Applied Genetics

Section 11.1
Dihybrid Crosses

Pre-View 11.1

- **Dihybrid cross** – studying two traits crossed from parent organisms
- **Law of Independent Assortment** – a natural law that explains how traits are inherited independently of other traits

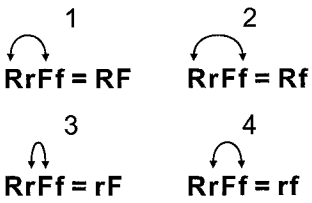
So far we have looked at monohybrid crosses, which only study one trait at a time. Many times scientists study two or more traits at a time. Studying two traits at a time is called a **dihybrid cross**.

When at least two traits at a time are studied, they illustrate the **Law of Independent Assortment**, which means that inheriting one trait doesn't affect the inheritance of another trait. For instance, having brown eyes has nothing to do with having freckles. The genes are inherited independently of each other. Again, Mendel was the first to make these observations, so this law is often called *Mendel's Law of Independent Assortment*. (You'll see later how this "law" isn't always true.)

To show a dihybrid cross, let's pick two different traits, such as handedness and freckles. Being right-handed (R) is dominant over being left-handed (r), and having freckles (F) is dominant over not having freckles (f).

Let's say that both the mother and the father are heterozygous for both traits. We can create a Punnett square of this dihybrid cross to analyze how these traits might appear in their children. The Punnett square is bigger and looks more complicated, but it really isn't difficult to do. Just follow these steps.

Step 1: First, pick a parent. The mother is heterozygous for both traits, (RrFf), so she has a dominant R gene and a recessive r gene. She also has a dominant F gene and a recessive f gene. She can pass on each of these traits independently to her children. What are the four possible combinations of these traits? They are RF, Rf, rF, and rf.



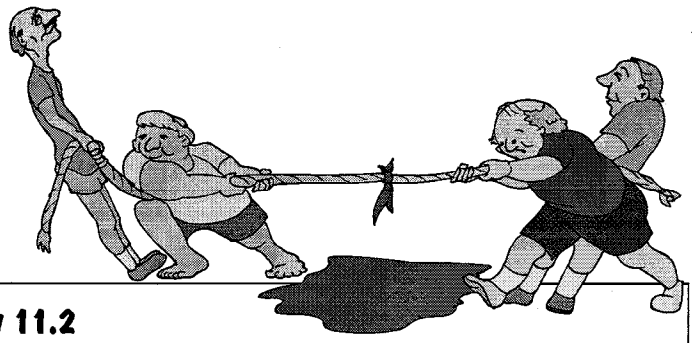
Step 2: Next, consider the father. He is also heterozygous for both traits, so the combinations of traits are the same: RF, Rf, rF, and rf.

Step 3: Now fill in a Punnet square with these combinations. Since there are four combinations for each parent, the square is 4 × 4 instead of 2 × 2. But you fill it in the same way.

		Mother			
		RF	**Rf**	**rF**	**rf**
Father	**RF**	RRFF	RRFf	RrFF	RrFf
	Rf	RRFf	RRff	RrFf	Rrff
	rF	RrFF	RrFf	rrFF	rrFf
	rf	RrFf	Rrff	rrFf	rrff

Applied Genetics

Section 11.2
Incomplete Dominance and Codominance



Pre-View 11.2

- **Incomplete dominance** – having alleles that do not have complete dominance so the resulting trait is a mix of two alternate traits; for example, red snapdragons crossed with white snapdragons produce pink snapdragons
- **Codominance** – having two or more alleles that are equally dominant for a trait so that both traits are expressed; for example, roan cattle have both red hair and white hair (not pink)
- **Sickle cell anemia** – a genetic disease caused by a codominant gene

Although Mendel learned a lot about genetics, all of the traits that he studied had one dominant allele and one recessive allele. Unfortunately, it's not always that easy. Scientists have learned a lot of new stuff that Mendel didn't know. There are many things that affect how characteristics are inherited. In this sub-section, we'll look at two cases that are exceptions to Mendel's rules: incomplete dominance and codominance.

