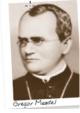
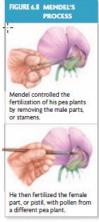
Mendel and Heredity Mendel Laid the Groundwork for Genetics



Traits are characteristics that are inherited such as eye color, leaf shape, or tail length. Scientists recognized that traits are hereditary, or passed on from one generation to the next, long before they understood how traits are passed on. **Genetics** is the study of biological inheritance patterns and variation in organisms.

<u>The study of Genetics started in the 1800's with an Austrian monk named **Gregor Mendel**. He recognized that there are separate units of inheritance – what we now call **genes** – that come from each parent. Mendel studied inheritance in pea plants.</u>

Mendel's Data Revealed Patterns of Inheritance



Three things about Mendel's experiments helped him develop his laws of inheritance:

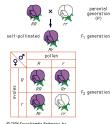
- He controlled the breeding of the pea plants he studied. Pea flowers have both male and female parts. They usually self pollinate (mates with itself). Mendel controlled the mating of his pea plants. He chose which plants to cross. In genetics the mating of two organisms is called a cross.
- 2. **He used "either-or" characteristics.** Mendel studied seven different pea traits, including flower color and pea shape. All of the characteristics he studied had only two forms, so all plants either had one form or the other.
 - **Ex:** All the flowers were purple or white. All the peas were wrinkled or round. All plants were tall or short.

3. He used purebred plants. If a line of plants self-pollinate for long enough, the plants become genetically uniform, or purebred. The offspring of a purebred parent inherits all of the parent organism's characteristics (they are identical to the parent's). Because Mendel started with purebred plants, he knew that any differences in the offspring were a result of HIS crosses.

Results

Mendel found that when he crossed purebred plants, one of the forms of a trait was hidden in the offspring. But the form would reappear in the next generation.

Mendel studied many plants and made many crosses. He found similar patterns in ALL of his results. In the figure above, you can see that the short plants disappeared in the first generation of offspring. In the second generation, though, he found that about 1/4 of the plants had the form of the trait that had disappeared in the first generation. The other 3/4 were tall plants. In other words there was a 3:1 ration of tall to short plants in the second generation. **Conclusions**



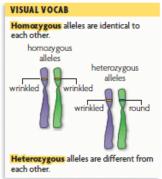
These observations helped Mendel form his first law called the **law of segregation**. There are 2 main parts to the law:

- 1. Organisms inherit two copies of each gene, one from each parent.
- 2. <u>Only ONE copy of the gene goes into an organism's gametes</u>. The two copies of a gene separate - or segregate - during gamete (egg, sperm) formation.

Vocabulary Check

- 1. The study of biological inheritance.
- 2. The mating of two organisms.
- 3. A characteristic that is inherited.
- 4. The law of segregation says that gametes receive only one chromosome from each homologous pair of chromosomes. Look back at meiosis. In which stage of meiosis do homologous chromosomes separate?f
- 5. Give two examples of traits that are NOT already mentioned.

Traits, Genes, and Alleles The same gene can have many versions.



As you learned, the units of inheritance that Mendel studied are now called **genes**. You can think of a **gene** as a piece of DNA that stores instructions to make a certain protein. Each gene is located at a particular place on a chromosome called a Locus. Just like a house has an address on a street, a gene has a locus on a chromosome. Many things come in different forms. For example, bread can be wheat, white or rye. Most genes have many forms too. An **allele** is any different form of a gene.

Ex: the gene for pea shape has two alleles - round and wrinkled.

Your cells, like the pea plant's cells, have two alleles for each gene – one on each chromosome of a homologous pair. The term **homozygous** means the two alleles of a gene are the same – **Ex:** both alleles are round, both alleles are tall. The term **heterozygous** means the two alleles of a gene are different – **Ex:** one allele is for round and one allele is wrinkled, one allele is tall and one allele is short.

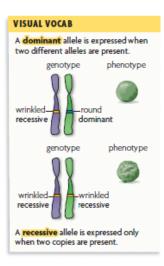
Genes influence the development of traits.

For Mendel's peas, if a plant was heterozygous for pea shape, the peas shape would be round. If it was heterozygous for height, the plant would be tall. This is because the allele for round and tall peas is **DOMINANT** or expressed when two Different alleles are present. A **recessive** allele is expressed ONLY when there are two copies of the recessive allele.

