can detect and treat genetic disorders.

VOCABULARY

pedigree
carrier
genetic disorder
polygenic
complex character
multiple allele
codominance
incomplete dominance
sex-influenced trait
Huntington's disease
amniocentesis
chorionic villi sampling
genetic counseling
gene therapy

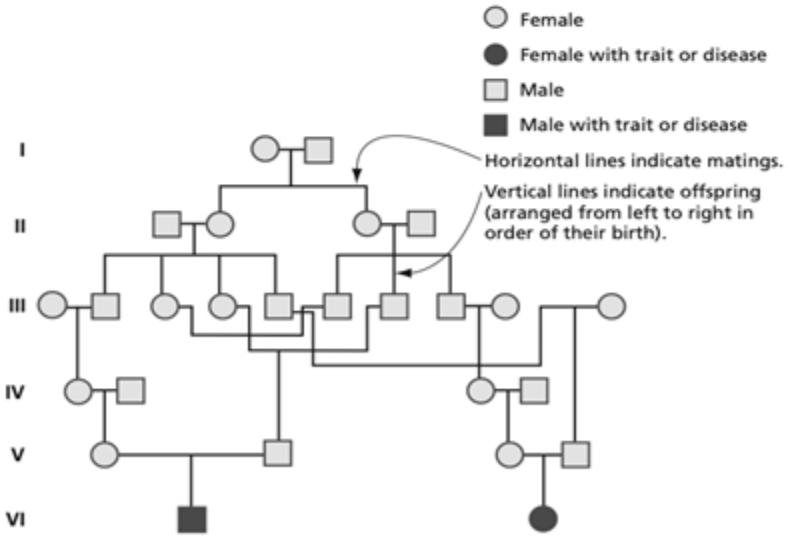


FIGURE 12-9
This pedigree for cystic fibrosis (CF) shows that each of the two affected individuals in the sixth generation has unaffected parents. Note that a cystic fibrosis allele from the first generation passed unexpressed through the next four generations. Marriages within the family during those four generations resulted in affected individuals who had two copies of the recessive disease allele for the CF gene.

Patterns of Inheritance

Biologists learn about genetic diseases by analyzing *patterns of inheritance,* the expression of genes over generations. Pedigrees help to interpret patterns of inheritance. For example, if a trait is autosomal, it will appear in both sexes equally. If a trait is sex-linked, it is usually seen only in males. Most sex-linked traits are recessive.

If a trait is autosomal dominant, every individual with the trait will have a parent with the trait. If the trait is recessive, an individual with the trait can have one, two, or neither parent exhibit the trait.

If individuals with autosomal traits are homozygous dominant or heterozygous, their phenotype will show the dominant characteristic. If individuals are homozygous recessive, their phenotype will show the recessive characteristic. Two people who are heterozygous carriers of a recessive mutation will not show the mutation, but they can produce children who are homozygous for the recessive allele.

The pedigree in Figure 12-9 shows that the condition of cystic fibrosis is inherited as an autosomal recessive. Individuals such as the four people in the fifth generation in the pedigree are called **carriers** because they have one copy of the recessive allele but do not have the disease. Although carriers do not express the recessive allele, they can pass it to their offspring.

Word Roots and Origins

polygenic

from the Greek *poly,* meaning "many," and the Greek *genesis,* meaning "origin"

GENETIC TRAITS AND DISORDERS

Genes controlling human traits show many patterns of inheritance. Some of these genes cause genetic disorders. **Genetic disorders** are diseases or disabling conditions that have a genetic basis.

Polygenic Inheritance

Single genes having two or more alleles can determine traits, such as blood type or cystic fibrosis. Geneticists have learned, however, that most human characteristics are **polygenic** (PAHL-ee-JEHN-ik) characters: they are influenced by several genes. Polygenic characters show many degrees of variation, as seen in Figure 12-10. Skin color, for example, results from the additive effects of three to six genes. These genes control the amount of the brownish-black pigment called *melanin* in the skin. The more melanin skin cells produce, the darker the skin. Each of three to six genes has an allele that produces low amounts of melanin and another allele that makes high amounts of melanin. The final amount of melanin in a person's skin that is not exposed to sunlight comes from the number of high-melanin alleles among these few skin-color genes. Eye color, height, and hair color are also polygenic characters.

