

11

Genetics

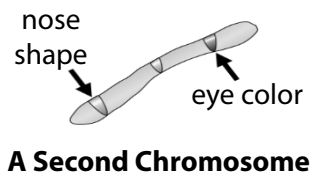
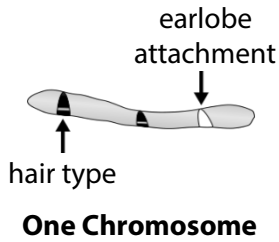


A generation before Darwin's theory of evolution, an Augustinian priest, Gregor Mendel, discovered that hereditary particles are passed from parent to offspring during the reproductive process. These particles are called *genes*, and the science of studying inheritance is called *genetics*.

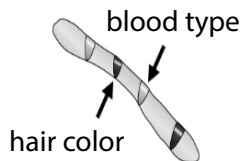
The investigation into the mechanics of inheritance—the mixing, the passing on, and the function of genes— is one of the greatest achievements in the 20th century. Genes can be traced backwards in time. When we do so, we find that all traits were new at some point in time, and that a gene's success correlates with adaptation, natural selection, and only partially understood chance events. However, genes do not last forever. Most have already gone extinct or been greatly changed. Understanding genetics has led to the prevention and treatment of several hereditary diseases. It has helped human beings to see their place in a family tree of life. In addition, genetics tells us that our genetic individuality is not the result of possessing a trait that no other individual has but is a result of a particular combination of genes. These genes came from our parents and their ancestors before them. Today's lab will explore some of the basic principles of genetics, introduce you to basic terminology, and help you apply genetic rules to some hypothetical inheritance questions.

Exercise #1	Basic Terminology
Exercise #2	Some Rules of Genetics
Exercise #3	How to Solve Genetic Problems
Exercise #4	Genetic Problems
Exercise #5	Sex-Linked Traits

Exercise #1 Basic Terminology



? Question



In order to understand the mechanics of inheritance, you must understand the terminology used to describe this very complex process. A **gene** is a segment of the DNA molecule that is responsible for manufacturing a protein. That protein either becomes part of the organism's structure or it becomes an enzyme that controls biochemical events. A second fundamental concept is the chromosome. The **chromosome** is a vehicle for moving genes around the cell during cell division. It is like a suitcase – a very unusual one. A particular chromosome of a species always carries the same genes.

Every organism has a certain number of chromosomes—the exact number depends on that species. Each chromosome carries many genes. DNA coils up into the form of a chromosome during cell division, and a gene is a distinct piece of that chromosome. Genes can be described by their exact location on a chromosome, and the process of locating genes is called **mapping**. The location of a gene is its **locus**, and geneticists go through great efforts to pinpoint these locations. Knowing where a gene is found on the chromosome is what allows scientists to start genetic research.

1. The particles that control inherited traits are called _____.
2. These particles are segments of _____, and are responsible for manufacturing a _____ that becomes either _____ or _____.
3. What is the basic purpose of a chromosome?
4. Every living thing on the planet has the same number of chromosomes. (T or F) Explain your answer.
5. Every chromosome has the same identical genes as every other chromosome. (T or F) Explain your answer.
6. The place where you find a particular gene on a chromosome is called the _____.

Homologous Chromosomes

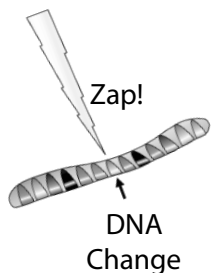
We have discussed homologous chromosomes before. This idea is essential to the understanding of genetics, so we will review it again.

- Very simple organisms have only one set of chromosomes and they are called haploid.
- More complex organisms have two sets of chromosomes and are called diploid.
- Haploid cells have one of each kind of chromosome and one of every kind of gene.
- Diploid cells have two of each kind of chromosome and two of every kind of gene.
- The two chromosomes of each kind in a diploid cell are called **homologous chromosomes** because they are carrying the same kind of traits (genes). Homo means “same.”
- A human has 23 different kinds of chromosomes that are given numbers from 1 to 23. Because we are diploid organisms, we have two of each of the different kinds. So, we have 46 chromosomes in all, made up of 23 homologous pairs.