Alleles are represented with letters – CAPITAL letters for DOMINANT ALLELES and lowercase letters for recessive alleles.

Ex: the dominant allele for pea shape is written as R for Round and the recessive is r for wrinkled.

Ex: the dominant allele for height is written as T for Tall and the recessive is t for short.
A Genotype is the set of alleles an organism has for a trait. Ex: a genotype can be homozygous dominant (RR, TT), heterozygous (Rr, Tt) or homozygous recessive (rr, tt). A Phenotype is what the resulting trait looks like. Ex: Round, Tall, wrinkled or short. A Genome is all of an organism's genetic material – all the genes on all of the chromosomes.
What is the difference between a genotype and a phenotype (give examples of each).



Vocabulary Check

- 1. What is the difference between a gene and an allele?
- 2. What is the difference between a dominant allele and a recessive allele?
- 3. Fill in the table based on the information given

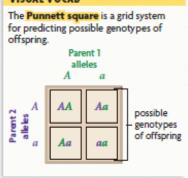
•			
	Genotype	Phenotype	Homozygous or Heterozygous
	RR		
		Round	
	Tt		
			Homozygous short
		wrinkled	

4. Which of the alleles in the table above are dominant?

Traits and Probability

Punnett Squares illustrate genetic crosses.

VISUAL VOCAB



A Punnett Square is a grid system for predicting all possible genotypes resulting from a cross. The outside edges of the grid represent the possible genotypes of gametes from each parent. The grid boxes show the possible genotypes of the offspring from those two parents.

Let's briefly review meiosis and segregation to see how Punnett squares work. Both parents have 2 alleles for each gene. These alleles are represented on the top and side of the Punnett square. During meiosis, the chromosomes – and alleles – are separated. Each gamete can receive only ONE of the alleles, but NOT both. When fertilization happens, gametes from each parent come together and for a diploid cell with two copies of each chromosome. The new cell has two alleles for each gene. This is why each box shows two alleles. One is from each parent.

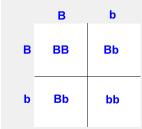
A monohybrid cross involves one trait.

Thus far, we have studied crosses of one trait. **Monohybrid crosses** are crosses that look at the inheritance of only one specific trait. **Ex: flower color, pod shape, or height**. If we know the genotypes of the parents, we can use a Punnett square to predict the genotypes of the offspring. The Punnett squares below show three different crosses.

1. Homozygous dominant crossed with homozygous recessive (RR x rr)

	R	R
r	Rr	Rr
r	Rr	Rr

2. Heterozygous crossed with heterozygous. (Bb x Bb)

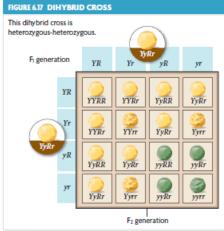


3. Heterozygous crossed with homozygous recessive. (Aa x aa)

	А	а
а	Aa	aa
а	Aa	aa

Suppose we had a purple flower pea plant but did not know its genotype. If could be Pp or PP. We could figure out its genotype by crossing the purple flower with a white flower plant. We know that the white flower plant is recessive so it's genotype is pp. If the purple flower plant is PP, all the offspring will be purple. If the purple flower plant is Pp, the half the offspring will have purple flowers and half white. Crossing a homozygous recessive organism with an organism of unknown genotype is called a **testcross**. What are the genotypes of offspring from an FF x ff cross?

A dihybrid cross involves two traits.



So far, we have examined monohybrid crosses, or crosses involving one trait. Mendel also performed **dihybrid crosses**, or crosses that look at two different traits.

Ex: Mendel crossed a purebred plant that had yellow round peas with a purebred plant that had green wrinkled peas. He wanted to see if the two traits were inherited together. The first generation offspring ALL looked the same – Yellow and Round (heterozygous). The second generation had Yellow Round, green Round, Yellow wrinkled and green wrinkled. In other words, Mendel found that pea shape and color were independent of each other – they were NOT inherited together. Mendel's Second Law of Genetics is the **Law of Independent Assortment** which states that alleles separate independently of one another during gamete formation, or meiosis. Different traits are inherited separately.