Incomplete Dominance

Some organisms have traits whose alleles show **incomplete dominance**, which means that neither allele for a particular trait is dominant over the other. Snapdragons are plants that show incomplete dominance in their flower color. If snapdragons with red flowers are crossed with snapdragons with white flowers, the phenotype of the offspring is between the two colors. They will have pink flowers, just like mixing red paint and white paint to get pink paint.

When you complete a Punnett square for incomplete dominance, you don't use lowercase letters since neither allele is recessive to the other one. Instead, a capital letter is used for one allele, and the same capital letter with an apostrophe or tic mark after it represents the other allele. (Sometimes two different capital letters are used.) Figure 11-1 shows a Punnett square for the incomplete dominance of snapdragons.

How can you tell if you are looking at a cross that shows incomplete dominance? First, there are three different phenotypes — one for each parent and a different one for the offspring. Second, the phenotype of the offspring will be a mix or blend of the parents' phenotypes.

Incomplete Dominance

	R	R
R′	RR′	RR′
R′	RR′	RR′

R – Red allele
R′ – White allele
RR′ – Pink flowers

Fig. 11-1

Codominance

At first, **codominance** looks somewhat the same as incomplete dominance, but in codominance, both alleles are expressed equally. Instead of neither allele being dominant, both alleles are dominant. Both parental phenotypes appear in the offspring together but not mixed. For example, in certain kinds of chickens, black feathers and white feathers have codominant alleles. When you cross a black chicken and a white chicken, you get offspring that are black and white speckled.

In a Punnett square, codominance does not use lowercase letters either. To distinguish codominance from incomplete dominance, sometimes superscripts are used. An example of a Punnett square for codominance is given in figure 11-2.

Codominance

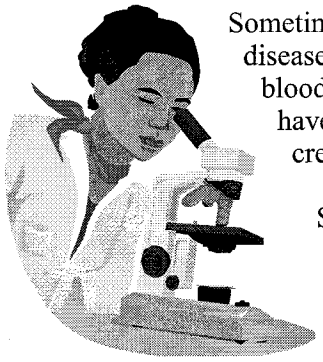
	F^B	F^B
F^W	F^BF^W	F^BF^W
F^W	F^BF^W	F^BF^W

F^B – Black feather allele
F^W – White feather allele
F^BF^W – speckled feathers

Fig. 11-2

Roan color in cattle and horses is another example of codominance. RR produces all red hairs, and WW produces all white hairs. RW produces roan fur, which has red and white hairs all mixed together. Notice the hairs aren't pink, like the snapdragon flowers' example of incomplete dominance. In codominance, some hairs are red and some are white. (Notice that in this case, two different capital letters are used. Two capital letters may be seen when showing incomplete dominance or codominance.)

Sickle Cell Anemia



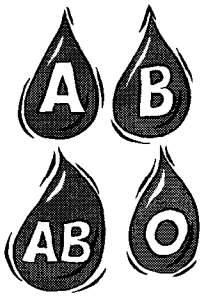
Sometimes genetic diseases can be caused by a codominant gene. An example of a codominance disease trait in humans is **sickle cell anemia**. The sickle cell trait causes the hemoglobin in red blood cells to be the wrong shape. Hemoglobin is a protein that carries oxygen. If it doesn't have the right shape, the red blood cell changes shape. Instead of being round, the cells are crescent shaped. The deformed cells can stick together and block small blood vessels.

Sickle cell anemia is most common in people of African ancestry, but it can occur in other ethnic groups, especially Mediterranean and Middle Eastern. An estimated 2 million Americans carry one gene for the sickle cell trait. If someone is heterozygous for the sickle cell gene (has one defective gene and one normal gene), then they produce enough normal red blood cells to live normal lives. They don't usually have sickle-cell problems unless they are at very high altitudes or other places where oxygen is less available.

People who are homozygous for sickle cell, however, have very serious health problems. The affected person may feel tired and weak, and the disorder increases the risk of stroke and infections. The sickle cells also die quickly, so the person can become anemic. Similar to testing for PKU, testing for sickle cell anemia is now required in almost every state. Although there is no cure for sickle cell, doctors are able to treat many of the symptoms.

