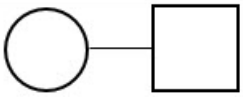
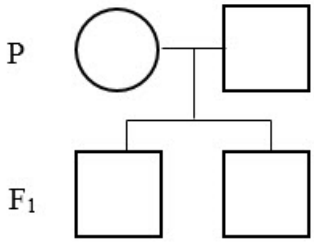
PEDIGREES

Now that you understand different types of inheritance, we can now discuss a diagram which is used to show the occurrence of a trait over several generations of a family. A **Pedigree** is the diagram that shows how a trait is inherited over several generations within a family. A genetic disorder can be traced within a family using a pedigree. Sometimes parents may want to know the likelihood of their children inheriting a disorder that may run within their family's past. Using a pedigree can also help parents determine if they are carriers, because, remember, carriers do not show any symptoms of a genetic disorder while they carry the allele for the disorder.

Symbols used in Pedigrees

Table 26-1: *Pedigree Symbols*

	Male
	Male with a trait
	Female
	Female with a trait
	Female Carrier (not always used)
	A cross or mating
	Offspring of parents

Refer to **Table 26-1** as you read this section of the symbols used in pedigrees. A male is drawn as a square. If the square is empty or not shaded he is considered normal or as not having a trait. A square that is shaded represents a male with the particular trait. A female is drawn as a circle. If the circle is empty or not shaded she is considered normal or as not having a trait. A circle that is shaded represents a female with the particular trait. A female can also be drawn as half-shaded. If her symbol is half shaded, she is considered a carrier. A carrier symbol is not always used. If the carrier symbol is not used, then she will be drawn as an empty symbol because she is a carrier. Remember, carriers are considered normal and only carry the trait with no symptoms or signs of carrying it. A horizontal line connecting a male symbol and female symbol represents a crossing or mating between the two parents. A vertical line drawn down from the horizontal line of crossing or mating connects the offspring of the parents. The offspring are normally listed in order of birth from left to right. Notice in Table 26-1 how "P" and "F_1" are used in the example next to *Offspring of parents*. The "P" identifies the parents of the pedigree. The P generation may also be listed as the Roman numeral "I" for the first generation. The "F_1" (known as first filial generation) identifies the offspring of the parents. The "F_1" may also be listed as the Roman numeral "II". Every generation after the F_1 would be given the next number in sequence whether it is listed as filial generations (F_2, F_3, etc.) or roman numerals (III, IV, etc.).

We will now examine three different pedigrees: autosomal recessive, autosomal dominant, and x-linked (sex-linked on the X chromosome) recessive. After completing the remainder of this unit, you should be able to determine the genotype and phenotype of each individual in a pedigree, explain how an individual acquired their alleles and predict possible future generations.

Autosomal Recessive Pedigree

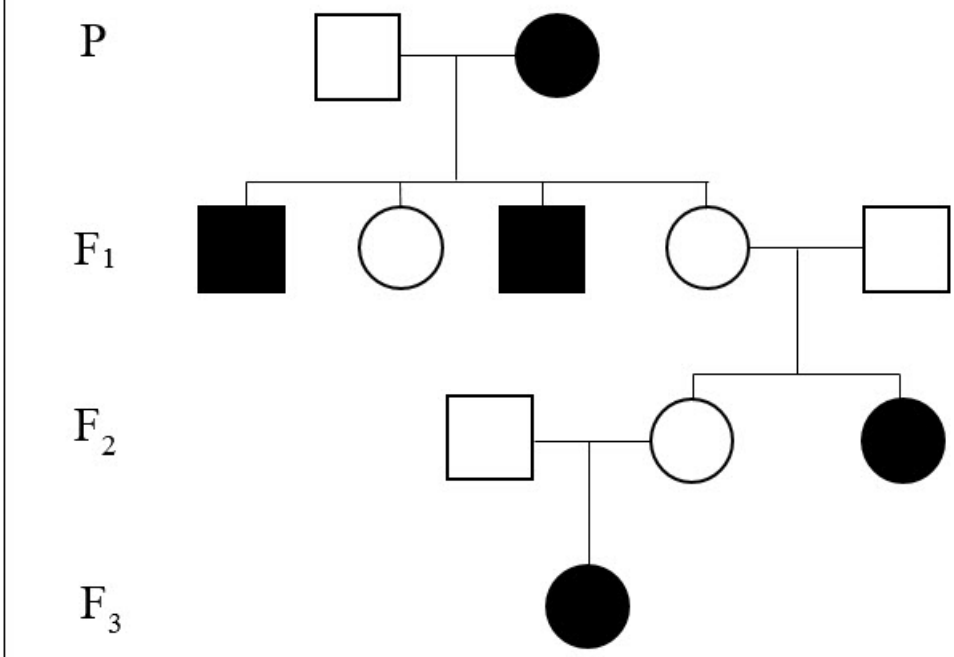
The pedigree below displays how the autosomal recessive allele for attached earlobes has been passed from generation to generation within a particular family. Free Earlobes are earlobes that hang below the point of attachment to the head. An Attached Earlobe is one that is directly attached to the skin of the jaw. An attached lobe is recessive and free lobe is dominant.

