

5. Inheritance

OBJECTIVES

- Understand the relationship between Mendel's laws, molecular genetics, and gamete production
- Understand the concept of probability and its role in transmission of genetic traits
- Understand modes of transmitting genetic traits determined by genes on autosomes and sex chromosomes
- Understand the difference between simple and complex inheritance
- Learn to predict outcomes from parental crosses in offspring
- Learn steps of segregation analysis in determining the mode of inheritance demonstrated in a pedigree

“Have you ever wondered...?”

- 👉 Why do I look so much more like my father than my mother, if each contributed 50% of my genes?
- 👉 How can a set of parents with brown eyes produce a child with blue eyes?

Gregor Mendel

Although Darwin and Wallace knew that variation was vital for natural selection to occur, they didn't know how that variation came to be. The mystery of how traits were passed from parent to offspring was solved by Johann Mendel (he took the name Gregor later while at a monastery) in the 1860s. Mendel's breeding experiments with pea plants led to his discovery of the laws of inheritance, which later were verified with knowledge of chromosomal behavior during meiosis.

Mendel was successful partly through luck but mainly because of carefully planned experiments, a large sample size (about 28,000 individual plants!), use of a statistical approach (probability), and his fortuitous choice of genetically **simple traits** (those controlled by only one gene). These traits are now called **Mendelian traits**.

Mendel's experiments included observing the transmission of a single trait at a time and also observing two traits at a time. From these experiments, he concluded these two “laws” of inheritance:

1. For any given trait, members of a pair of “characters” separate (segregate) from each other during the formation of gametes, so only one copy (one gene) is passed on from each parent. This is his **principle of segregation**. We now know that this explains the separation of members of a gene pair from each other during the formation of gametes (when homologous pairs separate).
2. Genes on one set of homologous chromosomes don't influence the distribution of gene pairs on other chromosomes (for example, the chance for a pea seed to be round or wrinkled is independent of its chance of being yellow or green). This is because genes from homologous chromosomes separate independently from each other during meiosis and are randomly assorted in the gametes. This is the **principle of independent assortment**.

Recall from Chapter 2 that a **gene** is a segment of a chromosome's DNA coding for a specific protein, and an **allele** (*G allele*: one another, parallel) is an alternative form of a

gene. For any trait, an individual inherits one allele from each parent so each individual always has two alleles for each trait. The two alleles at a given locus make up an individual's **genotype**. The alleles are represented by letters (see Figure 5.1). As shown in Chapter 2, chromosome 9 has genes for melanin production and for blood type for the ABO blood group.

If an individual inherits two alleles coding for the same form of the trait, they are said to be **homozygous** for that trait. If the alleles code for different forms of the trait, the genotype is **heterozygous**. Thus, for example, if someone inherits one allele that does not result in normal melanin production and the other allele that does code for melanin production, what determines which form the trait will take? Will the person produce melanin or not?

Many traits have a consistent pattern of expression such that one allele may be expressed whenever it is present and the other allele is expressed only if it has been passed on by both parents. If an allele is always expressed when present, this allele codes for the dominant form of the trait. The **recessive** form of a trait is expressed only when both "recessive" alleles are present and there is no allele for the **dominant** form of the trait present. Genotypes can be **homozygous dominant**, **heterozygous**, or **homozygous recessive**.

The physical expression of a trait is the **phenotype** (*G pheno*: show, seem, appear). Thus, the **genotype** consists

of the genes (alleles) present at a particular locus on a homologous pair of chromosomes, and the **phenotype** is the resulting observable form of the trait.

If a specific trait is coded for by a gene on a chromosome numbered 1–22 (the autosomes), it is an **autosomal trait**. A trait coded for by a gene on the 23rd pair (the sex chromosomes) is a **sex-linked trait**.

Autosomal Traits

In the following discussion, we will first consider transmission of one trait, then two traits.

Consideration of One Trait

It is straightforward to follow the transmission of *one* autosomal trait from parents to offspring. We'll use albinism as an example.

Albinism (lack of production of the pigment melanin) is coded for by a recessive autosomal gene. This gene has two alternative alleles: *A* (the dominant allele) and *a* (the recessive allele). Therefore, using the letters *A* and *a* to represent the dominant and recessive alleles, the genotypes *AA* and *Aa* (homozygous dominant and heterozygous, respectively) would result in normal melanin production, and therefore in phenotypes with normally pigmented coloration. The genotype *aa*, by contrast, would result in the recessive form and thus an albino phenotype.

The following explains how to predict the probability of offspring from a cross between an *albino woman* and a *man heterozygous for albinism*.

1. List the genotype of the mother as *aa* and the father as *Aa*.
2. Figure out what alleles for this trait will be carried by the gametes in individuals with the above genotypes. Remember that during meiosis the homologous chromosome pairs are separated from each other during the first division (review in Chapter 4, if necessary, and see Figure 5.2). Although an individual has two alleles for each trait in each somatic cell (one maternally and one paternally derived), when they produce gametes, each gamete will have only *one* allele representing each trait (to potentially combine with an allele from an individual of the opposite sex).

In our example, the mother has only *a* alleles, so all of her eggs will carry one of these recessive alleles (see Figure 5.2A). The father is heterozygous, so he will produce sperm carrying dominant and recessive alleles in approximately equal numbers. Therefore, half of his sperm will carry an *A*, and half an *a* (see Figure 5.2B). In this process, alleles are passed on to daughter cells, which develop into gametes.