What is the difference between a monohybrid cross and a dihybrid cross?

Heredity patterns can be calculated using probability.

Probability is the likelihood, or chance, that a particular event will happen. It predicts the average number of times something happens, NOT the exact number of times.

Probability = <u>number of ways a specific event can occur</u> Number of total possible outcomes

Suppose you flip a coin. There is a $\frac{1}{2}$ chance it will land on heads, and a $\frac{1}{2}$ chance that it will land on tails. Suppose you flip two coins. For each one, the chance it will land on heads is $\frac{1}{2}$. But for both to land on heads the chance is

 $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$

These probabilities can be applied to meiosis, too. Suppose a germ cell has heterozygous alleles for a trait (Ff). A gamete has a $\frac{1}{2}$ chance of getting an F and a $\frac{1}{2}$ chance of getting an f. If two heterozygous plants are crossed what is the chance the offspring will be FF? There is $\frac{1}{2}$ chance of getting and F from mom and a $\frac{1}{2}$ chance of getting F from dad. Therefore it is $\frac{1}{2} \times \frac{1}{2}$ or $\frac{1}{4}$ chance of getting FF. Probability can be used to determine all the possible outcomes of a cross.

If you flip 2 coins, what is the probability that BOTH will land on tails?

Vocabulary Check

- 1. Crossing an organism of unknown genotype with a homozygous recessive organism.
- 2. A cross to examine one trait only.
- 3. A cross to examine two different traits.
- 4. Create a punnett square and list the genotype, phenotype and probability of each for the following: Ff x ff

Meiosis and Genetic Variation

Sexual Reproduction creates unique gene combinations.

Sexual reproduction produces a lot of variety within a species. This genetic variety comes from the events of meiosis and from the fertilization of gametes, which is random. Recall that humans have 23 pairs of chromosomes, and that each pair assorts independently from the others. As a result, there are about 8 million different combinations of chromosomes that can be produced during meiosis of one human cell.

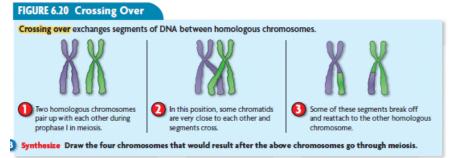
Suppose a human sperm cell that has one of 8 million different possible combinations fertilizes a human egg cell that has one of 8 million different possible combinations. Since any sperm can fertilize any egg, more than 64 trillion possible combinations can result.

For all sexually reproducing organisms, sexual reproduction results in unique combinations of the two parents' genes. Therefore, their offspring have unique phenotypes. This variety helps some organisms of a species survive and reproduce in conditions where other organisms and species cannot.

What are two parts of sexual reproduction that produce genetic variation?

Crossing over during meiosis increases genetic diversity.

Crossing over is a process that occurs during meiosis and also contributes to genetic variation. **Crossing Over** is the exchange of chromosome pieces between homologous chromosomes. This happens during prophase I of meiosis I. The process is shown below. Crossing over can happen many times – even within the same pair of homologous chromosomes.



Recall that a single chromosome has many genes, each with its own place on the chromosome. Two genes on the same chromosome may be close together or far apart. **Ex:** in the picture below, genes A is far away from genes B and C. When crossing over occurs, it is likely that A will move and B and C may be separated as well. Genes located close together tend to be inherited together which is called **genetic linkage**.



Vocabulary Check

- 1. Draw a picture that shows two chromosomes crossing over.
- 2. Draw a picture that shows genetic linkage of two genes on a chromosome.
- 3. How is genetic diversity beneficial to a species?

4. How does crossing over contribute to genetic diversity?

Chapter 6 Review

- 1. Which pair of sex chromosomes makes a person a male?
- 2. Which cells in a multicellular organism undergo meiosis?
- 3. What is the final product of meiosis?
- 4. In pea plants, the allele for tall stems, T, is dominant to the allele for short stems, t. Draw and fill in a Punnett square that shows the cross of a heterozygous plant with a homozygous dominant plant. Be sure to include genotype, phenotype and ratio in your answer.
- 5. What part of meiosis is responsible for Mendel's law of segregation?
- 6. Which human cells are haploid?
- 7. Explain in words or drawings how the processes of fertilization and crossing over contribute to genetic diversity.