FIGURE 12-10
Many characters, such as height, weight, hair color, and skin color, are polygenic. Often, the environment strongly influences polygenic characters.

Complex Characters

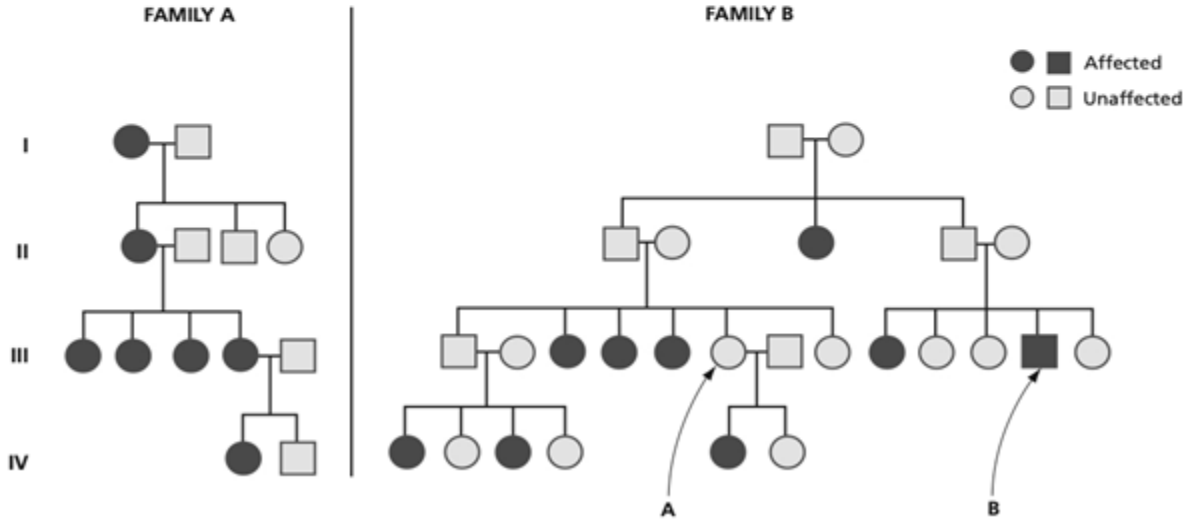
Many human conditions are **complex characters**—characters that are influenced strongly both by the environment and by genes. Skin color is both polygenic and complex. Exposure to sunlight generally causes the skin to become darker, no matter what the skin-color genotype is. Human height is another polygenic character that is controlled by an unknown number of genes that influence the growth of the skeleton. Height, however, is also influenced by environmental factors, such as nutrition and disease.

Other complex characters play a role in diseases or conditions such as breast cancer, diabetes, heart disease, stroke, and schizophrenia. Most breast cancer, for example, occurs in people with no familial history of the disease. But breast cancer also runs in some families.

Geneticist Marie-Claire King studied the genetics of breast cancer in families in which several individuals had the disease at younger ages than the average breast cancer patient does. Figure 12-11 shows the pedigrees of two of the families she studied. In Family A, each affected person has an affected parent, which is the inheritance pattern of a dominant trait. Family B demonstrates that additional genetic and environmental factors can influence whether or not a person expresses a trait. In Family B, a female, individual III-A, does not develop breast cancer herself but has a child who goes on to develop breast cancer. Notice also, individual III-B in Family B is a male who has breast cancer. This again shows that many factors in addition to the central gene influence the onset of a genetic disease.

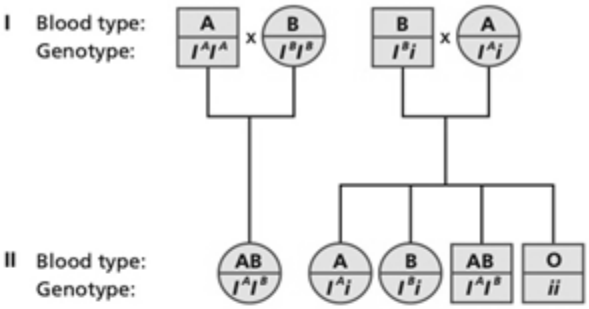
Biologists hope that by identifying the environmental components that contribute to a disease, they can educate people in ways that minimize their risk of developing the disease. For breast cancer, for example, non-genetic risk factors include a diet high in saturated fat.

FIGURE 12-11
These diagrams show the pedigrees of families in which there is hereditary breast cancer.