Phenotype	Genotype
Free Earlobe	EE or Ee
Attached Earlobe	ee

Interesting Note:
There is currently a debate about whether free vs attached earlobes is truly a complete dominance autosomal recessive trait. Some are arguing that there are many variations between these two phenotypes and that this character is not controlled by a single gene. For our purposes, we will go with the idea that this character is controlled by one gene and there are only two phenotypes.

Autosomal Recessive Pedigree

Dominant Trait = Free Lobes , Recessive Trait = Attached Lobes

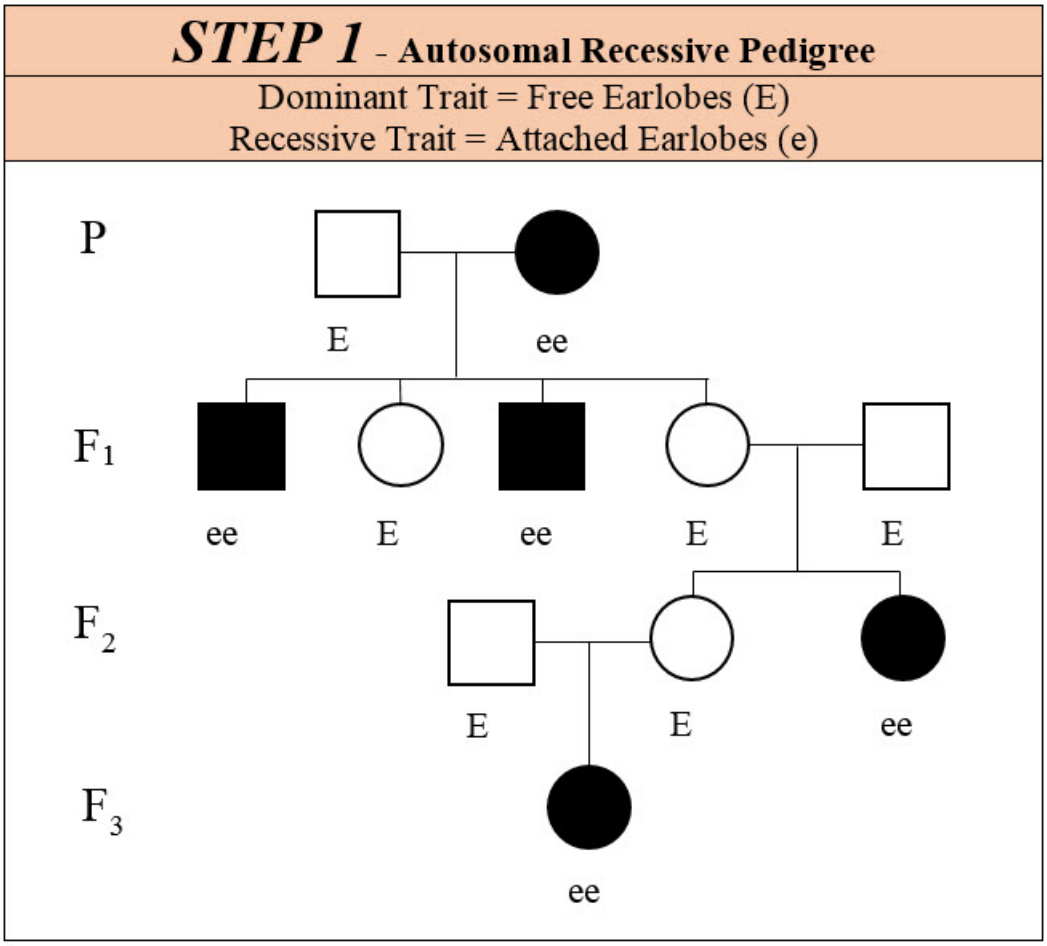


Notice in the autosomal recessive pedigree above that there are 4 generations. The parents that began this pedigree had 4 children (2 males and 2 females). The second female, from the parents in the first generation, had 2 children (2 