Extending Mendelian Genetics

Chromosomes and Phenotype

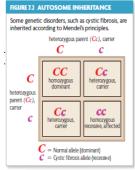
About 99.9% of everyone's DNA is identical. But look around you and you'll see a huge variety of traits such as eye color, hair color and texture, height and weight. Mendel's peas are a good place to start learning about genetics. But the great variety in living things is not just a result of dominant and recessive alleles. There are many complexities of genetic inheritance.

Two copies of each autosomal gene affect phenotype.

Recall that autosomes are all of an organism's chromosomes except the sex chromosomes. Sexually reproducing organisms have two of each chromosome, one from mom and one from dad. The two chromosomes have the same genes, but may have different alleles. Different alleles can produce different phenotypes.

Most traits are the result of genes on autosomes. Many human genetic disorders (diseases) are also caused by autosomal genes. The chance that a person will have one of these disorders can be predicted, just as we predicted the phenotypes in Mendel's peas.

Disorders caused by recessive alleles.



Two copies of the allele must be present for a person to have a disorder caused by recessive alleles. Someone who is homozygous for the recessive allele will have the disorder. Someone who is heterozygous does NOT have the disorder but is a **Carrier**. A **Carrier** is someone who does not have the disorder but Carries the recessive allele, and therefore can pass the allele on to offspring. If both parents are heterozygous, neither will have the disorder, but they can still have children with the disorder.

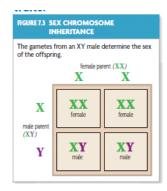
Disorders Caused by Dominant Alleles

Dominant genetic disorders are less common than recessive disorders. Because the disorder is caused by a dominant allele, there is a 50% chance that a child will have it even if only one parent has one of the alleles. If both parents are heterozygous for a dominant disorder, they both have symptoms of the disorder, and there is a 75 percent chance that a child will inherit the disorder. What is the genotype of a carrier of a recessive disorder?

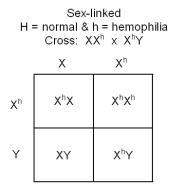
Males and Females can Differ in Sex-linked Traits

So far, we have examined the expression of autosomal genes. The expression of genes on sex chromosomes – the X and Y chromosomes – is different. Recall that for humans and other mammals, females have an XX and males have an XY genotype. A female can pass on only X chromosomes to offspring. A male can pass on either an X or a Y chromosome to offspring.

In humans and many other organisms, the Y chromosome is smaller and has many fewer genes than the X chromosome. In addition to sex determination, genes on the X chromosome affect many traits. Genes on the sex chromosomes are called **Sex-linked genes**. When people talk about sex-linked genes, they are usually talking about genes on the X chromosome.



Expression of Sex-Linked Genes in Males



Genotypic ratio: 1:1:1:1 (XⁿX =25% XⁿXⁿ=25% XY=25% XⁿY=25%)

Phenotypic ratio: 1:1:1:1 Female carrier =25% Female hemophilia =25% Male normal =25% Male hemophilia =25%

Vocabulary Check

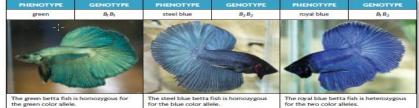
- 1. Which term describes something that happens in female mammals?
- 2. Which term describes an organism that is a heterozygote for a recessive autosomal disorder?
- 3. Which term describes a gene that will always be expressed in male mammals?
- 4. Look back at autosomal inheritance. The two parents are carriers of a recessive disorder called cystic fibrosis. What is the probability that any of the offspring of these two parents will have the disorder?
- 5. Look back at the figure with sex chromosome inheritance. Explain why a female can pass on only an X chromosome to offspring.

Complex Patterns of Inheritance

Phenotype can depend on interactions of alleles.

In many cases, alleles are not simply dominant or recessive. Alleles may interact in many different ways. For Example, alleles might have a range of dominance. There might be more than just two alleles for a gene. Or there might be many different genes that all affect one trait. **Incomplete Dominance**

Sometimes, neither allele is completely dominant or completely recessive. In this case, the heterozygous phenotype is somewhere between the two homozygous phenotypes. In other words, the alleles show **Incomplete Dominance**. One example of incomplete dominance is the flowers of the four o'clock plant. When plants are homozygous for red flowers are crossed with plants that are homozygous for white flowers, the offspring have pink flowers. Neither of the phenotypes of the parents can be seen separately in the offspring.