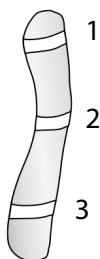
? Question

1. How many sets of DNA molecules or chromosomes does a diploid organism have?
2. How many sets of DNA molecules or chromosomes does a haploid organism have?
3. Humans are (haploid or diploid)?
4. Which of your cells are haploid?
5. How many homologous pairs of chromosomes does a human have?
6. Because chromosomes occur in pairs in a diploid organism, how many genes for one trait would a diploid organism possess?
7. How many genes for a trait would a haploid organism (or one of your gametes) possess?

Alleles: The Various Forms of a Gene

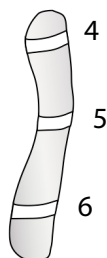


Mutation Event

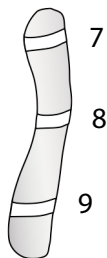


Chromosome #3

? Question



Chromosome #7



Chromosome #3

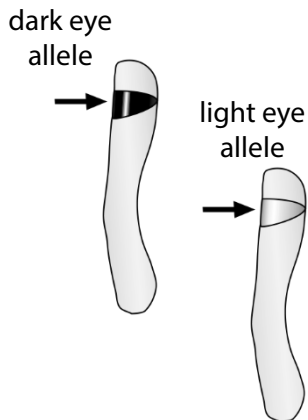
Humans are diploid, and they have two copies of every kind of gene. One of the purposes of genetics is to figure out which form (variation) of these two genes you have and what expression of those genes you can expect. The alternate forms of a particular gene are called *alleles*. For example, there are three alternate forms (three alleles) for blood type: A, B, and O.

The reason all species have various alleles (forms of genes) is that *mutation* events change the structure of genes. A gene can be mutated (changed) by radiation, by chemicals in the environment, or by other spontaneous events that are surprisingly common. There may have been a time when all the genes for eye color were identical and resulted in brown eyes. But over time, mutations occurred and changed the DNA of this eye color gene, creating a new “alleles” for the eye color trait. Perhaps this new allele was for blue eyes. Alleles are always for the same trait and are located at the exact same spot on homologous chromosomes. They have the same *locus*. This is how we know that they are truly alleles of each other and not different genes. Remember: Alleles are variations of the same gene.

1. What is an allele?
2. Where are alleles located?
3. What process creates the various alleles in a species? Explain how.
4. Which of the genes (1 through 9) on the chromosomes to the left are alleles of each other?

Genotype

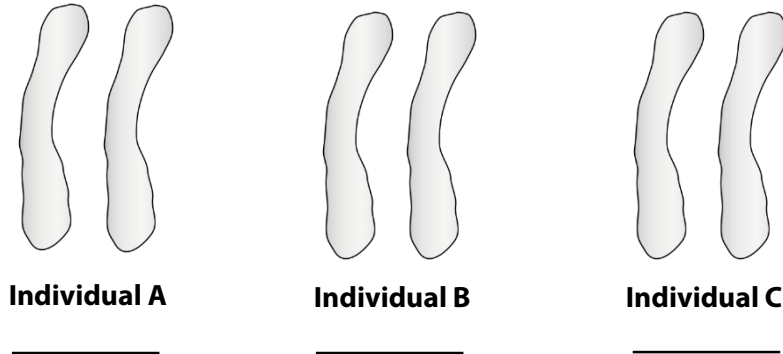
Genotype = Your Genes



Homologous Chromosomes

A **genotype** is a description of the alleles that you have for a particular trait. Even though two genes can look different, they are alleles of the same gene if they are at the same locus on homologous chromosomes.

Considering the two chromosomes on the left, draw and label the three combinations of eye color alleles that are possible.



If an organism has two identical alleles, we say it is **homozygous** for that trait (meaning the “same” two alleles). If an organism has two different alleles, we say it is **heterozygous** for that trait (meaning “different” alleles). Go back to the diagram of the three individuals above and label each as to whether it is homozygous or heterozygous.

Phenotype

Phenotype = What you look like.

The physical expression of the alleles—what an organism looks like—is called the **phenotype**. Because there are different possible combinations of alleles (genotypes), there are also different possible phenotypes for a trait that can be expressed in a population.

- The phenotype is the description of the physical expression of a trait (brown eyes), whereas the genotype is the description of the exact combination of alleles (for example; 1 allele for brown eyes + 1 allele for blue eyes).
- The genotype results from the combination of genes you inherited from your parents.
- The phenotype results from the physical expression of the genotype, and it may be influenced by the organism’s environment. In some cases there are only two phenotypes for a trait, and in other cases there are more than two phenotypes for a trait.