Multiple Alleles

So far we've seen examples of traits that are determined by only two different alleles. Although each individual will have only two alleles per trait, it is possible to have more than two alleles that can determine a trait. Let's look at a common example below, human blood type.



Do you know what type of blood you have? Humans have four main blood types: A, B, AB, and O. These blood types are determined by multiple alleles. There are three alleles for the blood type gene instead of two, but you have only two in your cells — one from your mother and one from your father. How do you get four blood types from three alleles?

1. Each person has two alleles — one from the mother and one from the father.
2. There are three alleles for blood type — I^A, I^B, and i.
3. I^A and I^B are codominant to each other.
4. Both I^A and I^B are dominant over i, which is recessive.

Figure 11-3 shows the different human blood types and the possible genotypes for each.

Blood Type	Genotypes
A	I^AI^A or I^Ai
B	I^BI^B or I^Bi
AB	I^AI^B
O	ii

Fig. 11-3

Blood typing is important because a person getting a blood transfusion can die if the wrong type of blood is given. The wrong blood type causes the red blood cells to clump together, and the person dies. Blood typing is also important in paternity disputes. It can't prove that a specific man is the father of a child, but sometimes it can prove that a certain man is *not* the father of a child.

Example 1: If a child has type AB blood and the mother has type A blood, could a man with type O blood be the father?

We know that the child's genotype is I^AI^B and that the man's genotype is ii. The mother's genotype is either I^AI^A or I^Ai. This man could not be the father because he couldn't give the child an I^B allele to make the AB blood type. The possibilities may be easier to see by using a Punnett square. The Punnett square shows that a mother having a genotype of I^AI^A or I^Ai and a father having a genotype of ii will NOT give a child the genotype I^AI^B.

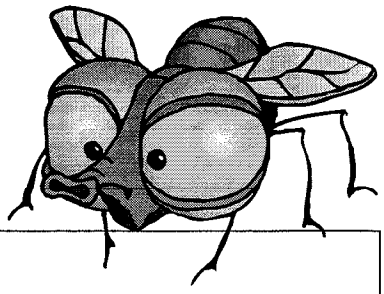
	I^A	I^A or i
i	I^Ai	I^Ai or ii
i	I^Ai	I^Ai or ii

Example 2: A mother has type A blood, and a father has type B blood. Can they have a child with type O blood? Why or why not?

This couple can have a child with O blood IF the parents' genotypes are I^Ai and I^Bi. In this case, each parent could contribute the recessive i gene, which would result in type O blood.

Applied Genetics

Section 11.3
Linked and Sex-Linked Genes



Pre-View 11.3

- **Law of Independent Assortment** – a natural law that explains how traits are inherited independently of other traits
- **Sex chromosomes** – the one pair of X and Y chromosomes that determines gender
- **Sex-linked genes** – genes that are found on either the X or the Y sex chromosome
- **Red-green color blindness** – a sex-linked disorder that is carried on the X chromosome
- **Hemophilia** – a sex-linked disease that is carried on the X chromosome

Revisiting the Law of Independent Assortment

Mendel looked at seven different traits in pea plants: pea shape, seed color, flower color, flower position, pod color, pod shape, and plant height. As he studied the inheritance of two traits, he hypothesized that each allele would segregate (separate) independently during gamete formation. In the case of seed color and seed shape, he determined that the alleles do separate independently of each other. As you saw in Section 11.1, his hypothesis became known as the **Law of Independent Assortment**.

Mendel was no dummy; the Law of Independent Assortment is true most of the time, but there are some exceptions. Remember, Mendel didn't know about genes and chromosomes. The Law of Independent Assortment is true when the genes are on different chromosomes. It is not always true for genes on the same chromosome because two genes that are close enough together on the same chromosome may be **linked** — that is, they may be inherited together.