Codominance

FIGURE 7.	FIGURE 7.6 CODOMINANCE				
PHENOT	PHENOTYPE (BLOOD TYPE)				
A	antigen A	I ^A I ^A or I ^A i			
В	antigen B	I ^B I ^B or I ^B i			
AB	both antigens	I ^A I ^B			
0	no antigens	ii			

Sometimes, both alleles of a gene are expressed completely, and neither is dominant or recessive. In this case, alleles show **Codominance**. With incomplete dominance, recall that the heterozygous flowers were pink – a blend of the two homozygous phenotypes. Codominance is different because BOTH traits are expressed separately. The heterozygous phenotype would have some red areas and some white areas.

Human blood types are an example of codominance. Blood type is also a **Multiple Allele Trait**, because there are three different alleles. The three alleles are called IA, IB, i. BOTH IA and IB produce a protein called an antigen on the surface of red blood cells. IA and IB are codominant. Allele i is recessive and does NOT produce an antigen. Four different phenotypes are possible, shown below.

What is the difference between incomplete dominance and codominance?

Recall that when there are two copies of an allele, the expression of a recessive allele can be masked, or covered up, by the expression of a dominant allele. For an XY male, there is only ONE copy of each gene on the Y chromosome. Likewise, there is only ONE copy of each gene on the X chromosome. There are no second copies of the alleles. This means that any recessive alleles on the X or Y chromosomes will be expressed in males.

Ex: hemophilia, color-blindness, male-patterned baldness.

Expression of Sex-linked Genes in Females

All of the body cells in a female have two X chromosomes. But, each cell uses only one of the two X chromosomes. The other X chromosome gets "turned OFF". Which of the two X chromosomes remains active and which gets turned off is random. In other words, some cells use one X chromosome and other cells use the other X chromosome.

X chromosome inactivation is the process by which one of the two X chromosomes in every cell in female mammals gets turned off. Even though only one of the two alleles is expressed in each cell, overall, both alleles for each gene on the X chromosome affect a female's genotype.

Why do males have only one allele for all genes on the X chromosome?

Many genes may interact to produce one trait.

As you have seen, some phenotypes are a result of incomplete dominance, codominance and multiple alleles. But most traits in plants and animals, including humans, are the result of several genes that interact.

Polygenic Traits

Traits produced by two or more genes are called **polygenic traits**. For example, eye color and skin color are both determined by the interaction of multiple genes. At least 3 genes affect eye color. Each gene has two alleles. Scientists think that there may be even more genes that affect eye color.

Epistasis

Fur color in mice and other mammals is also a polygenic trait. In mice, at least five different genes interact to produce the fur color phenotype. One of the genes is called an eipistatic gene. This gene can prevent the expression of all of the other genes. Albinism – the lack of pigment (color) in skin, hair and eyes – is the result of epistasis. If a mouse is homozygous for the alleles that prevent pigmentation, the fur will be white no matter what alleles the mouse has for the other four genes. Albinism occurs in humans too.

What is the difference between a multiple-allele trait and a polygenic trait?

At least three different genes interact to produce the range of human eye colors, such as in the examples on the right.

GENE NAME	DOMINANT ALLELE	RECESSIVE ALLELE
BEY1	brown	blue
BEY2	brown	blue
GEY	green	blue

Order of dominance: brown > green > blue.



The environment interacts with genotype.

Phenotype is NOT determined only by genes. The environment – the conditions surrounding an organism – also affects phenotype. For Example: the sex of sea turtles depends on the temperature of the environment in which the egg develops. Female turtles make nests on beaches and bury their eggs. Eggs in warmer parts of the nest become females. Eggs in cooler parts become male.

Genes and environment also interact to determine human traits. For Example: genes influence height, but they do not completely control height. One way scientists study the interaction between genes and environment is by comparing twins raised in different environments. Identical twins can have height and size differences depending on environmental conditions such as nutrition and health care.

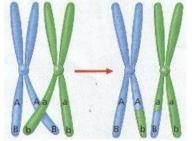
Why might genetically identical twins have different phenotypes?