Draw Table 11.1 on the whiteboard, and record your phenotype for each of the six traits. After everyone in the class has recorded their phenotypes, write the class totals in Table 11.1. Your instructor will tell you the genotypes of these traits at the end of the next Exercise.

Table 11.1. Frequency of Various Traits in Lab Class.

Trait	Class Phenotype Totals		
Eye Color	*Dark =		Light =
Earlobes	Attached =		Unattached =
PTC Paper	Can Taste =		Cannot Taste =
Hairline	Widow's Peak =		Straight Forehead =
Hair Type	Straight =	Wavy =	Curly =
Fingers	Five =		Six =
Little Finger	Bent =		Straight =
Tongue	Roller =		Non-Roller =
Long Palmer Muscle	Present =		Absent =

Exercise #2 Some Rules of Genetics

Nine times out of ten, in the arts as in life, there is actually nothing to be discovered; there is only error to be exposed.

—H. L. Mencken American editor and critic (1880–1956)



**You must have the gene
to pass it on.**

Rule of the Gene

The parent must possess the gene in order to pass it on. The source of all genes in the offspring is the parents. Always look to the parents to figure out what genes the sperm or egg can possibly carry. A parent does not have all of the alleles found within a reproducing population of a species.

? Question

1. How many different alleles for a single trait can a homozygous parent pass on?
2. How many different alleles for a single trait can a heterozygous parent pass on?

Rule of Segregation

Only one gene of the two alleles that you possess is put into each gamete that you make. Alleles are located on homologous chromosomes, and since homologous chromosomes are segregated during meiosis, the genes are also segregated. Numerous gametes are formed during gamete production, and if the alleles are different (heterozygous), 50% of the gametes will carry one allele and 50% of the gametes will carry the other. When alleles are the same (homozygous), 100% of the gametes will carry the same allele.

? Question

1. A parent possesses two copies of each gene. When this parent passes on its alleles for a gene, how many does it contribute to each of the offspring?
2. How many copies of a gene does the other parent contribute to each offspring?
3. How many copies of each gene for the trait does each offspring receive?

Rule of Dominant and Recessive Alleles

Some alleles control the phenotype even if they are paired with a different allele. If two different alleles are together in an organism, and only one phenotype is expressed, then the allele that is expressed is called **dominant**. The other allele that is “hidden” is called **recessive**. One example of a dominant allele is the dark-eye allele that will create the dark-eye phenotype in an individual even if the allele for light eyes is also present.

? Question

1. Can the individual carry an allele that is not expressed? Explain.
2. What word is used to describe the genotype condition in which two different alleles occur together in the same organism?
3. What word is used to describe the genotype condition in which two of the same alleles occur together in the same organism?

Since dominance and recessiveness have intricate biochemical explanations, the only easy way of determining dominance is to cross two individuals that are homozygous (pure) for the two different phenotypes. This produces the heterozygous condition. Whichever phenotype is exclusively expressed is said to be the *dominant phenotype*.