females) with a male from another pedigree. The first female in the F_2 generation (generation III) had one child (female) with a male from another pedigree. Recognize that the parents that started this pedigree had 4 children (2 males and 2 females - F_1), 2 grandchildren (2 females – F_2), and 1 great grandchild (female - F_3).

Some generalities about autosomal recessive pedigrees:

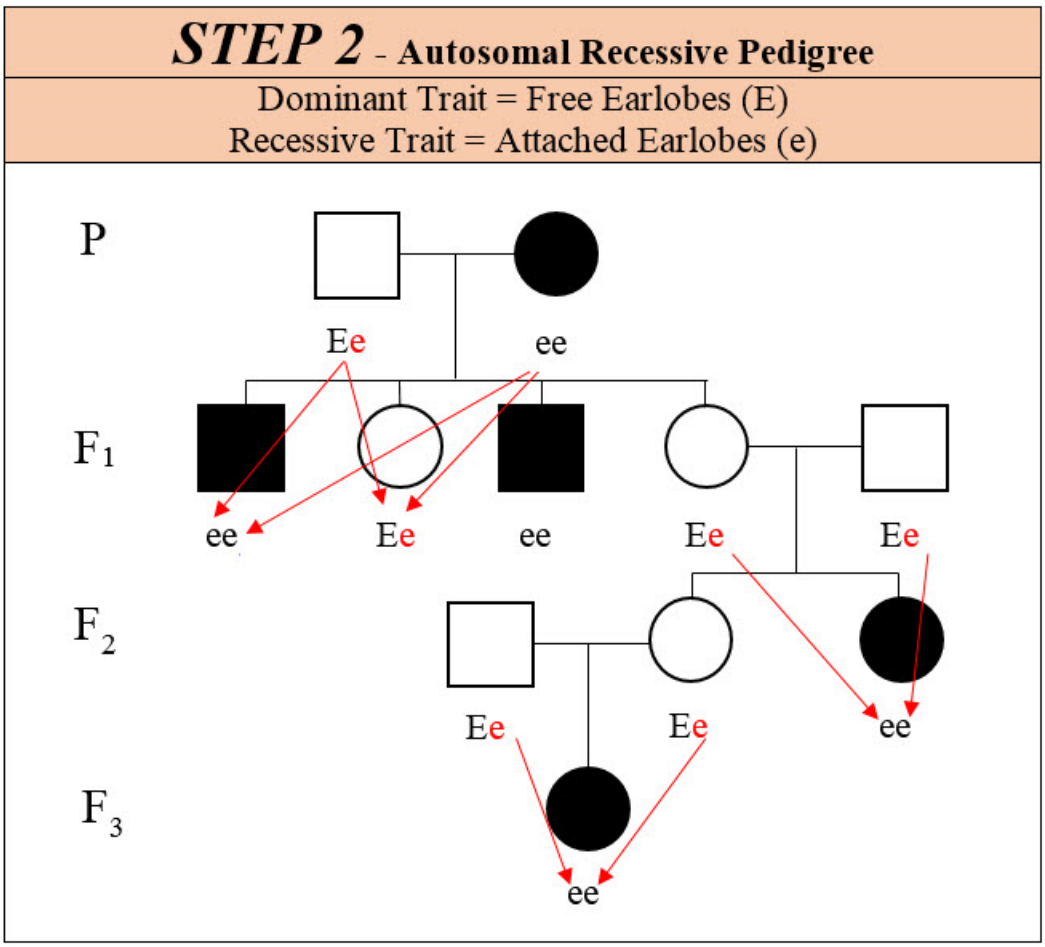
- Unaffected parents can have affected offspring *(notice how the parents in the F_2 generation did not have the trait but their child does have the trait)*.
- Affected offspring can be male and female *(notice how two males and two females are affected in this pedigree)*.
- The recessive phenotype/genotype can skip generation *(notice how the trait skipped the F_2 generation then reappeared in the F_3 generation)*.