3. Set up a **Punnett square**. This is a simple tool, developed in the early 1900s by geneticist Reginald Punnett, to

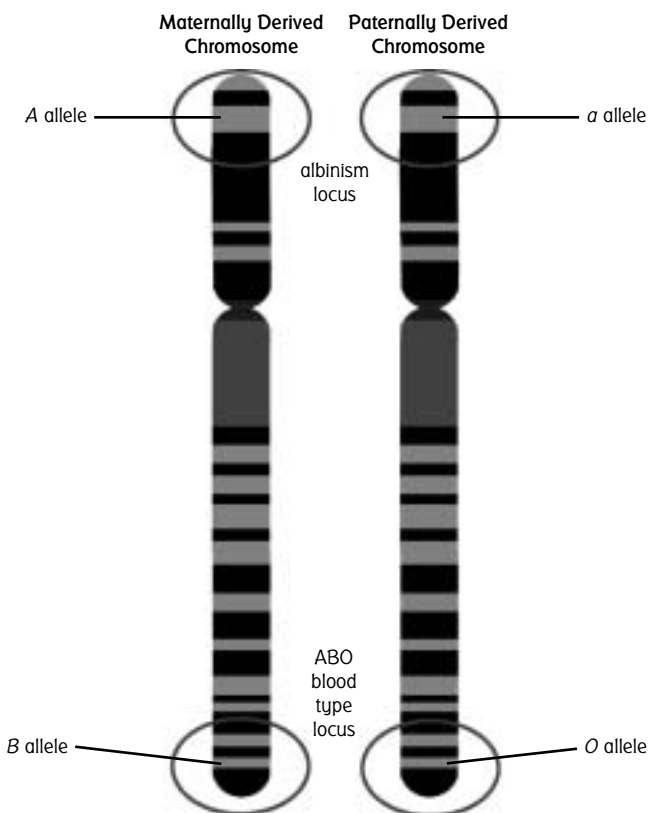
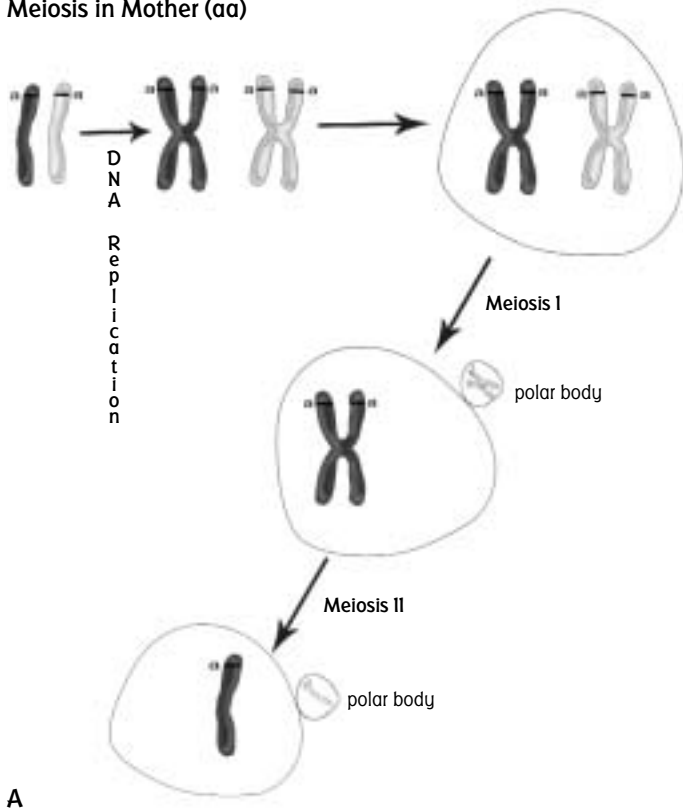


FIGURE 5.1 Chromosome 9 for a Hypothetical Individual, Showing Samples of Loci for Two Traits

Meiosis in Mother (aa)



Meiosis in Father (Aa)

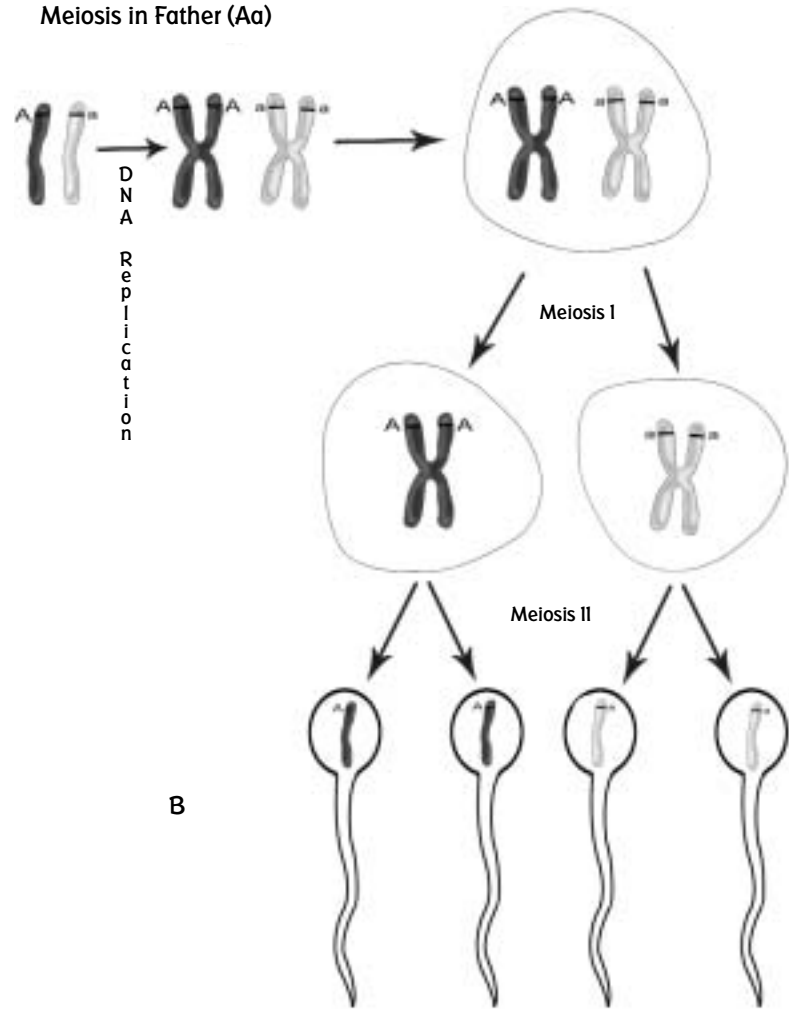


FIGURE 5.2 Process of Meiosis in Mother (A) and Father (B)

predict offspring outcomes depending upon the parental genotypes.

List the alleles possessed by one parent along the top of the square and those of the other parent along the left side of the square—in our example, we'll list the mother's alleles along the top. Thus, these will represent the kinds of alleles present in the eggs of the mother and the sperm cells of the father (see Figure 5.3). Keep in mind that we are only focusing on one trait—actually, in each gamete there will be alleles present for *all* traits.

- Fill in your Punnett square. The results represent possible outcomes of allele combinations (genotypes) in the zygote (see Figure 5.4).
- List the possible genotypes and phenotypes of the offspring, as well as the probability of each type.

genotypes	phenotypes	probability
Aa	nonalbino	1/2 (50%)
aa	albino	1/2 (50%)

- List the ratio of the phenotypes and the genotypes, in terms of probabilities:
 - genotypic ratio**—number of homozygous dominant to heterozygous to homozygous recessive: 0:2:2