Phenotype		
Blood type	**Molecule on red blood cell surface**	**Genotype**
A		I^AI^A or I^Ai
B		I^BI^B or I^Bi
AB		I^AI^B
O	Neither A nor B	ii

(a) Blood Types



(b) Inheritance of Blood Type Alleles

FIGURE 12-12
The ABO gene has three alleles (a). Allele I^A causes a type A sugar to appear on the surface of red blood cells. Allele I^B, causes red blood cells to display a type B sugar. The third allele, *i*, does not cause the display of any sugars. In (b), a pedigree shows two examples of how blood type can be inherited.

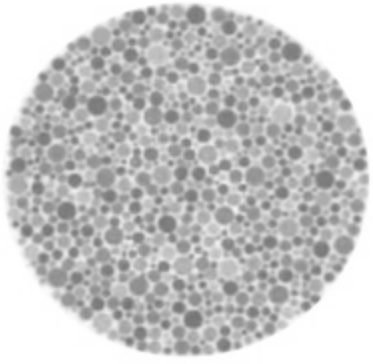
Multiple Alleles

Many genes have more than three alleles. Genes with three or more alleles are said to have **multiple alleles.** For example, in humans, the ABO blood groups (blood types) are controlled by the three alleles I^A, I^B, and i. The alleles I^A and I^B are codominant. In **codominance,** both alleles are expressed in the phenotype of a heterozygote. Both I^A and I^B are dominant to the recessive i allele. The I^A and I^B alleles encode variants of an enzyme that cause two different sugar molecules to appear on the surface of red blood cells. The i allele lacks the activity of the enzyme entirely, so neither of the sugars appear on the red blood cell surface. Figure 12-12a shows how combinations of the three different alleles can produce four different blood types—A, B, AB, and O. Notice that a person who inherits two i alleles has type O blood. Figure 12-12b shows how blood type can be inherited.

Incomplete Dominance

Sometimes, an individual displays a trait that is intermediate between the two parents, a condition known as **incomplete dominance.** For example, in Caucasians, the child of a straight-haired parent and a curly-haired parent would have wavy hair. Straight hair and curly hair are homozygous traits. Wavy hair is heterozygous and is intermediate between straight and curly hair.

FIGURE 12-13
A person who has a red-green colorblindness might not be able to see the number 5 in the center of the circle in this color-vision test chart.



X-Linked Traits

Some complex characters are determined by X-linked genes, and a pedigree will usually reveal many affected males and no affected females. A male inherits his X chromosome from his mother. One form of *colorblindness* is a recessive X-linked disorder in which an individual cannot distinguish certain colors, such as red and green. Several X-linked genes encode proteins that absorb red or green light in the eye. Red-green colorblindness occurs because mutations disrupt these genes, so the eye cannot absorb some colors of light. Eye doctors often test for colorblindness by using a chart similar to the one in Figure 12-13.

Sex-Influenced Traits

Sex-influenced traits are involved in other complex characters. Males and females can show different phenotypes even when they share the same genotype. Sex-influenced traits are usually autosomal. For example, an allele that is dominant in males but recessive in females controls pattern baldness, the type of baldness usually found in men. The difference is due to higher levels of the hormone testosterone in men, which interacts with the genotype to produce pattern baldness.

Single-Allele Traits

A single allele of a gene controls single-allele traits. Geneticists have discovered that more than 200 human traits are governed by single dominant alleles. **Huntington's disease** (HD) is an autosomal dominant condition characterized by forgetfulness and irritability. It develops as an affected person reaches 30 or 40 years of age and progresses to muscle spasms, severe mental illness, and, finally, death. Because a dominant gene exists in every HD heterozygote, each affected person has at least one affected parent. Unfortunately, many HD patients have already had children by the time their symptoms appear. Direct DNA testing is beginning to allow for earlier diagnosis of the HD allele.

V. Friday, March 27

Unit – Ch 12: Inheritance Patterns and Human Genetics
Lesson 5: Human Genetics (Part 2)

Lesson 4 Socratic Guiding Question: Keep this question in mind as you study!
What might be the advantages and the disadvantages of being able to detect genetic disorders in a fetus?

Objectives: Be able to do this by the end of this lesson.
1. Identify and describe genetic disorders.
2. Identify and describe types of genetic disorder treatment.