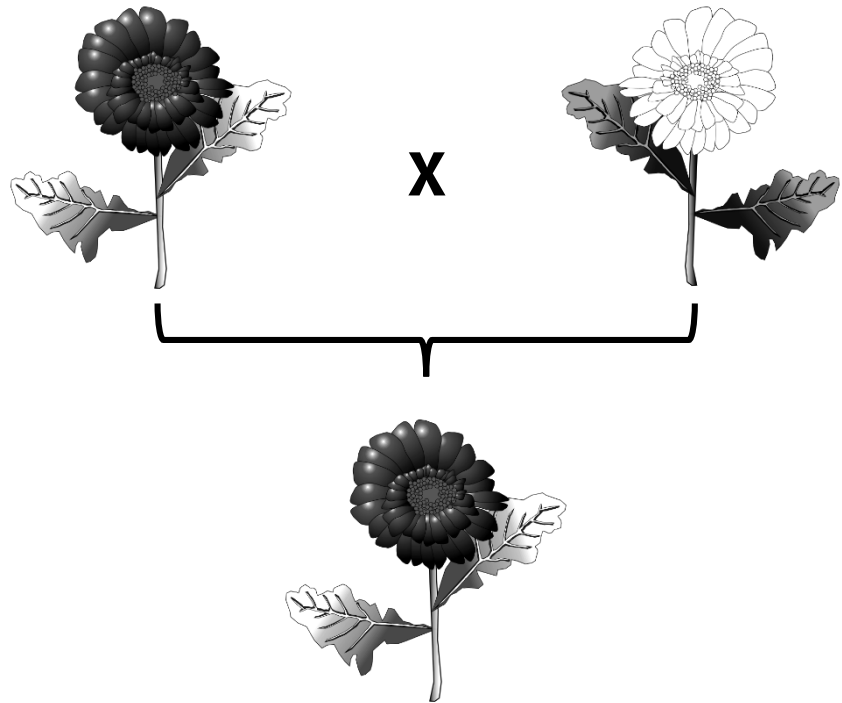


Figure 11.1. In some flowering plants when red flowers are crossed with white flowers, the result is all red flowers. This is an example of complete dominance.

? Question

1. A homozygous blue-eyed mouse with short whiskers mates with a homozygous brown-eyed mouse with long whiskers. All of their offspring have brown eyes and short whiskers. Which alleles are dominant?
2. A homozygous five-clawed cat is crossed with a homozygous six-clawed cat and all of the kittens have six claws. Which allele is dominant?
3. In humans, the five-fingered condition is recessive to the six-fingered condition. Yet, most people have five fingers. Explain how this can happen.

Dominant does not mean most common.

4. Ask your instructor which alleles are dominant in the class Phenotype Chart (Table 11.1). It is a common mistake to assume that the allele found most frequently is always the dominant allele. The success of an allele is usually determined by how successful the trait is in response to natural selection.

Rule of Incomplete Dominance

When two different pure-breeding strains are crossed, and their offspring show a blending of phenotypes, then neither allele is dominant.

Incomplete Dominance is easily recognized whenever there is a phenotype somewhere between two extremes. Including the parents, there are three phenotypes (red, white, and pink) being expressed in these flowers instead of only two, and that third phenotype is intermediate between the other two. This heterozygous condition is called ***incomplete dominance***.

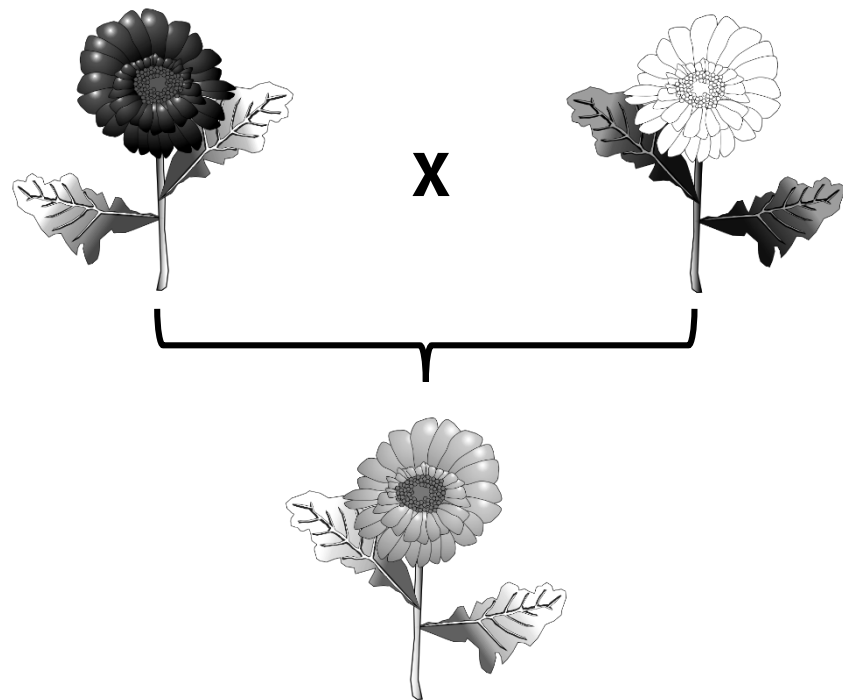


Figure 11.2. In some flowering plants when red flowers are crossed with white flowers, the result is all pink offspring. This is an example of incomplete dominance.

? Question

1. On the chart you did earlier, which of the three hair types (wavy, curly, or straight) represents incomplete dominance—the blended heterozygous condition?
2. You cross a herd of red cattle with white cattle and all of the calves appear to be roan (reddish white). Is this an example of incomplete dominance? How do you know?
3. You cross a blue flowering pea plant with a white flowering pea plant and all of the offspring are blue flowered. Is this an example of incomplete dominance? How do you know?