FIGURE 5.3 Punnett Square, Showing Alleles in Gametes to be Passed On

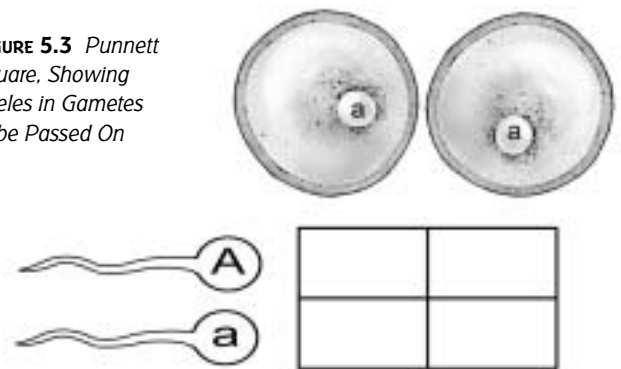
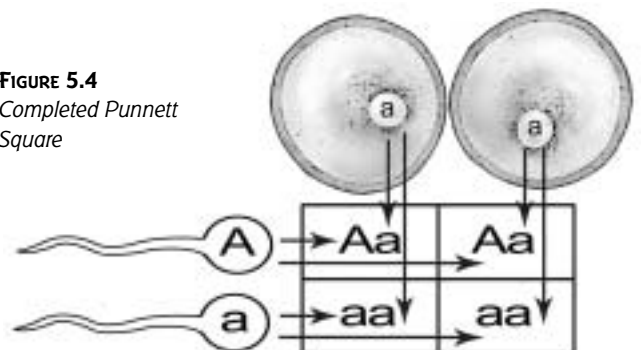


FIGURE 5.4 Completed Punnett Square



- b. **phenotypic ratio**—number expressing dominant form of trait to number expressing the recessive form: 1:1

Consideration of Two Traits

If we consider *two traits* at once, Mendel's principle of segregation becomes important in demonstrating that the chances of inheriting one trait has no effect on the chances of inheriting the other. That is because, during gamete formation, the alleles (on their respective chromosomes) segregate/assort independently from one another and are assorted randomly in the gametes. Thus, the chances for someone's bloodtype to be A or B is independent of his or her chances of being an albino or not.

The Punnett square also can be used to determine the probabilities of outcomes of more than one trait considered together. We'll use the traits of tongue-rolling and albinism in this example. Remember that albinism is inherited recessively by a gene on one of the autosomes. Tongue-rolling is inherited as an autosomal dominant trait. That is, the ability to tongue-roll is dominant, so someone who is heterozygous (Rr) for the trait can roll the tongue, and someone who is homozygous recessive (rr) does not have the musculature to tongue-roll. Actually, tongue-rolling may be controlled by genes at more than one locus, and it has some environmental

component as well (e.g., Martin, 1975). Still, the trait serves as a useful example.

Work through this example of a woman who cannot roll her tongue and is not an albino (she's homozygous dominant for albinism) who marries a man who can roll his tongue (he's heterozygous for tongue-rolling) and is an albino.

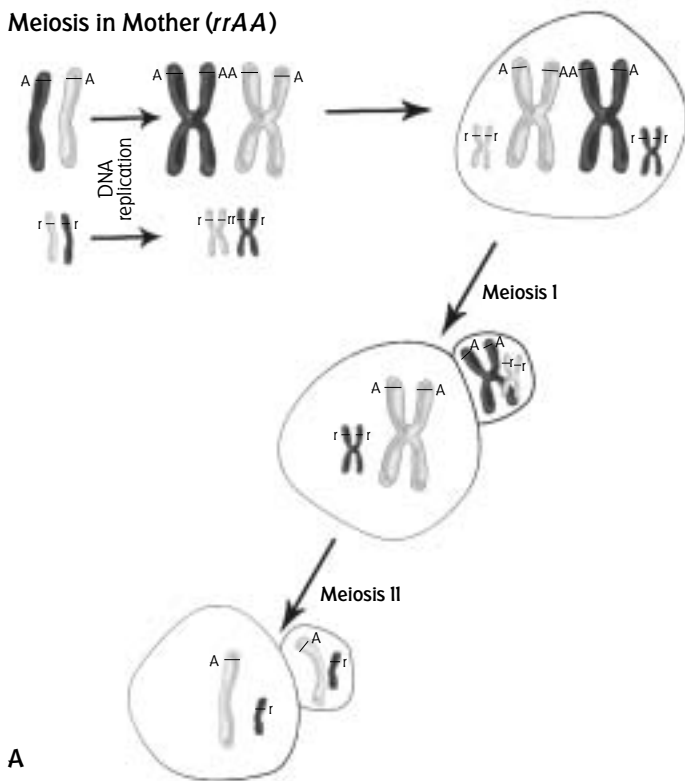
1. List the genotypes of the parents for both traits:

Mother: $rrAA$ Father: $Rraa$

2. Next, figure out what alleles are present in the gametes of the mother and father. *This is a crucial step! Do it carefully, and ask your instructor if you do not understand!* See Figure 5.5. The process of meiosis will result in the following:

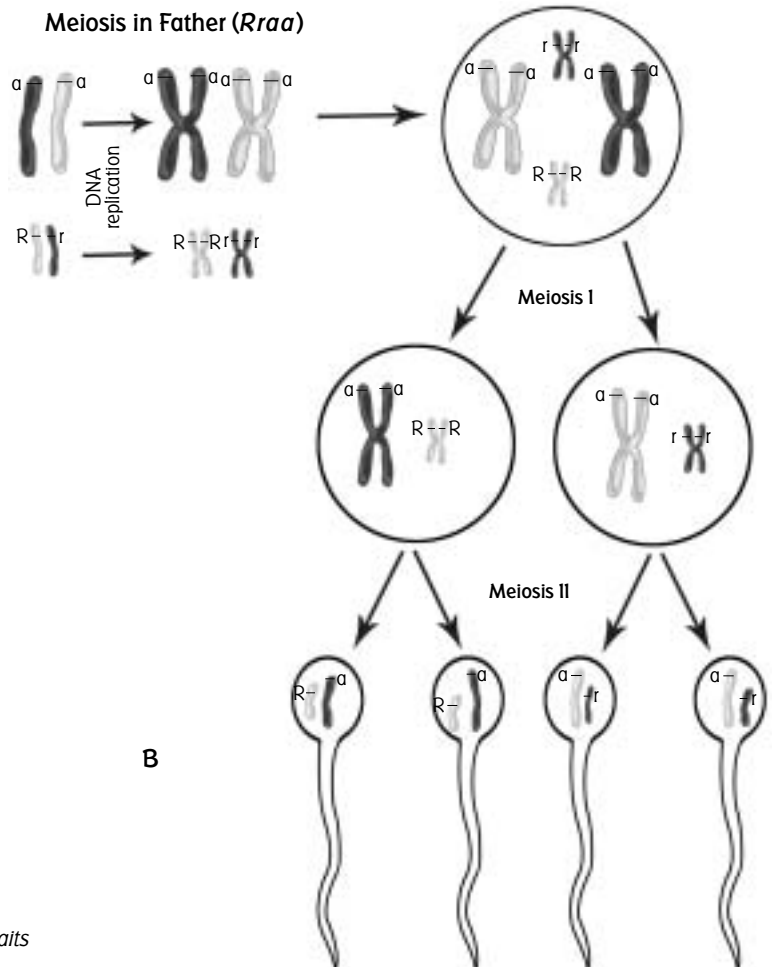
- a. In the mother's eggs, the tongue-rolling trait will be represented only by the r allele (the mother's genotype is rr), and the albinism trait will be represented only by the A allele (the mother's genotype is AA). Thus, all eggs she produces will have an r and an A to represent those two traits. This can be written as rA (see Figure 5.5A).
- b. The father's sperm will have two possible combinations of alleles. Because he is heterozygous for tongue-rolling, half of his sperm will have the R allele and

Meiosis in Mother ($rrAA$)



A

Meiosis in Father ($Rraa$)



B

FIGURE 5.5 Process of Meiosis in Female (A) and Male (B), Considering Two Traits

half the r allele. All of his sperm will be produced carrying the a allele for albinism. Thus, half his sperm will have Ra and half will have ra (see Figure 5.5B).