Genes are found at different locations along a chromosome. Look at figure 11-4. To use a very simple example, Section A may represent the location of gene A. Section B would have gene B, and so on. Remember that during meiosis, homologous chromosomes (the ones from each parent) line up side by side (as seen in figure 11-4). The genes C and D are close together. Even if crossing over occurs, these two genes may be exchanged together. See how C and D stay together? Genes A and D, on the other hand, may end up on different chromosomes if crossing over occurs. The closer two genes are on the same chromosome, the more likely they are to be linked together. For example, in humans the characteristics for red hair and freckles seem to be linked.

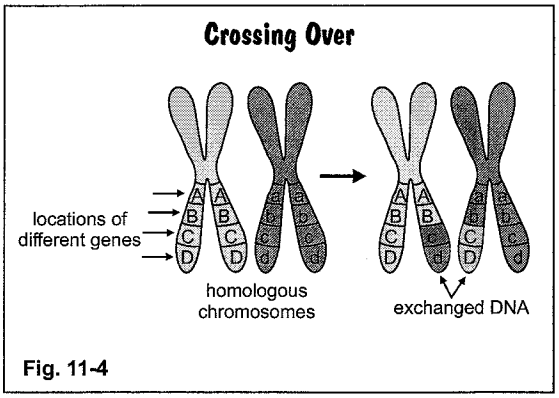


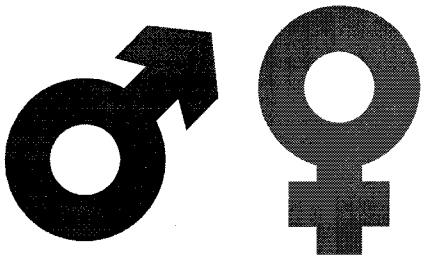
Fig. 11-4

The Study of Linked Genes

Who discovered linked genes? In the early 1900s, a scientist named **Thomas Morgan** and his graduate students were studying a little insect called *Drosophila*, or fruit fly. They found that the fruit flies had several genes that did not show independent assortment. Fruit fly body color could be gray (G) or black (g), and wings could be long (L) or vestigial (l). (Vestigial wings are so small that they don't really function as wings.) Morgan found that the flies that were gray tended to have long wings and the flies that were black tended to have vestigial wings. He found that these genes are so close on the same chromosome that they tend to be inherited together. Later studies found that many other organisms, including humans, have linked genes.

Sex Chromosomes

Humans have 23 pairs of chromosomes (for a total of 46 chromosomes). You've already seen how the autosomes pass on traits. Now let's look at the other pair of chromosomes, the sex chromosomes. Humans have 22 pairs of autosomes and one pair of sex chromosomes. The **sex chromosomes** are called the X and Y chromosomes, and they determine gender. Human females have two X chromosomes, and males have one X chromosome and one Y chromosome. The X chromosome is larger than the Y chromosome, so it has more genes on it.



Sex-Linked Genes

In humans the X and Y chromosomes determine gender, but they also have genes on them that determine other characteristics. These genes are called **sex-linked genes** because they are found on either the X chromosome or the Y chromosome. Because the X chromosome has more genes on it, most sex-linked genes are X-linked. Don't confuse sex-linked genes with the linked genes mentioned earlier. Linked genes are on *autosomes*.

Color Blindness

A defective gene on either the X or the Y chromosome may result in a sex-linked disorder or disease. A common sex-linked disorder is red-green color blindness. **Color blindness** is more correctly called *color-deficiency* because most people with this disorder can see some colors. It occurs when there is a problem with the cone cells in the eye, which sense colors. The most common form is red-green color deficiency, where a person has trouble telling the difference between red and green. Another form is blue-yellow deficiency. The deficiency may be so mild that a person doesn't know if he is affected or so severe that the person can't see any color at all.

Both males and females can be color-deficient although it is rare in females. Why? The gene for color vision is carried on the X chromosome. Since color deficiency is a recessive trait, a female would have to inherit one gene from her mother and one gene from her father to be color deficient. If she has one normal color vision gene, then she will have normal color vision. A male, on the other hand, has only one X chromosome. If he inherits the color deficient gene from his mother, he automatically will have color deficient vision since his father can't give him a dominant gene for normal color vision. (The Y chromosome doesn't have a gene for color vision at all.) As a result, about 10% of males have some form of color deficiency.