Exercise #3
How to Solve Genetic Problems

Using Letters for Alleles

For convenience, the genes of an allele pair are usually symbolized by a letter from the alphabet. A **Capital letter is used for the dominant trait** and **a small letter for the recessive trait**. When we want to describe the genotype of an organism, we use letters to represent the alleles inherited from the parents. For example, free earlobes is a dominant allele and attached earlobes is recessive. You would use a capital “F” to indicate the dominant allele and a small “f” to indicate the recessive allele in describing an individual.

? Question

1. Write the three genotypes for earlobe attachment as it applies to the following individuals.
 - a. Heterozygous _____
 - b. Homozygous Dominant _____
 - c. Homozygous Recessive _____
2. When it comes to symbolizing incomplete dominance with letters, it is best to use the letter “C” for one allele and “c” for the other allele. List the three possible genotypes for hair type.
 - a. Curly _____
 - b. Wavy _____
 - c. Straight _____

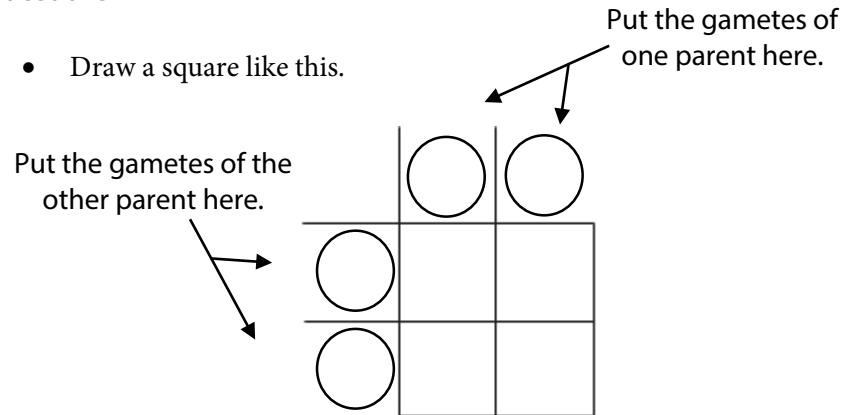
Why not use a small letter “c” for the heterozygous genotype in this case?

Using the Punnett Square

The *Punnett Square* is a simple method of predicting the probable outcome of genetic crosses.

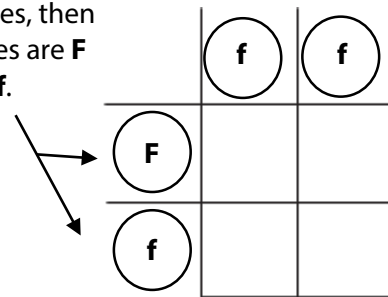
Procedure

- Draw a square like this.



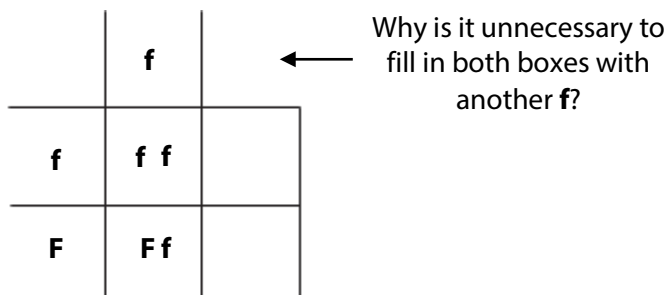
- Determine the kinds of gametes that are made by each of the parents in this cross ($Ff \times ff$), and put those gametes into the parent boxes of the Punnett Square.

If your spouse is heterozygous (Ff) for free-ear lobes, then her gametes are **F** and **f**.



If you are homozygous recessive (ff) for attached earlobes, then all of your gametes are **f**.

- Fill in the offspring boxes of the Punnett Square.



In this example there are only two possible offspring genotypes. The Punnett Square tells us to expect about 50% ff and 50% Ff. Sometimes the Punnett Square is more complex than this example, and you must figure out more than one trait at a time. Nevertheless, you use the same basic method.

- Make up your own genotype example and work out the crosses.