Introduction to Lesson 4
As a result of mutations, inherited or not, individuals may be susceptible to developing genetic disorders and diseases. Knowing a person's genetic makeup can help to determine their probability of passing on genetic disorders to offspring or of developing a disease. Treatments for these genetic disorders and diseases vary based on the severity of the symptoms and the conditions associated with the disorder or disease.

Read pages 245-248 from your text (*found on pgs. 27-30 in this packet*). After reading complete the guided outline and questions below.

- Detecting Genetic Disease
  - What is amniocentesis? How is it useful?

    _____

    _____

    _____

  - Using Table 12-1 on p. 246, list three examples of genetic disorders and describe the mutations involved in them.
    Disorder: _____
    Mutation Involved: _____

    _____

    Disorder: _____
    Mutation Involved: _____

    _____

    Disorder: _____
    Mutation Involved: _____

    _____

  - **Genetic Counseling** – the process of informing a person or couple of their _____ _____. What might be the benefit of genetic counseling?

    _____

    _____

❖ Treating Genetic Disease

➢ Physicians can treat genetic diseases in several ways.

- Treat symptoms through diets, medication, therapies, and lifestyle changes.
- Another level of treatment - **gene therapy**, which is a _____ that places a _____ copy of a _____ in the cells of a person whose copy of the gene is _____.
  - Uses viruses by implanting the DNA into the virus to invade cells.
  - Short-term success, but requires continuing treatments
  - What is the difference between *somatic cell gene therapy* and *germ cell gene therapy*?

    _____

    _____

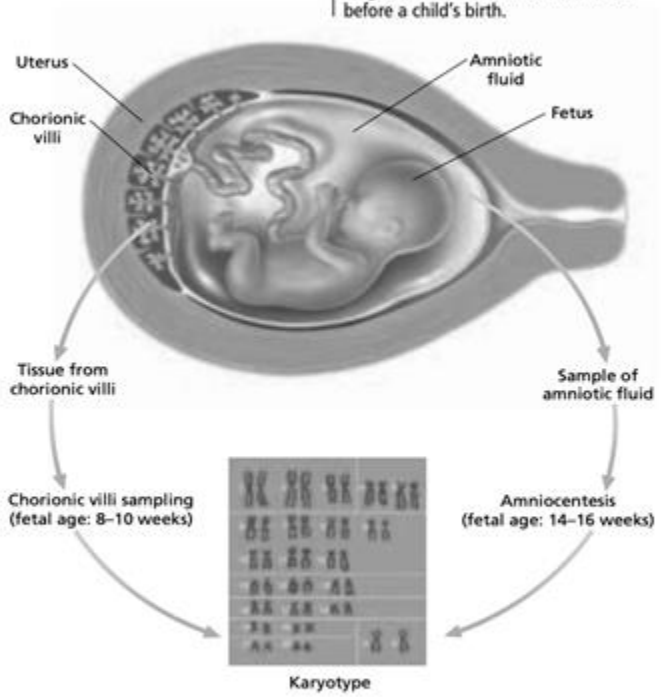
    _____

FOR USE DURING AT-HOME INSTRUCTION SPRING 2020 ONLY

DETECTING GENETIC DISEASE

Many people with a family history of genetic disease seek genetic screening before having children. *Genetic screening* is an examination of a person's genetic makeup. It may involve karyotypes, blood tests for certain proteins, or direct tests of DNA. Physicians can now also detect more than 200 genetic disorders in the fetus. The technique called **amniocentesis** (AM-nee-oh-sen-TEE-sis), shown in Figure 12-14, allows a physician to remove some amniotic fluid from the amnion, the sac that surrounds the fetus, between the 14th and 16th week of pregnancy. Geneticists can analyze fetal cells for genetic disease by examining chromosomes and proteins in the fluid.

FIGURE 12-14
Geneticists can use fetal cells obtained by amniocentesis or by chorionic villi sampling to prepare fetal karyotypes that might display chromosome mutations. This allows physicians to diagnose chromosomal abnormalities before a child's birth.



In **chorionic villi** (KAWR-ee-AHN-ik VIL-ie) **sampling,** also shown in Figure 12-14, the physician takes a sample of the chorionic villi—cells derived from the zygote that grow between the mother's uterus and the placenta—between the 8th and 10th week. Both procedures allow technicians to analyze fetal cells, chromosomes, proteins and detect genetic disease.