The allele for normal color vision is shown by X^B, and color blindness is shown by X^b. The chart below shows the possible genotypes and phenotypes:

	GENOTYPE	PHENOTYPE
Males	$X^B Y$	normal color vision
	$X^b Y$	color blind
Females	$X^B X^B$	normal color vision
	$X^B X^b$	normal but a carrier
	$X^b X^b$	color blind

If a male gets the color blindness gene, he will be color blind. He would inherit the gene from his mother. Females are color blind only if they get two genes for color blindness — one from each parent.

Using capital and lowercase letters as superscripts, as shown above, is a common way to indicate sex-linked traits. However, a sex-linked gene may be indicated by X′. For example, a normal female would be shown as XX, a carrier female as XX′, a normal male as XY, and an affected male as X′Y. The apostrophe indicates the presence of a sex-linked gene.

Example: If a woman who has normal color vision but who carries the recessive allele for color blindness marries a man with normal color vision, will their children have normal color vision?

To answer this question, fill in a Punnett square to see the possible genotypes in the offspring. Notice that two out of the four offspring represent daughters and the other two represent sons.

	X^B	X^b
X^B	X^BX^B normal daughter	X^BX^b carrier daughter
Y	X^BY normal son	X^bY affected son

To consider the daughters, look at only the possibilities with two X's. Since both possibilities for daughters has one normal X^B gene, all of the girls would have normal color vision. However, one out of two of the daughter possibilities has an X^b gene, which means that there is a 50% chance that a girl will carry the color blind gene.

Now consider the sons, which have an XY. Can you see that if the couple has a boy, there is a 50% chance that he would be color blind?

Hemophilia

Human blood contains proteins called *clotting factors* that stop bleeding when a person gets a cut or a scratch. People with **hemophilia** don't have enough of one of these proteins and will bleed for a longer period of time than those with the normal level of proteins. They don't bleed faster, just longer. They have to be concerned about internal bleeding, especially into muscles and joints. Like color vision, the gene is carried on the X chromosome, so it affects men more than women although women are carriers.

Applied Genetics

Section 11.4
Pedigrees

Pre-View 11.4

- **Pedigree** – a diagram used by geneticists to chart a trait from one generation to another

Have you ever seen your family tree? You know — a family tree is a diagram that shows different generations and various members of your family. Geneticists use a diagram called a **pedigree** that is similar to a family tree to show genetic inheritance. Pedigrees can help geneticists determine if a trait is inherited, they can show how a trait is passed from one generation to the next, and they can help determine whether an allele for a trait is dominant or recessive.

Pedigrees always use certain symbols that you should know. Study the sample pedigree given below.

Sample Pedigree of a Family with Color Blindness

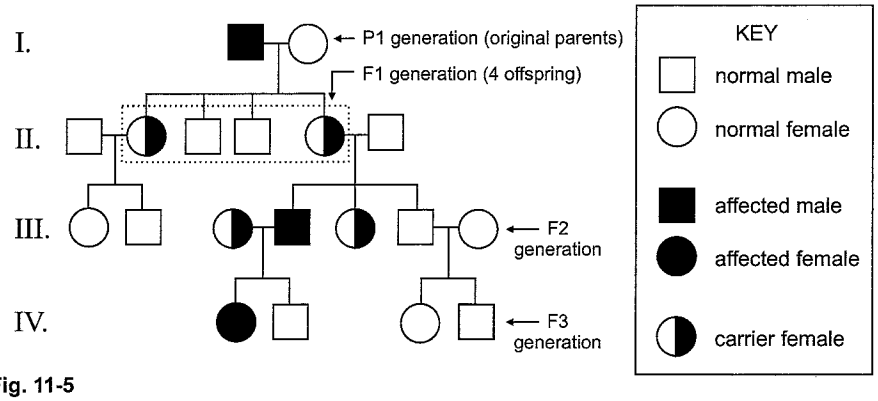


Fig. 11-5

Each horizontal row represents one generation, and the youngest generation is at the bottom. The rows are usually labeled with Roman numerals. Row I corresponds to the first generation (P1), row II corresponds to the children of the first generation and their spouses (F1), row III represents their grandchildren (F2), and so on. The horizontal line between a square and a circle represents parents. The vertical line between the parents that link to the next generation shows the offspring of the parents.