Vocabulary Check

- 1. Eye color, which is determined by at least three genes, is an example of _
- 2. The flowers of a heterozygous four o'clock plant are pink, which is between flower color of each homozygous plant. This is an example of _
- 3. Human blood type is determined by a protein, called an antigen, on the surface of red blood cells. Someone with BOTH the IA and IB alleles will have both the A and B antigens on their red blood cells. This is an example of ______.
- 4. List at list three patterns of inheritance that are different than the dominant-recessive patterns of Mendel's peas.

Gene Linkage and Mapping

Gene linkage was explained through fruit flies.



You may have seen fruit flies in the kitchen, buzzing around ripe fruit. Fruit flies are also used in genetic research because they have variations that are easy to observe, for example, in eye color, body color and wing shape.

A scientist named Thomas Hunt Morgan and his students did experiments similar to Mendel's, but with fruit flies. They found that some traits were inherited together. They called the traits linked traits. They concluded that linked traits are caused by linked genes – genes that are on the same chromosome. Chromosomes, NOT genes, assort independently during meiosis. So genes on the same chromosome move together. But they also found that linked genes were not always inherited together. They concluded that this is because of crossing over between homologous chromosomes.

What are linked genes?

Linkage maps estimate distances between genes.

Recall that homologous chromosomes cross over, or exchange pieces, during meiosis. The closer together two genes are, the less likely it is that they will be separated by crossing over. The farther apart two genes are, the more likely it is that they will be separated by crossing over. One of Morgan's students kept track of the number of times that linked genes were inherited separately. In other words, he kept track of the frequency of cross-overs. He converted the percentage of cross-overs into a measurement called map units. Using this information, he made **Linkage Maps** that showed the relative location of genes on a chromosome.

Linkage maps show the relative locations of genes, but not the actual physical distances between the genes.

Why does the frequency of cross-overs give information about the relative location of genes?

Vocabulary Check

- 1. Use the following data to label the linkage map below for genes R, S, and T
 - Gene R and gene S cross over 5.0 percent of the time.
 - Gene S and Gene T cross over 10.0 percent of the time.
 - Gene R and Gene T cross over 15.0 percent of the time.
 5.0
 10.0
- 2. How were fruit flies used in genetic research?
- 3. How are linked genes different from sex linked genes?

Human Genetics and Pedigrees

Human genetics follows the patterns seen in other organisms.

Fruit flies and pea plants may seem simple. And they are certainly different than humans. But human genetics follows the same patterns of heredity. The process of meiosis happens the same way in humans as in fruit flies. Humans, like other organisms that reproduce sexually, have the same relationships between alleles: dominant-recessive interactions, polygenic traits, and sex-linked genes among others.

Although many traits are complex, single-gene traits are helpful in understanding human genetics. A downward-pointed hairline (widow's peak) is a single-gene trait with a dominant and recessive inheritance pattern. Many human genetic disorders are, too. Much of what is known about human genetics comes from studying genetic disorders.

In what ways are human genetics similar to fruit fly or pea plant genetics?

Females can carry sex-linked disorders.

Recall that some genetic disorders are caused by genes on autosomes. Both males and females can be carriers of a recessive autosomal disorder. That is, they can have one recessive allele but have no symptoms of the disorder.

In contrast, only females can be carriers of a sex-linked disorder. Recall that the X chromosome has far more genes than the Y chromosomes, they can be heterozygous and have one recessive allele, but not have symptoms of the disorder. A male has an XY genotype. A male who has an allele for a disorder located on the X chromosome will NOT have a second, normal allele to mask it.

Why can't males be carriers of sex-linked disorders?

A pedigree is a chart for tracing genes in a family.

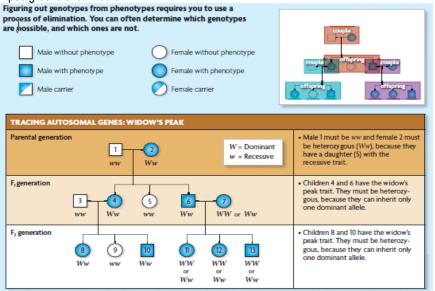
A **pedigree** chart can help trace the phenotypes and genotypes in a family to determine the chance that a child might have a certain genetic disorder. The genotypes can often be figured out using enough family phenotypes. Using phenotypes to figure out the possible genotypes in a family is like solving a puzzle. You have to use logic and clues to narrow the possibilities for each person's genotype.