Traits:

Symbols:

Male Genotype:

Female Genotype:

Offspring Genotypes:

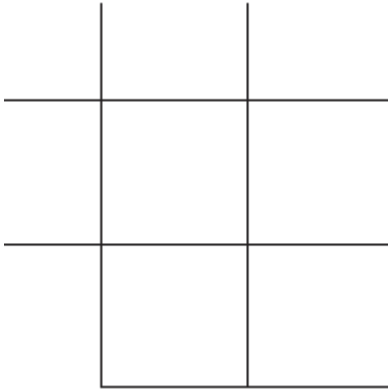
Exercise #4 Genetic Problems

Cases of Complete Dominance

Gregor Mendel grew different varieties of pea plants in his garden. When he crossed yellow-seed plants with green-seed plants, he always got yellow pea seeds.

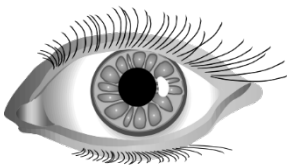
1. What is the dominant allele?
2. What is the genotype of all green-seed plants?
3. Use the Punnett Square to show Mendel's cross.
4. Do the parent yellow-seed plants have the same genotype as the offspring yellow-seed pea plant?
Parent: _____ Offspring: _____
5. What genetic fact do you know about any yellow-seed pea plant?
6. If yellow-seed pea plants are dominant to green-seed pea plants, why are there mostly green pea seeds in nature?

A dark-eyed man has children with a light-eyed woman, and they have ten dark-eyed children.



1. What is the dominant allele?
2. What is the genotype of all light-eyed people?
3. What are the genotypes of the two parents?
4. What is the genotype difference between the dark-eyed parent and the dark-eyed offspring?
Parent: _____ Offspring: _____
5. When two heterozygous dark-eyed people (Dd) are crossed, what is the phenotype ratio of dark-eyed offspring to light-eyed offspring?
Use the Punnet Square to show your answer.

Eye color is due to multiple alleles and more than one gene pair. The numerous phenotypes are determined by genes that control both the amount and the distribution of a dark pigment called *melanin*. Except for albinos, everyone has some eye pigmentation. The actual eye color is determined mainly by the location of melanin in the iris of the eye. There aren't different colored eye pigments.



All eye colors are determined by where the brown pigments are located in the iris.

Blue Eyes: No melanin in the front part of the iris. The color is due to minimal amounts of melanin in the rear of the iris with the clear front portion scattering light reflected off the melanin. This scattering is greatest in the blue spectrum giving the iris its blue color.

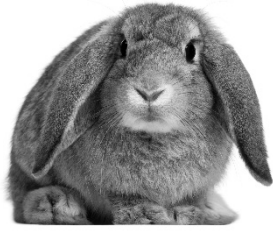
Grey Eyes: The same as blue, but with a slight amount of melanin in the front of the iris which tones down, or greys, the blue reflected from behind.

Green Eyes: A bit more melanin particles scattered in the front part of the iris creates a yellow appearance. Blended with the light blue from the rear of the iris, it produces an overall green color.

Hazel Eyes: Even more melanin particles in the front of the iris gives a slight brown color, and dilute melanin particles scattered throughout the iris add some yellow.

Brown Eyes: Melanin particles are in the front part of the iris and throughout the iris. The amount of melanin varies, leading to gradations of brown color in the eye.

Black Eyes: Large amounts of melanin in front and throughout the iris.



Floppy Ears is a recessive trait.

Test Cross to Check Genotype

If an organism shows the dominant phenotype, then one of its genes has to be the dominant allele, but you cannot be sure of the identity of the other allele unless you do a **test cross** to see if the dominant parent will breed pure. Imagine that you are in the rabbit breeding business. You know that straight ears on a rabbit is a dominant allele and floppy ears is recessive. You purchase a male straight eared rabbit. How do you figure out if your male rabbit is homozygous or heterozygous for straight ears?

Is this Straight Eared Rabbit homozygous or heterozygous?



1. Which genotype of female should you breed him to?
2. If a proper test cross is used, what phenotypes of rabbits would you see if your male rabbit is heterozygous dominant?
3. What ear phenotypes would you see if your male dog is homozygous dominant?
4. Complete the Punnett Square to show the test cross that would convince someone that your rabbit is homozygous for straight ears.



Wavy Hair is incomplete dominance.

Cases of Incomplete Dominance

When a straight-haired mouse is crossed with a curly-haired mouse, the result is always wavy hair. If two wavy-haired mice cross:

1. What are the genotypes of the two wavy-haired mice?
2. Draw the Punnett Square of a cross between two wavy-haired mice, and show the probable genotypes of their offspring.
3. What is the expected phenotype ratio of the offspring?
 _____% _____% _____%
4. What is the expected genotype ratio of the offspring?