- Set up your Punnett square. The number of columns and rows will be dependent upon the number of possible combinations in the parents' gametes. Because there is only one possible type of egg, the mother will produce (rA), and the father will have two types of sperm (Ra and ra). The Punnett "square" can be as small as a 1×2 table (see Figure 5.6).
- Fill in the Punnett square with the expected types of offspring (Figure 5.7).
- List the possible genotypes and phenotypes of the offspring, as well as the probability of each type.

genotype	phenotype	probability
$RrAa$	tongue-roller, non-albino	$\frac{1}{2}$ (50%)
$rrAa$	non tongue-roller, non-albino	$\frac{1}{2}$ (50%)

We have focused on traits that are coded for by only one gene—called **monogenic**, or simple traits. Although many traits are coded for by more than one gene—called **polygenic**—and also may have an environmental contribution, we continue to use as examples these simple, or Mendelian, traits for the purpose of illustrating patterns of inheritance.

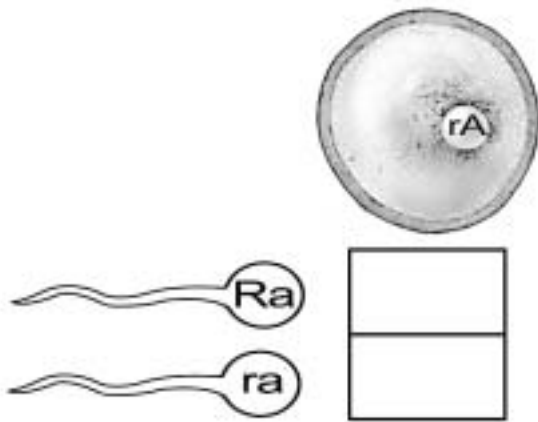


FIGURE 5.6 Punnett Square, Showing Alleles in Gametes to be Passed On for Two Traits

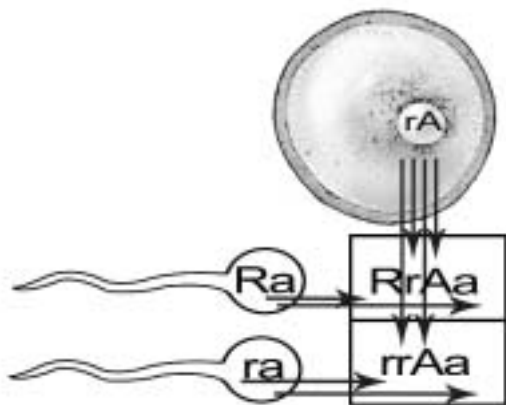


FIGURE 5.7 Completed Punnett Square for Two Traits

Blood Typing

Although one usually hears about type A, B, O, and AB, as well as positive and negative blood, we each actually have not two, but about 24 blood types! Each different blood type is coded for by genes at different loci on our chromosomes. A blood type is determined by the kind of antigen present on the surface of our red blood cells. Blood types are useful for tracking genetic traits within and between populations, investigating their biomedical effects, and learning about inheritance.

ABO Blood Group

In the blood serum, each person has **antibodies** against foreign blood antigens. By determining the type of antibodies in the blood, you can determine the individual's blood type. The simplest way to test for the type of antibody present is to add **antigens**. If the antibodies attack the antigens, the result is a clumping affect, called **agglutination**. The surface of the red blood cells has antigens with the same common name on their surface as the person's type of blood (A or B).

The **ABO blood group** is coded for at a locus on chromosome 9; alleles called I^A , I^B , and I^O (hereafter referred to simply as A, B, and O alleles) determine enzymes that are responsible for the type of antigens that are produced and reside on the surface of red blood cells. These antigens are composed of a chain of simple sugar molecules and differ just slightly from each other depending upon the alleles present. The difference between A and B antigens (determined by A or B alleles) is that each has an extra sugar molecule (different from each other) on the end of the chain, which is lacking in the chain produced by someone with only O alleles.

The various genotypes for this trait are: AA, AO, AB, BO, OO, and BB. A and B alleles are both dominant over O but are **codominant** with regard to each other. Thus, if both an A and a B allele are present, both A-type and B-type antigens are produced (and the genotype is type AB). O alleles are expressed only if they occur without an A or a B allele to "mask" their appearance in the phenotype: An OO genotype (type O blood) is the only one to produce a sugar chain with no end sugar as in blood types A, B, and AB. Table 5.1 lists the genotypes, phenotypes (blood types), antigens, and antibodies for each blood type in the ABO group.

Blood types for the ABO blood group must match if blood from one person (a donor) is to be used for another (a recipient), as a result of the reaction of anti-A and anti-B antibodies against the A and B antigens, respectively, on the surface of the red blood cells. Because type O blood has neither A nor B antigens on its cell surface, it has both anti-A and anti-B antibodies in its serum. For that very reason (lack of A or B antigens), however, type O blood does not provoke a reaction (**agglutinate**) with type A, B, or AB blood. Therefore, individuals with type O blood are known as **universal**

TABLE 5.1: Characteristics of ABO Blood Group

Genotype	Blood type	Type of Antigens on Red Blood Cell Surface	Type of Antibodies in Serum
AA or AO	Type A blood	A antigens	Anti-B
BB or BO	Type B blood	B antigens	Anti-A
OO	Type O blood	None	Anti-A and anti-B
AB	Type AB blood	A and B antigens	None

donors. Type AB blood has neither anti-A nor anti-B antibodies with which to attack incoming blood cells, so it can receive any blood—type A, type B, type AB, or type O. Thus, type AB individuals are referred to as **universal recipients**.

Rh Blood Group

The blood types associated with the **Rh blood group** are Rh⁺ and Rh⁻ (A⁺, B⁻, etc. really represent blood types from two different blood groups, ABO and Rh). There are actually several alleles coding for Rh blood types, determined by simple dominant/recessive inheritance. If an individual is homozygous dominant or heterozygous, he or she is Rh⁺; the individual is Rh⁻ if homozygous recessive. A person who

is Rh⁺ has antigens on the surface of the red blood cells that can provoke the production of antibodies in the serum of someone without those antigens (an Rh⁻ person).