Table 12-1 lists some important genetic disorders, symptoms, and their patterns of inheritance.

TABLE 12-1 ***Some Important Genetic Disorders***

Disorder (Gene)	Symptom	Normal protein; function; effect	Pattern of inheritance and chromosome location	Frequency among human births
Huntington's disease (gene HD)	gradual deterioration of brain tissue in middle age; shortened life expectancy	huntingtin protein; involved in movement of vesicles in nerve cells; mutation causes extra copies of the codon CAG in the gene	autosomal dominant on chromosome 4	1 in 10,000
Cystic fibrosis (gene CFTR)	mucus clogs lungs and pancreas; victims today live to early adulthood or longer	cystic fibrosis transmembrane conductance regulator; regulates the transport of chloride ions in epithelial cells	autosomal recessive on chromosome 7	Up to 1 in 900 French Canadians; 1 in 2000 Europeans
Sickle cell anemia (gene HBB)	organ damage due to impaired blood flow	beta globin; carries oxygen in blood; mutation causes red blood cells to change shape and clog capillaries	autosomal recessive on chromosome 11	1 in 500 African Americans
Tay-Sachs disease (gene HEXA)	deterioration of central nervous system in infancy; death in early childhood	hexosaminidase A; breaks down cellular wastes in lysosome; mutation allows waste buildup, causing nerve-cell death	autosomal recessive on chromosome 15	1 in 600 Jews of European descent
Phenylketonuria (gene PAH)	infant brain fails to develop normally; death in childhood	phenylalanine hydroxylase; catalyzes change of the amino acid phenylalanine to tyrosine; without the enzyme, a toxic substance accumulates	autosomal recessive on chromosome 12	1 in 18,000 Americans
Marfan syndrome (gene FBN1)	long limbs, loose joints, deformed vertebral column, crowded teeth, rupture of large arteries	fibrillin-1; a major component of connective tissue; lack of this protein causes weakness of ligaments and blood-vessel sheaths	autosomal dominant on chromosome 15	1 in 20,000 Americans
Breast cancer (gene BRCA1)	malignant tumors in breast tissue	breast cancer-1; inhibits growth of breast and ovarian tumors, probably by encouraging repair of DNA damage	autosomal dominant on chromosome 17	About 8 percent of breast cancer patients
Hemophilia (gene F8)	prolonged bleeding due to ineffective blood clotting	coagulation factor 8; helps cause blood to clot; the mutant protein does not function in clotting	X-linked recessive on chromosome X	1 in 7,000

Careers in BIOLOGY

Genetic Counselor

Job Description
A genetic counselor is a health professional who has a specialized graduate degree and experience in the areas of medical genetics and counseling.

Focus On a Genetic Counselor
Robin Bennett is a senior genetic counselor and clinic manager at the Medical Genetics Clinic of the University of Washington. Individuals and couples who seek genetic counseling include people whose family members have birth defects or genetic disorders and people who may be at risk for an inherited condition. Clients may be expecting a child or considering parenthood, or they may have been recently diagnosed with a disease. "The whole family becomes your patient," says Bennett. At the first meeting with a client, Bennett usually draws up a pedigree, obtains medical records of affected relatives, and, in some cases, orders blood tests. "After the appointment, I send the client a written report, which helps patients and their families and healthcare providers to understand the information."

Education and Skills
- **High school**—a focus on science and math courses
- **College**—bachelor's degree, including course work in genetics, chemistry, statistics, psychology, and developmental biology; M.S. degree in genetic counseling; national certification is available
- **Skills**—strong verbal skills, emotional stability, and strong writing skills

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For more about careers, visit go.hrw.com and type in the keyword **HM6 Careers.**

Genetic Counseling

Many people with a family history of a genetic disease also undergo **genetic counseling,** the process of informing a person or couple about their genetic makeup. Genetic counseling is a form of medical guidance that informs individuals about problems that might affect their offspring. By studying the data from genetic screening tests and the family's pedigree, a genetic counselor can predict the likelihood that a couple will produce an affected child. For diseases that have both genetic and environmental influences, such as diabetes, physicians and counselors can advise families on how to lower risk factors.