Now we will attempt to determine every individual's genotype and phenotype. Step 1, we will determine the genotype of every individual that we are certain of by how the symbols are drawn for the recessive trait. Therefore, every shaded symbol will be of the genotype "ee" because it is a recessive trait and if they are shaded that means they have the trait. Every other symbol that is not shaded will have the genotype of either "EE" or "Ee". For now, all we know is that each empty symbol has an "E" in common.



Step 2, we need to determine the second allele of every empty symbol. Remember that each individual needs two alleles, one from the mother and one from the father. To determine the second allele we need to examine the offspring and parents together. Follow the arrows in the pedigree below to understand how the second allele is determined.

Again, it is very important to understand that the two alleles each individual has are the result of one allele from the mother and one allele from the father. Once you have determined that a parent gives one specific allele, you can no longer look to that parent to give another allele to the same child, **for example** the first female in the F_1 generation. We know she has an "E" because her symbol is empty. Looking at her parents we know she inherited the "E" from her father, because the mother does not have an "E" to give. When we then look to the mother we see that all she can give to her daughter is an "e", so the daughter inherited "E" from her father and "e" from her mother to make her have the genotype "Ee".



Autosomal Dominant Pedigree

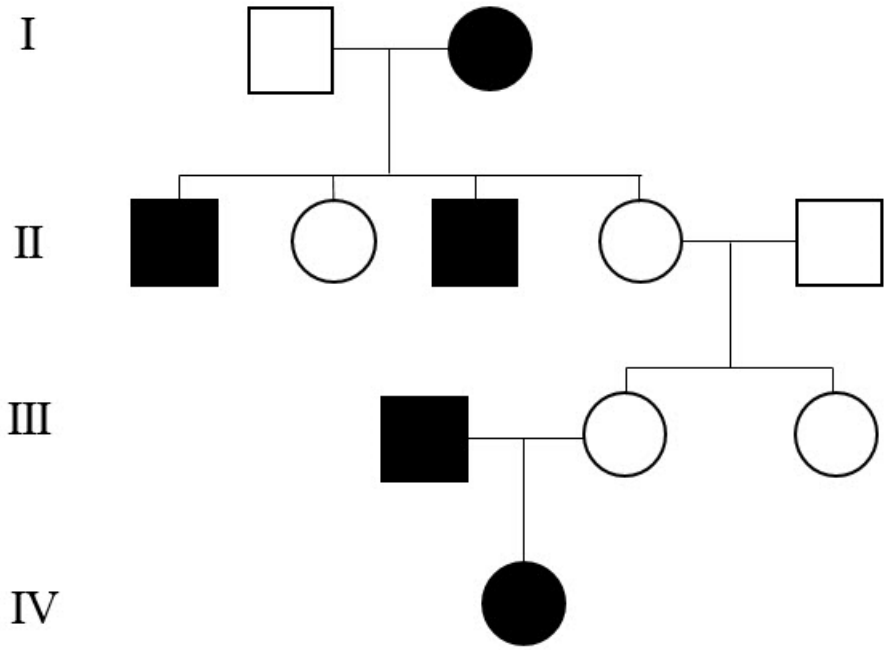
The pedigree below displays how the autosomal dominant allele for widow's peak has been passed from generation to generation within a particular family. Widow's Peak is a V-shaped point in the hairline in the center of the forehead. Widow's Peak is dominant and Straight Hairline (no widow's peak) is recessive.

Phenotype	Genotype
Widow's Peak	WW or Ww
Straight Hairline	ww

Interesting Note:
There is currently a debate about this character also, just as with the earlobes. For our purposes, we will go with the idea that this character is controlled by one gene and there are only two phenotypes.