The Rh blood group has great significance medically, because of potential problems with **Rh incompatibility** between a mother and her developing fetus. If the antibodies are produced by a (Rh⁻) mother's immune system in response to antigens on her (Rh⁺) fetus' red blood cells, the mother's antibodies can attack the fetal cells and break them open, releasing the hemoglobin within and resulting in severe **anemia** in the infant around the time of its birth. If the mother receives prenatal care, medical advances have virtually eliminated the problem in our society today.

LAB EXERCISE 5.1

NAME _____ SECTION _____ DATE _____

1. Can you roll your tongue? Can you tell what your genotype is? What are the possibilities for your genotype for this trait? Make a Punnett square for the following exercise to answer these questions.
 - a. If you can roll your tongue, let's (for the purposes of this exercise) assume that you're heterozygous. If you produce offspring with another heterozygote, what are the possible genotypes of these offspring? Their phenotypes? (Remember, use the letter *R* and *r* to represent the dominant and recessive alleles.)

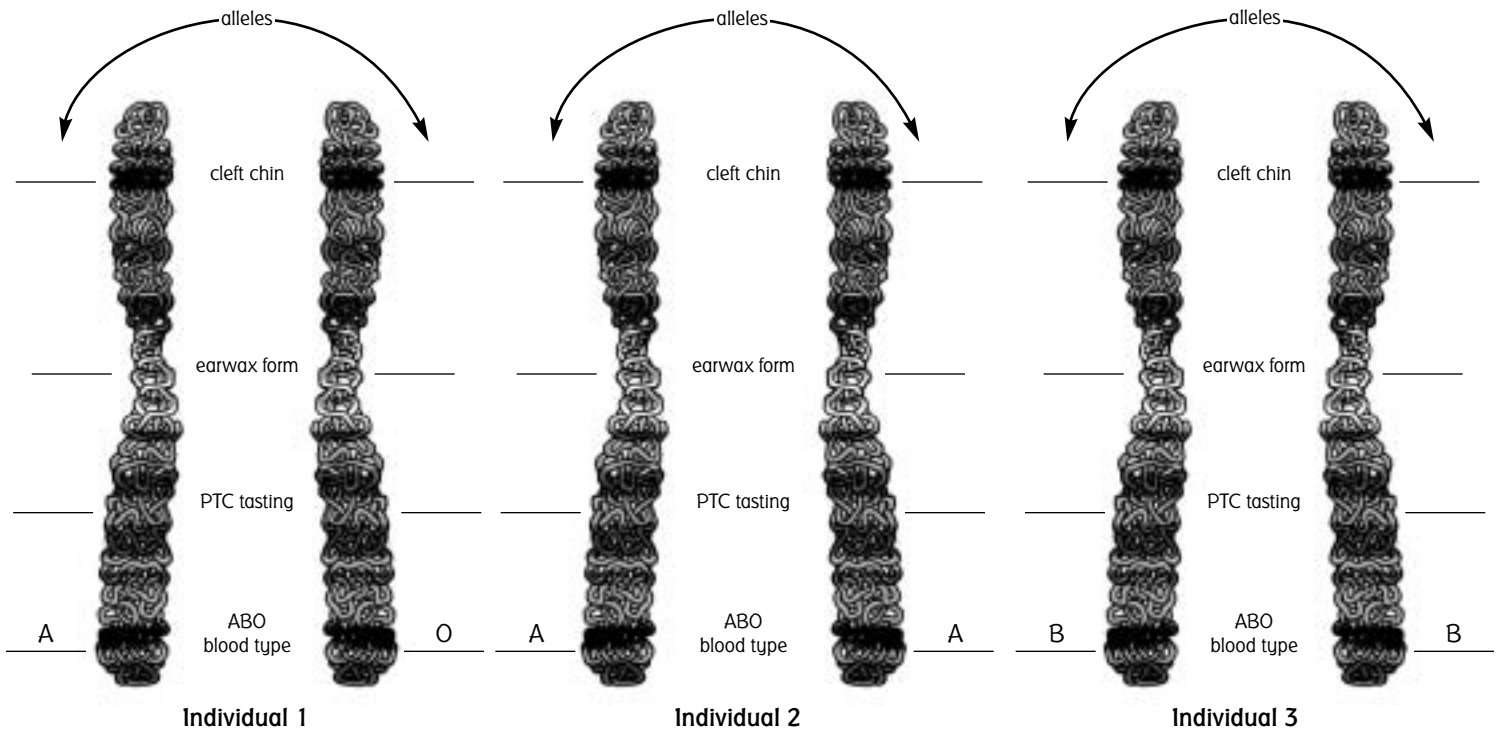
- b. If you cannot roll your tongue, and you produce offspring with someone who is heterozygous for tongue-rolling, what are the possible genotypes of these offspring? Their phenotypes?

2. Represented in the illustration on the next page is chromosome 11 (it hypothetically shows several Mendelian traits to illustrate; the actual genes present on chromosome 11 are different from these). This chromosome pair is taken from the karyotype of three different individuals—Individuals 1, 2, and 3.

Using the information given in the chart below and previously in this chapter, you will create genotypes for each chromosome pair for four traits: ABO blood type, cleft chin, earwax form, and PTC tasting.

Trait	Description	Dominant form of trait	Alleles
Cleft chin	Dimple in center of chin	Possession of cleft chin	<i>D, d</i>
Earwax (cerumen) form	Sticky yellow/brown vs. dry grayish earwax	Sticky earwax	<i>C, c</i>
PTC (phenylthiocarbamide) tasting	Ability to taste bitterness	Ability to taste PTC	<i>T, t</i>

- a. Fill in the alleles on the chromosomes illustrated below, using whatever combination of dominant and recessive alleles you wish. (One example is filled in for you; you may change them if you wish.)



- b. Fill in the chart below, making the genotype for each trait different for individuals 1, 2, and 3.

	Individual 1		Individual 2		Individual 3	
	Genotype	Phenotype	Genotype	Phenotype	Genotype	Phenotype
ABO blood type	AO	Type A	AA	Type A	BB	Type B
Cleft chin						
Dwarfism						
PTC tasting						

- c. Compare the genotypes and phenotypes for your three individuals with a classmate's results. Are your results the same?

This demonstrates the variation that can result from the different combinations of alleles produced by the parents and randomly passed on to the next generation (Mendel's principles). There are several hundred thousand possible combinations of genotypes for these four traits for any group of three individuals randomly chosen from the population! Imagine the possible allelic combinations if we consider all approximately 25,000 traits represented by the genes on all 23 chromosomes!