Red orchids with straight petals are crossed with white orchids that have curly petals. The results are all pink orchids with wavy petals.

1. What are the genotypes of the two parent orchid plants? Remember: You are dealing with two different traits.

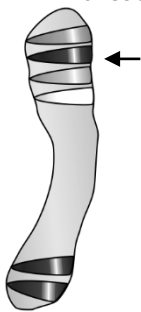
First parent: _____ (color) _____ (shape)

Second parent: _____

2. What is the genotype of the offspring orchids?

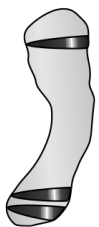
Offspring: _____ (color) _____ (shape)

The Y chromosome is missing these genes.



X Chromosome

"male-making" genes



Y Chromosome

**Exercise #5
Sex-Linked Traits**

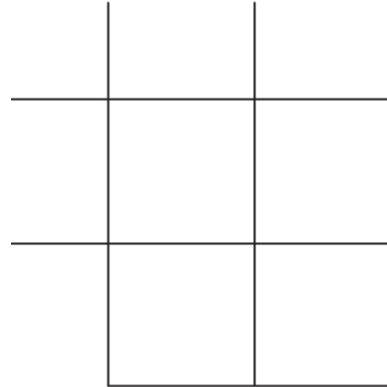
Sex Determination

Humans have 23 homologous pairs of chromosomes. Twenty-two of these pairs are named using the numbers 1 through 22. The 23rd pair is individually labeled with the letters "X" and "Y". These labels distinguish them as the *sex chromosomes*. They are not exactly the same. The Y chromosome has the "male making" genes, and the X does not. Also, the Y chromosome is missing some of the genes that are carried on the X chromosome. The female has two X chromosomes, and the male has one X and one Y chromosome.

During meiosis in the male two types of sperm are produced: those carrying the X and those carrying the Y chromosome. Females produce eggs carrying only the X chromosome. If a Y chromosome is present in the cells of an embryo, then the child becomes a male. If the Y is not present, the child becomes a female. It is the presence or absence of the Y chromosome that determines the sex of a child. This means that a male child receives a Y chromosome from his father and an X chromosome from his mother. A female child receives an X chromosome from her father and the other X chromosome from her mother.

? Question

Draw a Punnett Square to show a cross of X and Y chromosomes in the fertilization of male and female gametes. The offspring boxes should reveal why we have about a 50% male to 50% female ratio within the human population.



Sex-Linkage

The X and Y chromosomes are not exactly identical (Y is missing some genes that are on the X), and sometimes there are unequal frequencies of phenotypes in male and female offspring. If any phenotype is distributed unequally between male and female offspring and those differences are due to X and Y chromosome differences, then we call those traits *sex-linked*. Actually, “sex-linked” means that the gene is carried on the X chromosome and not on the Y chromosome. It could be more accurate to call them *X-linked*. It is easier to understand sex-linkage by looking at the sex chromosomes. See Figure 11.3.

“Sex-linked” means carried on the X chromosome.

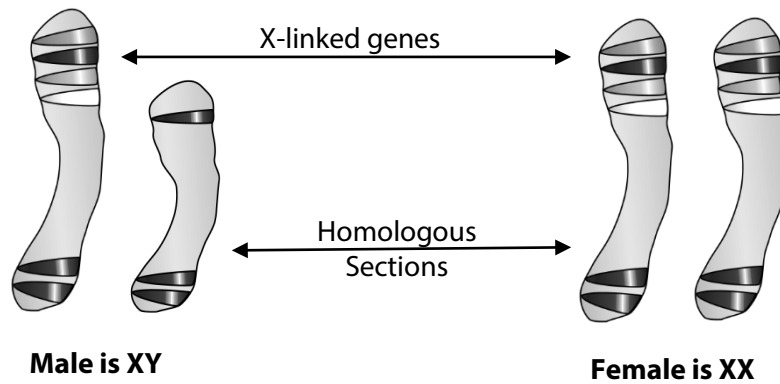


Figure 11.3. Differences between the X and the Y chromosomes.

There is a homologous section of the X and Y chromosomes that is the same, and these genes are inherited just like the genes on any other homologous chromosome pair. Notice that the Y chromosome is very short. It lacks some of the genes that are carried on the X chromosome. The X-linked section on the X chromosome carries genes that are missing from the Y chromosome.