Autosomal Dominant Pedigree

Dominant Trait = Widow's Peak
Recessive Trait = Straight Hairline

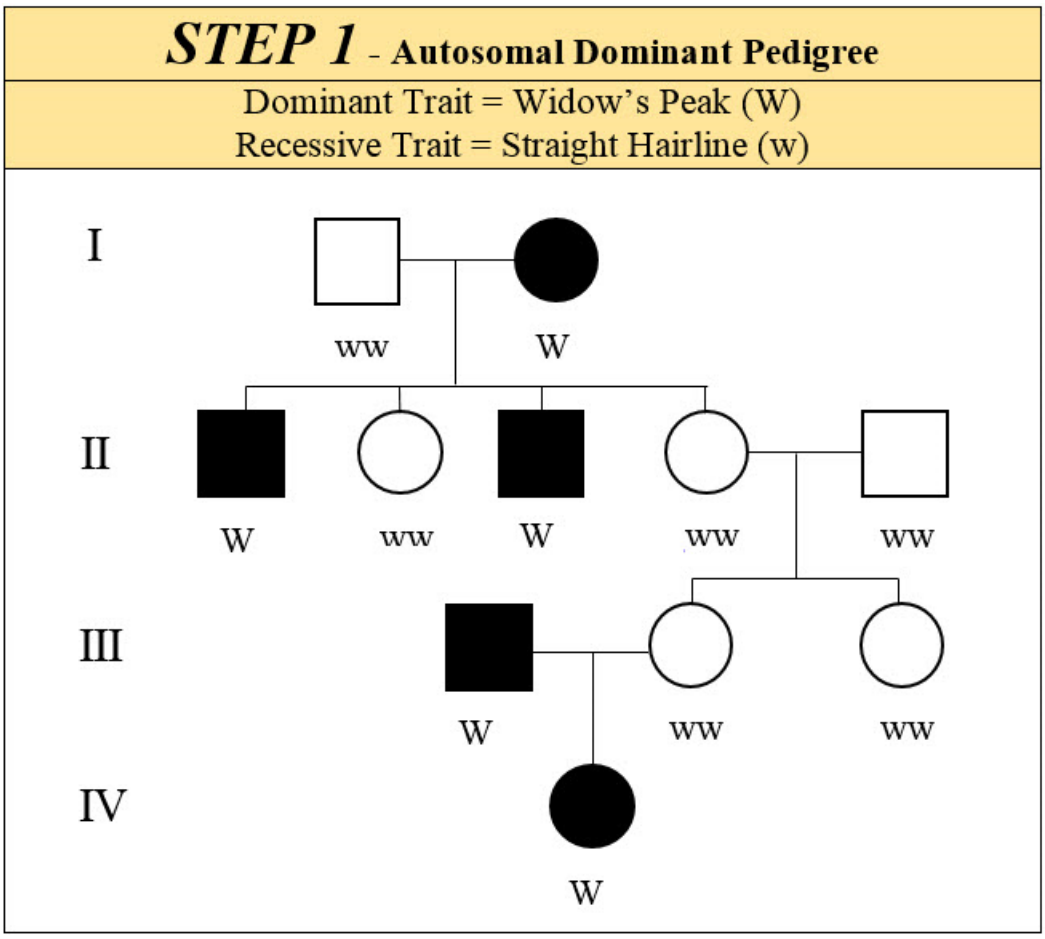


Notice in the autosomal dominant pedigree above that there are 4 generations. The parents that began this pedigree had 4 children (2 males and 2 females). The second female, from the parents in the first generation, had 2 children (2 females) with a male from another pedigree. The first female in generation III had one child (female) with a male from another pedigree. Recognize that the parents that started this pedigree had 4 children (2 males and 2 females – Generation II), 2 grandchildren (2 females – Generation III), and 1 great grandchild (female – Generation IV).

Some generalities about autosomal dominant pedigrees:

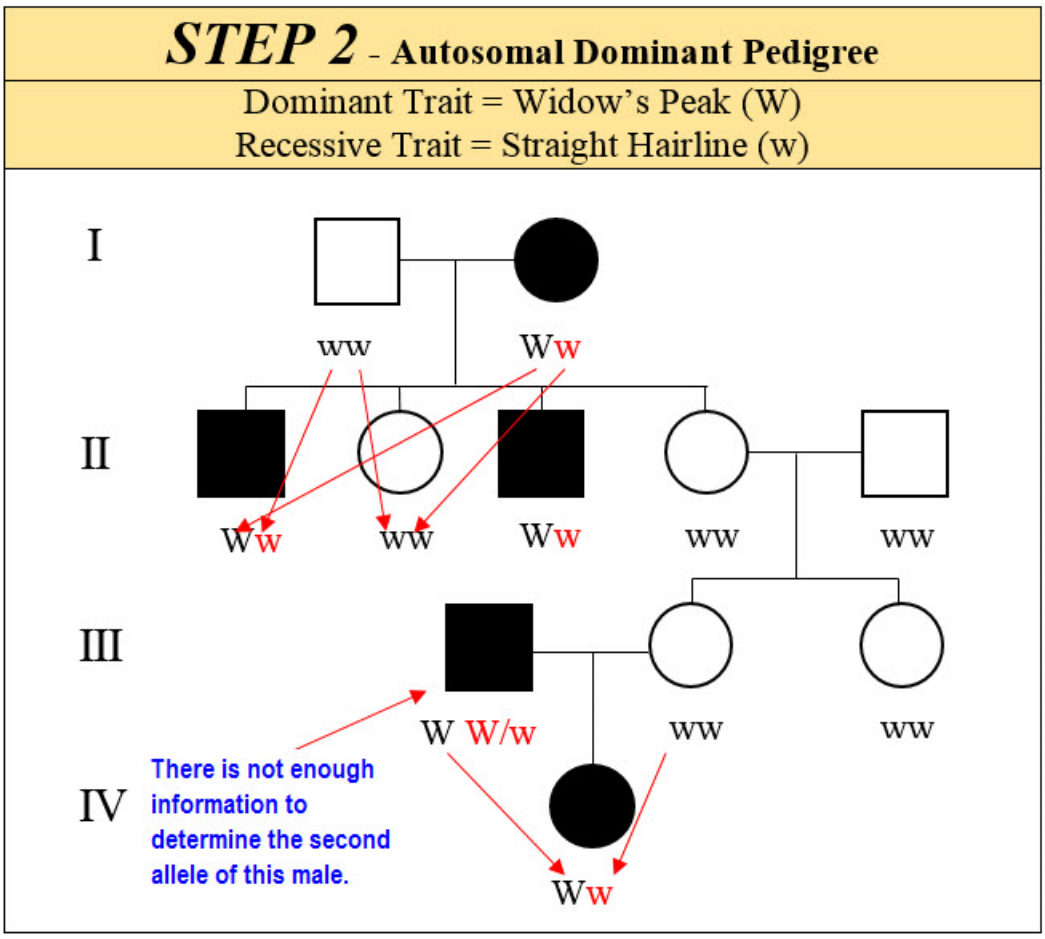
- Affected individuals have at least one affected parent *(notice how the two males in generation II have a mother with the trait, and the female in generation IV has a father with the trait)*.
- Two unaffected parents can only have unaffected offspring *(notice how the parents in generation II had two female offspring (generation III) neither of which have the trait)*.
- The dominant phenotype/genotype usually appears in every generation *(notice how every generation has at least one individual with the dominant trait to keep it running in the family)*.

Now we will attempt to determine every individual's genotype and phenotype. Step 1, we will determine the genotype of every individual that we are certain of by how the symbols are drawn for the dominant trait. The fact that this is a dominant trait means that we need to switch our thinking from what we did in the last recessive pedigree. Therefore, every shaded symbol will now be of the genotype "WW" or "Ww" because it is a dominant trait and if they are shaded that means they have the trait. Every other symbol that is not shaded will now have the genotype "ww". For now, all we know is that each empty symbol is "ww" and every shaded symbol has a "W" in common.

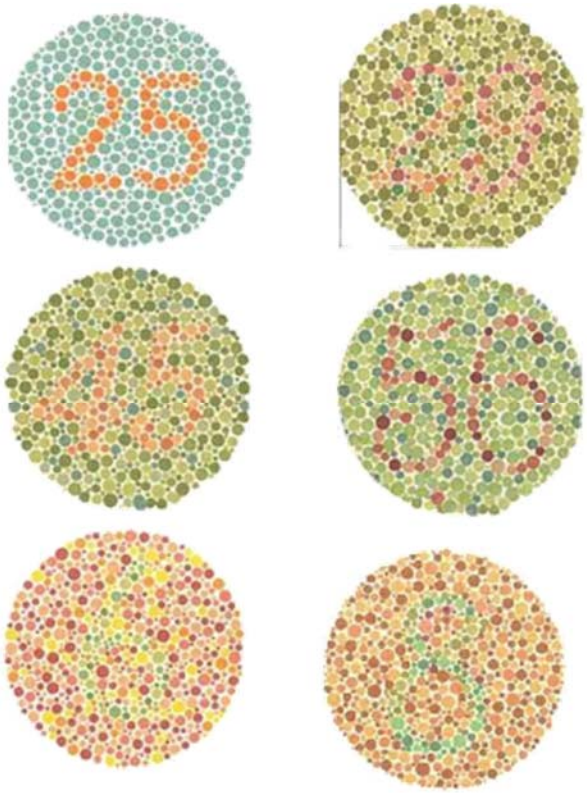


Step 2, we need to determine the second allele of every shaded symbol. Remember that each individual needs two alleles, one from the mother and one from the father. To determine the second allele we need to examine the offspring and parents together. Follow the arrows in the pedigree below to understand how the second allele is determined.