LAB EXERCISE 5.1 (continued)

3. At DuPont Company in 1931, a chemist instigated an accident, in which some synthesized compound phenylthiocarbamide (PTC) exploded into the air (*Gadsby, 2000*). Some of the workers actually could taste bitterness in the air and others couldn't. This led to the "taste test" for PTC. When the compound was handed out as crystals at the 1932 American Academy for the Advancement of Sciences (AAAS) conference, about a quarter of the people could not taste it ("non-tasters"), and the others ("tasters") said it was incredibly bitter.

It was quickly noted that the ability to taste was determined genetically, and it was transmitted in a simple Mendelian fashion. Phenotypes were divided into non-tasters and tasters. The ability to taste is dominant to the inability, so a non-taster would be homozygous recessive.

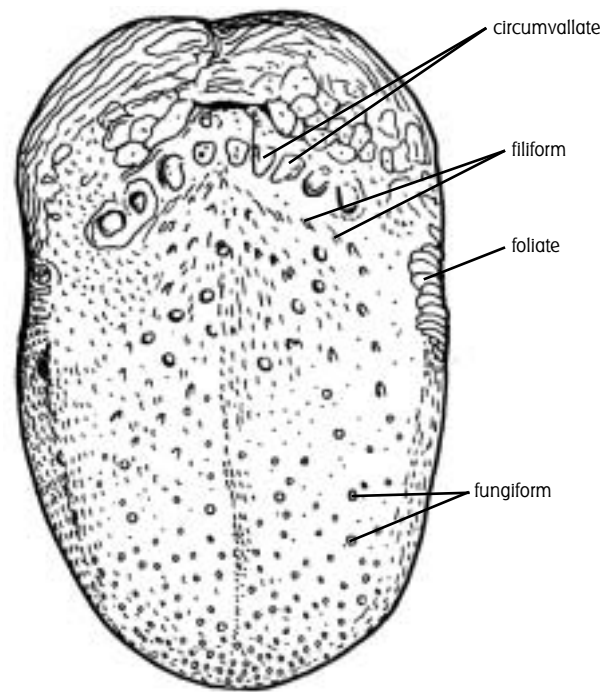
- a. Taste the PTC paper, and note your:

phenotype **genotype (or at least any known alleles—use the letters *T,t*)**

More recently, human taste specialist Linda Bartoshuk (1994), found a third level of tasters, and thus divided people into super-tasters, tasters, and non-tasters. These may correlate with homozygous dominant, heterozygous, and homozygous recessive genotypes.

We have four types of **papillae** on the tongue. The papillae house the receptor cells (taste buds). Molecules with sweet, salty, sour, or bitter tastes stimulate the receptors, which stimulate nerve endings inside the tongue, and the message of the type of taste is carried along the nerve cells to the brain. The number of taste buds embedded in the tips of the tongue are controlled by our genes. The types of papillae are **circumvallate**, **foliate**, **filiform**, and **fungiform** (the mushroom-shaped bumps concentrated more at the tongue's tip), as illustrated.

- b. Drip a small drop of food coloring (green or blue works best) onto a thin strip of paper, then use it to paint the tip of your tongue (*Scientific American Frontiers Archives, 1999*).
- c. Punch a hole in an index card and place the hole over the tip of your tongue. This small area of your tongue will serve as your sample of taste buds (*Shahbake et al., 2005*). The edge of the hole should be on the tip of the tongue, in the center.
- d. Have someone else count (using a magnifying glass, if available) the fungiform papillae (these should appear pale against the food-colored tongue background). *Super-tasters* may have up to 50 fungiform papillae. *Tasters* usually have 15 to 30, and *non-tasters* may have as few as 10 (*Bartoshuk et al., 1994, 1998; Discover magazine, 2000*).
- e. Are you a PTC taster? According to fungiform-counting, what is your tasting level? Do your taste preferences match the results of PTC tasting and fungiform papillae-counting? Super-tasters often avoid black coffee, grapefruit, and very sweet desserts. (More females than males tend to be super-tasters.)



- f. Take a count of the phenotypes of all of your classmates so you have a total number of supertasters, tasters and non-tasters.

Supertasters: _____

Tasters: _____

Non-tasters: _____

S E L F - T E S T 5 . 1

NAME _____ SECTION _____ DATE _____

1. Two normally pigmented parents have an albino child. What are the parents' genotypes? The child grows up and marries another albino. What is the probability of their having an albino child?
2. A zebra population has a mutant allele for spots. The spot trait (S) is dominant to the striped allele (s).
 - a. A spotted male mates with a striped female. Assuming that he is homozygous, what is the probability that they will have striped offspring?
 - b. If one of their daughters mates with a spotted (heterozygous) male, what is the probability the offspring will be striped?
3. If a male who is Rh^- mates with a female who is Rh^+ (heterozygous), what are the possible genotypes and phenotypes of their offspring? (Use the letters D and d to represent the dominant and recessive alleles, respectively; use a Punnett square.)
4. A normally pigmented woman and an albino man have nine normally pigmented children and one albino child. What is the woman's genotype?
5. The ability to taste the chemical PTC is transmitted as a dominant allele, represented by T ; the recessive allele is t . Two normally pigmented taster parents have an albino son and a non-taster daughter with normal pigmentation.
 - a. What are the genotypes of the parents?
 - b. What is the chance that the albino son is a taster?
 - c. What is the chance that the non-taster daughter is heterozygous for the gene controlling albinism?
 - d. The non-taster, non-albino daughter marries a taster man with normal pigmentation; his mother was a non-taster albino. What is his genotype?
 - e. What is the chance that a child of theirs will be a taster albino if the wife is heterozygous for albinism?

6. There is a case of disputed paternity, involving a woman (W) and her children (i, ii, iii, iv) and two men (Y and Z). The analysis is limited to the ABO and MN blood group systems. The phenotypes are as follows:

		father:
W: A, MN	i: A, MN	_____
Y: B, MN	ii: A, M	_____
Z: AB, N	iii. AB, M	_____
	iv. O, N	_____

Assuming that only Y or Z could be the father of these children, assign the children to their appropriate father. In the ABO blood group system, A and B are codominant, and both are dominant over the recessive O allele. In the MN blood group system, M and N are codominant. You can solve this problem either with Punnett squares for both possible parental crosses (W with Y; W with Z) or without a Punnett square, working with a process of elimination of possibilities.

7. You are typing your blood. What is your blood type if:

- a. There is no agglutination?
- b. There is agglutination with anti-A but not anti-B?
- c. There is agglutination with both anti-A and anti-B?

Which blood type is known as a universal donor? _____

Why?

- e. Which blood type is known as a universal receiver? _____

Why?