? Question

1. How many copies of an X-linked gene does a male have?
2. Will a male be able to give his X-linked genes to his daughter? Explain.
3. Will a male be able to give his X-linked genes to his son? Why or Why not?
4. How many copies of an X-linked gene does a female have?
5. A male child gets X-linked genes from which of his parents?
6. A female child gets X-linked genes from which of her parents?
7. If a father is carrying an X-linked allele, then how many of his sons will get that allele?
8. How many of his daughters will get that allele?
9. If a mother has a defective X-linked allele on one of her chromosomes and the other chromosome is normal, then how many of her sons will get that defective allele?
10. Will any of her daughters get the defective allele? How many?
11. If we found that none of the daughters actually showed the defective phenotype, how could we explain it?

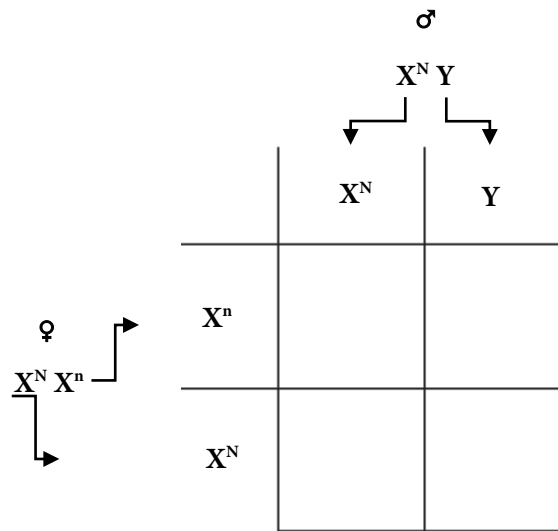
Tips for Solving Sex-Linked Genetic Problems

Follow the X, and follow the Y.

There is a sex-linked gene on the X chromosome that causes a disorder called *hemophilia*, where the blood fails to clot properly when a person is injured. This disorder is recessive and can be symbolized by the small letter “n.” Normal blood clotting is dominant and can be symbolized by the capital letter “N.” In solving sex-linked cases we not only use letters to symbolize the genes, but we also include the X and Y chromosomes and follow the sex chromosomes into the next generation.

- Using these symbols we can indicate a female who is heterozygous for clotting as $X^N X^n$.
- A homozygous female for normal clotting would be $X^N X^N$.
- A hemophilic male would be $X^n Y$.

We would diagram the Punnett Square showing the cross between a heterozygous female and a normal clotting male like this:



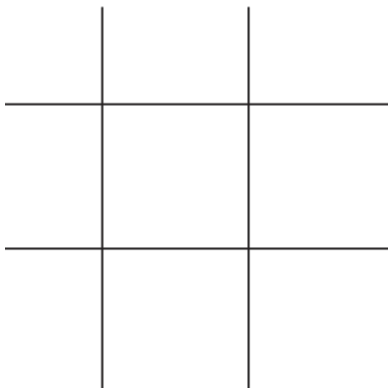
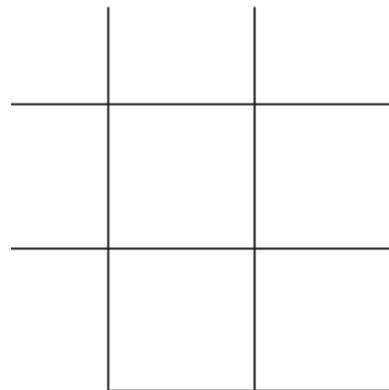
? Question

1. Complete the Punnett Square above showing the offspring.
2. What is the genotype for the female parent?
3. What is the genotype for the male parent?
4. What are the genotypes for their offspring?

5. What are the chances that any child will be a hemophiliac?
6. Is it the father or the mother that passes the hemophilia gene to the male child?
7. Failure to distinguish between red and green colors is caused by a recessive allele and is a sex-linked gene carried on the X chromosome. A red-green color-blind male marries a normal female. Of their six children (four boys and two girls), all have normal vision.

What is the most probable genotype of the mother?

8. Will any of their four male children pass this disorder on? Explain.
9. Draw a Punnett Square of this cross to prove your answers.



10. A normal-vision female gave birth to a color-blind daughter. Her husband has normal vision. He claims that the child is not his. Does the genetic information suggest someone else is the child's father? Explain and prove your answer using a Punnett Square.