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www.scilinks.org
Topic: Genetic Diseases, Screening, Counseling
Keyword: HM60651
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TREATING GENETIC DISEASE

Physicians can treat genetic diseases in several ways. For many diseases they can treat just the symptoms. For example, an individual with the genetic disease phenylketonuria (PKU) lacks an enzyme that converts the amino acid phenylalanine into the amino acid tyrosine. Phenylalanine builds up in the body and causes severe mental retardation. Physicians prescribe strict food regimens for phenylketonuria (PKU) patients to eliminate the amino acid phenylalanine from their diets. PKU can be detected by means of a blood test administered to infants during the first few days of life.

FIGURE 12-15
Jeff Pinard discusses with a colleague the results of his DNA analyses of cystic fibrosis.

For cystic fibrosis patients, physicians prescribe 45-minute-long sessions of pounding on the back and chest to dislodge sticky mucus.

For some diseases, physicians can implement symptom-prevention measures. For example, a physician might prescribe insulin injections to patients with diabetes. For patients with hemophilia, a doctor might prescribe injections of a missing blood-clotting protein. Physicians can even do some types of surgery to correct genetic defects in a fetus before birth.

Gene Therapy

Another level of treatment currently in development involves replacing the defective gene. This type of therapy, called **gene therapy,** is a technique that places a healthy copy of a gene into the cells of a person whose copy of the gene is defective. Gene therapy relies on knowing gene sequences like the one Pinard is viewing in Figure 12-15. Medical researchers place a functional allele of the gene, such as the *CFTR* gene, into the DNA of a virus. They then introduce the modified virus into a patient's lungs where the virus infects the cells and brings along the functional gene. This improves the patient's symptoms, but only until the infected cells slough off. Then the patient must undergo the procedure again. Researchers are working to increase the effectiveness of gene therapy.

Gene therapy, in which only body cells are altered, is called *somatic cell gene therapy*. This contrasts with *germ cell gene therapy,* the attempt to alter eggs or sperm. Bioethicists, who study ethical issues in biological research, generally view somatic cell gene therapy as an extension of normal medicine to improve patients' health. Germ cell gene therapy, however, poses more risks and ethical issues because future generations could be affected in unpredictable ways.

SECTION 2 REVIEW

1. A husband and wife have a son with cystic fibrosis. Their second child, a daughter, does not. Prepare a pedigree for this family.
2. Explain the difference between a polygenic character and a complex character.
3. A husband and wife have the ABO blood group genotypes I^AI^B and *ii*. What ABO blood types can their children have?
4. Use Table 12-1 to compare Huntington's disease with sickle cell anemia.
5. Describe the methods physicians can use to detect genetic diseases in an unborn fetus.

CRITICAL THINKING

6. **Predicting Patterns** A woman with cystic fibrosis marries a man who is heterozygous for cystic fibrosis. What is the likelihood that their children will have cystic fibrosis?
7. **Applying Information** Why is colorblindness less common among females?
8. **Analyzing Patterns** A man with blood type B marries a woman with blood type A. Their first child is blood type O. What is the probability their next child will be blood type AB? blood type B?

The following two pages contain your minor assessment for the week and <u>should be completed on Friday</u> only <u>AFTER</u> you have completed all previous work in this packet. You may use your notes in this packet. It should take you approximately 12-15 minutes.

Minor Assessment: Chapter 12 – Inheritance Patterns and Human Genetics

***Directions*: Complete the following tasks. You may use your notes from this packet only.**

1. The two kinds of chromosomes in general are _____ chromosomes and _____.

2. Humans with the chromosomes XX are _____ and XY are _____.

3. A _____-cell mutation would only affect an organism during its lifetime.

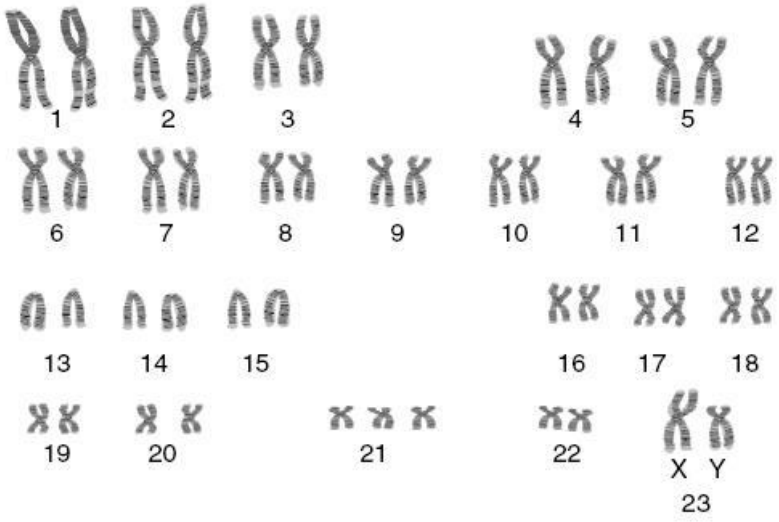
4. A _____-cell mutation would only affect an organism's offspring.