8. A female who is a PTC taster and a tongue-roller mates with a male who is also a PTC taster but is not a tongue-roller. Use the letters T,t (for PTC tasting) and R,r (tongue-rolling). The ability to taste is dominant, and the ability to tongue-roll is dominant.

- a. What do we know of their genotypes? Female _____ Male _____

(Fill in only the alleles you know at this point, without looking ahead.)

- b. They have three kids with the following phenotypes:

Kid 1: PTC taster and tongue-roller

Kid 2: PTC taster and non-tongue roller

Kid 3: PTC non-taster and tongue-roller

- c. With the knowledge of the next generation's phenotypes, now what do we know of the genotypes of the parents?

Female _____ Male _____

9. In a case of disputed paternity, a woman has a daughter with type A, Rh⁺ blood. The woman's blood type is A, Rh⁻. What blood types could a possible father have?

10. How is it possible that a set of parents with brown eyes produces a child with blue eyes?

Sex-Linked Traits

Sex-linked traits can be coded for by genes on either the X or the Y chromosome. Traits coded for by genes on the X chromosome are X-linked, and those on the Y chromosome are Y-linked. Most traits that are sex-linked are X-linked because X is a much larger chromosome, possessing many more genes than the Y chromosome. Genes on the Y chromosome relate to male sexual development. Most genes on the X chromosome have nothing to do with female sexual development but are vital for individuals of both sexes.

One example of an X-linked trait is red/green colorblindness. On the X chromosome is a gene coding for **opsin** proteins (proteins in cone cells of the retina that enable us to perceive color); these opsins bind to visual pigments in the red-sensitive cones, green-sensitive cones, or blue-sensitive cones, making the visual pigment/opsin complex sensitive to light of a particular wavelength. If the opsin protein (a product of a gene, via protein synthesis) is absent or defective, the color vision is affected.

The vast majority (93%) of people who are colorblind are males, because this trait is transmitted by a recessive gene on the X chromosome. When denoting the genotype for a sex-linked trait, *always include the sex chromosomes, X and Y*, as shown below. This is important because the pattern of transmission of a sex-linked trait differs for males versus females. Below, *C* denotes the normal, dominant allele, and *c* the recessive, faulty allele. The genotypes for males and for females are as follows:

Genotypes for Males

$X^C Y$

$X^c Y$

Genotypes for Females

$X^C X^C$

$X^C X^c$

$X^c X^c$

Phenotypes

Male with normal color vision

Colorblind male

Phenotypes

Female with normal color vision (non-carrier)

Carrier for colorblindness (but normal color vision)

Colorblind female

Note that females can be homozygous dominant, heterozygous, or homozygous recessive. Because males have only one X chromosome, and thus one allele for each trait coded for by this chromosome, they cannot be referred to as any of these! They are **hemizygous**. Recessive alleles on the X chromosome are always expressed in males, because there is no dominant allele to “compensate.”

The steps for predicting outcomes for a sex-linked trait are demonstrated by this example. A man with normal color vision and a woman who is a carrier for colorblindness are about to have a child. What are the possible genotypes for the child?

1. List the genotypes of each parent. Man: $X^C Y$

Woman: $X^C X^c$

2. Set up your Punnett square, remembering to include the sex chromosomes with the allele that is on it, as shown below.

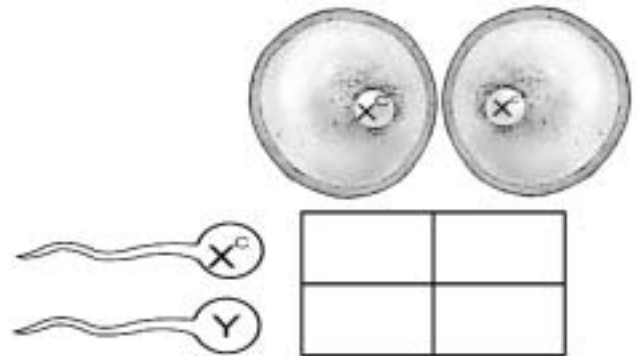


FIGURE 5.8 Punnett Square for X-linked Trait, Colorblindness

3. Fill in the Punnett square with the expected types of offspring.

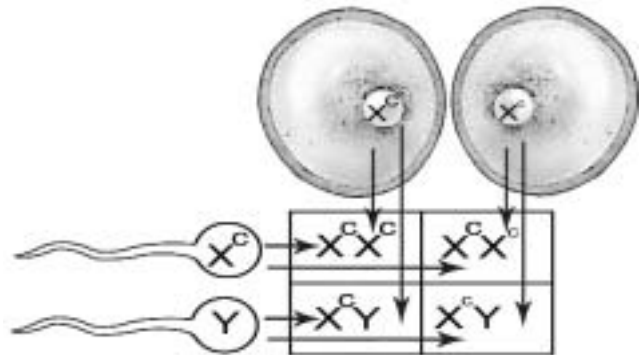


FIGURE 5.11 Completed Punnett Square for Colorblindness

4. List the possible genotypes and phenotypes of each, and their associated probabilities of occurrence.

Genotypes	Phenotypes	Probability
$X^C X^C$	normal female	1/4 (25%)
$X^C X^c$	carrier female	1/4 (25%)
$X^C Y$	normal male	1/4 (25%)
$X^c Y$	colorblind male	1/4 (25%)

Pedigrees

An important part of the study of inheritance is the search for patterns of transmission of traits from one generation to the next. Does a specific trait follow a definite and predictable pattern of transmission? Knowledge of the mode of inheritance has important implications for biomedical research, genetics studies, and genetic counseling.

We can visualize patterns of inheritance by constructing a **pedigree**, which is a diagram that delineates the genetic relationships of family members over two or more generations. **Segregation analysis** is used to determine which of several modes of inheritance is responsible for producing specific patterns in a familial line. This involves proposing a genetic hypothesis (a possible mode of transmission), testing it, and continuing the process until a hypothesis is proposed that

accounts for the observed patterns of inheritance with a high degree of accuracy and cannot be rejected.

The five modes of inheritance (which follow a Mendelian pattern) discussed here are: autosomal recessive, autosomal dominant, sex-linked recessive, sex-linked dominant, and Y-linked. Each has a set of characteristics that can be used to identify it from a pedigree, listed in the accompanying box.

The way to “test” a hypothesis is to provisionally assume a particular mode of inheritance, and try out associated genotypes to see which fits for parents and offspring. Below are examples of abbreviated pedigrees for autosomal and sex-linked traits, with sample genotypes and explanations. Keep in mind that to determine a pattern of inheritance, one normally would observe several generations of a family or extended family rather than only a set of parents and their offspring. Read over the boxed information, then try these out. The actual mode of inheritance for each is listed at the end of this section.