Tracing Autosomal Genes

Consider the widow's peak. The widow's peak is an autosomal trait. A widow's peak is dominant to a straight hairline.

- People with a widow's peak have either homozygous dominant (WW) or heterozygous (Ww) genotypes.
- Two parents without a widow's peak are both homozygous recessive (ww) and CANNOT have children with a widow's peak.
- Two parents who both have a widow's peak can have a child who does not (ww) if both parents are heterozygous (Ww).

A pedigree is shown below.



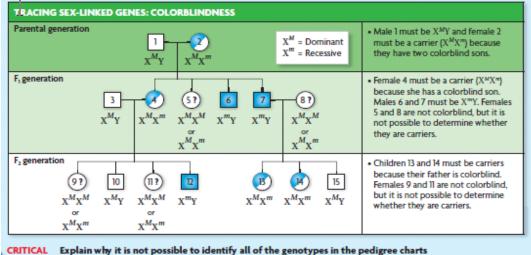
Tracing Sex-linked Genes

For sex-linked genes, you have to think about dominant and recessive alleles, but you also have to think about inheritance of the sex chromosomes. One example of a sex-linked trait is red-green colorblindness, a condition that causes a person to not be able to see the difference between some colors.

By using a process of elimination, you can often figure out the possible genotypes for a given phenotype.

- Colorblind females must be homozygous recessive (XmXm).
- Males who are colorblind must have the recessive allele (XmY)
- Females who are heterozygous for the alleles (XMXm) are not colorblind, but are carriers of the trait.

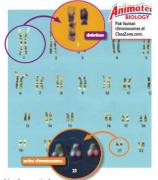
The inheritance of colorblindness is shown in the pedigree below.



VIEWING above. What information would you need to identify the genotypes of those people?

What is one difference between tracing the inheritance of autosomal traits and tracing the inheritance of sex-linked traits?

Several methods help map human chromosomes.



The human genome or all of the DNA in humans is so large that it is difficult to map human genes. In addition to pedigrees, other methods more directly study human chromosomes. A **Karyotype** is a picture of all of the chromosomes in a cell. Chemicals are used to stain the chromosomes. The stains produce a pattern of bands on the chromosomes. These patterns can be used to tell different chromosomes apart. Karyotypes can also show if there are extra or missing chromosomes, or pieces of chromosomes. Down Syndrome results from an extra copy of at least part of chromosome 21, and can be identified on a karyotype. The condition is caused by a mutation and is called Trisomy 21.

What is one example of a genetic disorder that can be seen on a karyotype?

Vocabulary Check

- 1. What is a pedigree?
- 2. What is a karvotype?
- 3. Why can the genetics of fruit flies be applied to humans?
- 4. What is the genotype of a female carrier of a sex-linked genetic disorder?
- 5. What are pedigrees used for?
- 6. What types of information can a karyotype provide?

Chapter Review

- 1. A certain disorder is recessive and sex-linked. Give two examples of genotypes that would show someone with the disorder.
- 2. A certain disorder is recessive and autosomal. Which genotype would show the trait?
 - a. RR b. Rr c. rr
- 3. Explain how the expression of genes on a sex chromosome differs from the expression of genes on an autosomal chromosome.
- 4. If two genes are linked, what does that mean about their physical location on chromosomes?
- 5. Which of the following shows the genotype of a normal human male? a. X b. YY c. XX
 - c. XX d. XY
- 6. The frequency of a certain biological process in which pieces of homologous chromosomes are exchanged is used to figure out gene linkage maps. What is the name of this process?
- 7. It is known that two fruit fly traits are linked. One trait is determined by gene A and the other by gene B. The two traits are usually, but not always, inherited together. What is the best explanation for this observation?

- a. The genes are not linked
 b. The genes are on different chromosomes
 c. the genes are far enough apart for crossing over to occur.
 d. the genes are too close for crossing over to occur.
 8. Some traits follow two-allele dominant-recessive inheritance patterns. Name at least TWO other genetic inheritance patterns.