5. The diagram represents the chromosomes of a person with a genetic disorder caused by nondisjunction. Which chromosome set displays nondisjunction? _____

How do you know?

_____

_____



6. Why are X-linked traits usually more commonly expressed in males?

_____

_____

_____

_____

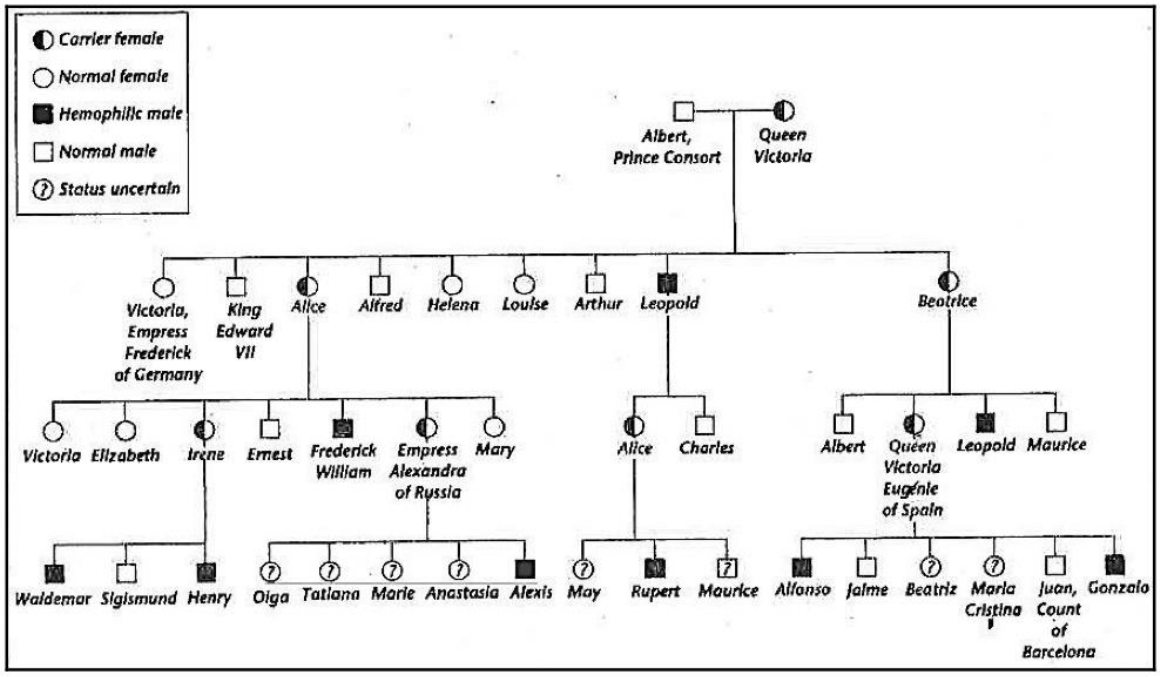
7. What are the benefits making and studying pedigrees? (Provide at least two.)

_____

_____

_____

Pedigree Practice with the Royal Family



Queen Victoria of England ruled the United Kingdom from 1837 until 1901. She and her husband had 9 children who married into other royal families around the world. She introduced the mutant gene for hemophilia into the royal family. **Hemophilia** is a genetic disorder that makes it hard for a person's blood to clot. This means a person with hemophilia will bleed for a lot longer than a normal person when they get hurt. It is a recessive X-linked disorder.

Answer the following questions based on the information above.

1. Which of Victoria's sons had hemophilia? _____
2. List Queen Victoria's grandchildren who had hemophilia.

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3. Do any women shown above have hemophilia? _____
4. List Queen Victoria's children and grandchildren who were female carriers.

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