In the first generation (P1) of the sample above, the father is affected by color blindness, and the mother is normal. The parents have four children, 2 sons and 2 daughters (enclosed by the dotted line). Both sons are normal, but both daughters are carriers.

Example 1: In the pedigree given in figure 11-5, determine the genotypes of the parents in the first generation. The sons are normal, but the daughters are carriers. If the first generation parents had additional children, could the genotypes of sons and daughters be different?

We know that color blindness is an X-linked gene. Since the man in the P1 generation is affected, we know that his genotype is X^bY. The woman in the first generation is normal, so her genotype is X^BX^B. Use a Punnett square to see the possible genotypes of offspring.

	X^b	Y
X^B	X^BX^b	X^BY
X^B	X^BX^b	X^BY

From the Punnett square we can see that these parents will always have daughters that are carriers and sons that are normal.

Example 2: In the pedigree given in figure 11-5, both carrier daughters in the second generation marry men who are normal. One of these women has a son and a daughter in the third generation who are both normal. The other woman has a son who is color blind, a son who is normal, and daughter who is a carrier. Use a Punnett square to show that all these genotypes are possible.

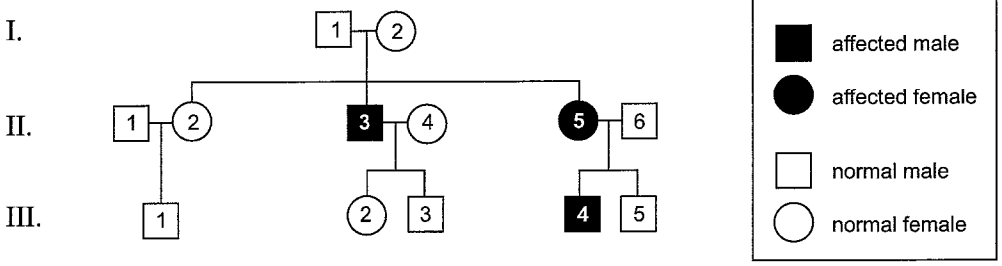
Both affected daughters in the second generation (F1) have a genotype of X^BX^b. The normal men that they marry have a genotype of X^BY. In the Punnett square, you can see that daughters born to these parents can either be normal (X^BX^B) or a carrier (X^BX^b). Sons can either be normal (X^BY) or color blind (X^bY).

	X^B	Y
X^B	X^BX^B	X^BY
X^b	X^BX^b	X^bY

The carriers of a genetic disorder may not be known and may not be shown on a pedigree. The purpose of a pedigree may be to discover who is a carrier. Look at another example.

Example 3: Study the pedigree below. What type of genetic disorder is indicated by this pedigree?

Pedigree for a Genetic Disorder



The parents in generation I have three children, two boys and a girl. One of the girls is normal, but the boy and the other girl are affected. The logical conclusion is that both parents are a carrier of the genetic disorder. If so, this disorder is autosomal recessive. You know that it isn't a sex-linked disorder since neither parent is affected and it is passed to both a son and a daughter. If it had been passed only to one or more sons, you may suspect that the mother is a carrier of a sex-linked disorder.

Example 4: For the pedigree given in Example 3, what must be true of individual II6? If the genotype of II5 is bb, what are the genotypes of individuals III2, III3, and III5?

Parent II5 is an affected female that has a son who is also affected. Parent II6 must be a carrier since this disease is recessive. Children of an affected individual, like individuals III2, III3, and III5, will be carriers of the disease since they will get one "disease" gene from the affected parent. Their genotype would be the same — Bb, or heterozygous for the disease.