Characteristics of Modes of Mendelian Inheritance

Autosomal Recessive

- Most affected individuals are children of unaffected parents
- All children of two affected parents (homozygous recessive) are affected
- Expressed in males and females to (approximately) same degree
- May skip generations

Autosomal Dominant

- Each affected individual has at least one affected parent
- Number of affected males and females is roughly equal
- Two affected individuals may have unaffected child (because affected individuals can be heterozygotes)

Sex-linked

X-linked Dominant

- Affected males produce all affected daughters and no affected sons
- A heterozygous-affected female will transmit the trait to half her children, with males and females equally affected.
- Twice as many females affected, on average, as males.

X-linked Recessive

- Hemizygous males and homozygous females are affected
- Males express the trait when present (because hemizygous)
- More common in males than in females
- Affected males get mutant allele from mother
- Males transmit allele to all daughters (via X chromosome), but not to sons (pass only Y to sons)
- May skip generations

Y-linked

- Appears only in males
- Affected males pass trait to sons but not to daughters
- Every Y-linked trait should be expressed

Key to Symbols

- | | |
|----------|-------------------|
| □ male | ■ affected male |
| ○ female | ● affected female |
| | mating □—○ |
| | sibship ○—□ |

$Aa Aa$

a. ○—□

●

aa

$Rr Rr$

b. ■—●

□

rr

$XY^{TDF} XX$

c. ■—○

■

XY^{TDF}

$X^H X^h X^h Y$

d. ●—□

□

$X^h Y$

$X^C X^C X^c Y$

e. ○—■

○

$X^C X^c$

- a. **Autosomal recessive:** If two unaffected parents have an affected offspring, the trait must be “hidden” in the parents’ genotypes, and thus be recessive. If the trait were dominant, at least one parent would be affected.
- b. **Autosomal dominant:** If two affected parents have an unaffected offspring, the trait must be dominant. Two affected parents with a recessive disorder would have all affected children.
- c. **Y-linked:** Father-to-son transmission rules out X-linked inheritance. All sons of affected fathers are affected; no females are affected.
- d. **X-linked dominant:** Affected woman with unaffected son rules out X-linked recessive, since both of her chromosomes would have recessive alleles—so son would be affected, but he is not.
- e. **X-linked recessive:** Unaffected daughter from affected father can’t be X-linked dominant, because father has only one type of X (with affected allele) to give a daughter, so she would also be affected.

Genetics Recap

By this point, you have gone through the major genetics processes. Try to keep the big picture in mind, from the DNA comprising the chromosomes in all of our cells, to the expression of our physical characteristics via protein synthesis, and their probability of occurrence from generation to generation. Remember also how we got our particular set of genes in the first place—from the meeting of the nucleus of our mother’s egg cell and that of our father’s sperm cell. The contents of the nucleus of all of our somatic cells are basically photocopies of that original cell, the zygote.

As for the genetic material we contribute to the next generation, in the process of producing our sperm or egg cells, we shuffle up the DNA sequences we received from our father and mother before passing it on to our children. Individuals together comprise populations, the sum total of our genetic contributions to the next generation results in the gene pool. The difference in the gene frequency between the gene pools of succeeding generations is evolution.

LAB EXERCISE 5.2

NAME _____ SECTION _____ DATE _____

- Your instructor should provide you with a colorblindness chart from the online Instructor's Manual. If you can see the reddish number against the green background, you have normal color vision. If you cannot distinguish the number, you are (red/green) colorblind.
 - Do you have normal color vision?
 - What is your genotype, or your possible genotypes, for the colorblindness trait?
(Remember, because it's a sex-linked trait, you will include the sex chromosomes in your genotype.)
- With a lab partner, you will use coin-tossing to simulate the random nature of allele combinations in offspring, and "create" a family for which you will construct a pedigree. Use the boxed information and the sample short pedigrees from earlier in this chapter for directions on the symbols to use. Although a Punnett square gives you all of the *possible* outcomes of various crosses and their probabilities of occurrence, this method will mimic the passing on of alleles for the *particular outcome* determined by your coin tosses.

Begin with a woman and a man with the following genotypes for the colorblindness trait:

Woman: X^cX^c Man: X^cY

- Start your pedigree by putting the symbols for this couple at the top (center) of a blank piece of paper. Fill in the blanks below as well as recording the genotypes next to the male/female symbols on the pedigree itself. Throughout the pedigree, remember to darken the symbol if an individual is affected by colorblindness.
- This couple will produce gametes with what kind of alleles?
 Woman: _____ Man: _____
- This couple has three children whose sex and color-vision acuity are determined by your coin tosses.
 - The *first coin toss* will be to determine which chromosome (X or Y) the father contributes. Use heads for the X chromosome and tails for the Y chromosome.
 - The *second coin toss* will be to determine which of the two X chromosomes the offspring receives from the mother. Use heads for an X chromosome with a dominant allele and tails for an X chromosome with a recessive allele.

Offspring 1

Chromosome contributed by the father? _____ (determines sex of offspring)

Chromosome contributed by the mother? _____

Genotype of offspring? _____

Offspring 2

Chromosome contributed by the father? _____ (determines sex of offspring)

Chromosome contributed by the mother? _____

Genotype of offspring? _____

Offspring 3

Chromosome contributed by the father? _____ (determines sex of offspring)

Chromosome contributed by the mother? _____

Genotype of offspring? _____

d. Add these three offspring to your pedigree, and write the genotypes next to the symbols for each.

e. "Create" mates for each of these offspring—you will know the sex of the mate, because it will be opposite from the offspring. Flip a coin to determine whether a male mate carries a *C* or a *c* on his X chromosomes, and to determine whether a female carries a *C* or a *c* on each of her X chromosomes. Draw these mates on your pedigree.

f. Produce one offspring for each of these second-generation couples by coin-tossing for sex and to determine which alleles are passed on for the color vision trait. Depending upon the parents' genotypes, you may have to toss either more or fewer coins.

Record the genotypes for your third generation.

Offspring Couple 1

Offspring Couple 2

Offspring Couple 3

g. Add them to your pedigree.

h. Compare your three generations to those of at least three lab partner pairs, and briefly comment on those comparisons.

SELF - TEST 5 . 2

NAME _____ SECTION _____ DATE _____

1. Hemophilia is a rare, sex-linked recessive trait. It may help to make a Punnett square to answer some of these questions. Use the letter *H*, *h* to represent the dominant and recessive allele.
 - a. What is the genotype of a male with hemophilia?

 - b. What is the genotype of a female who is a carrier?

 - c. If a female who is a carrier mates with a normal male, what are the chances that they will have an offspring with hemophilia?

 - d. Will this offspring (with hemophilia) be a male or a female?

 - e. What are the chances of their having a carrier daughter?

2. What is the probability that a colorblind male and a colorblind female will have an offspring with normal vision?

3. A cross between a colorblind man and a woman who is a carrier will result in what “kinds” of offspring, and in what proportions?