Again, it is very important to understand that the two alleles each individual has are the result of one allele from the mother and one allele from the father. Once you have determined that a parent gives one specific allele, you can no longer look to that parent to give another allele to the same child, **for example** the first male in generation II. We know he has a "W" because his symbol is shaded and this is a dominant trait pedigree. Looking at his parents we know he inherited the "W" from his mother, because the father does not have a "W" to give. When we then look to the father we see that all he can give to his son is a "w", so the son inherited "W" from his mother and "w" from his father to make him have the genotype "Ww".



X-Linked (sex-linked on the X chromosome) Recessive Pedigree

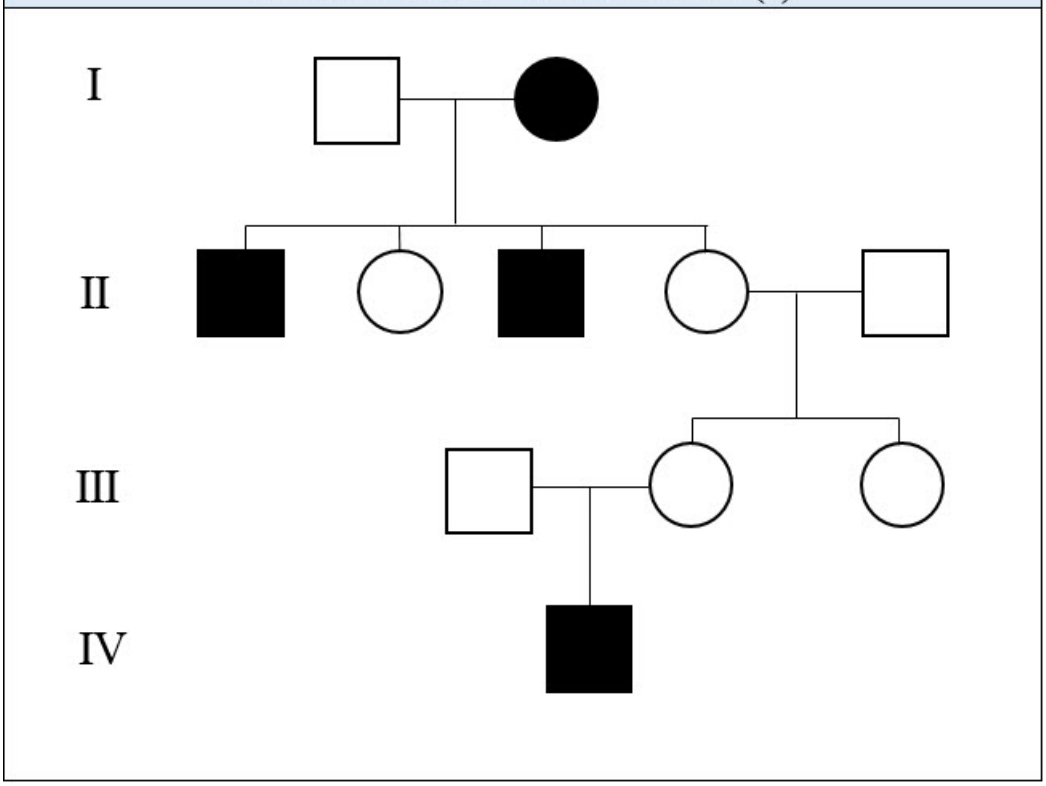


The pedigree below displays how the sex-linked recessive allele, which is carried on the X chromosome, for colorblindness has been passed from generation to generation within a particular family. Colorblindness (specifically red-green colorblindness) is the inability to distinguish red and green colors. Normal vision is dominant and colorblindness is recessive.

Keep in mind, this is a sex-linked trait, therefore, you must use the sex chromosomes (male XY and female XX).

Recessive Sex-Linked Pedigree

Dominant Trait = Normal Vision (C)
Recessive Trait = Colorblindness (c)

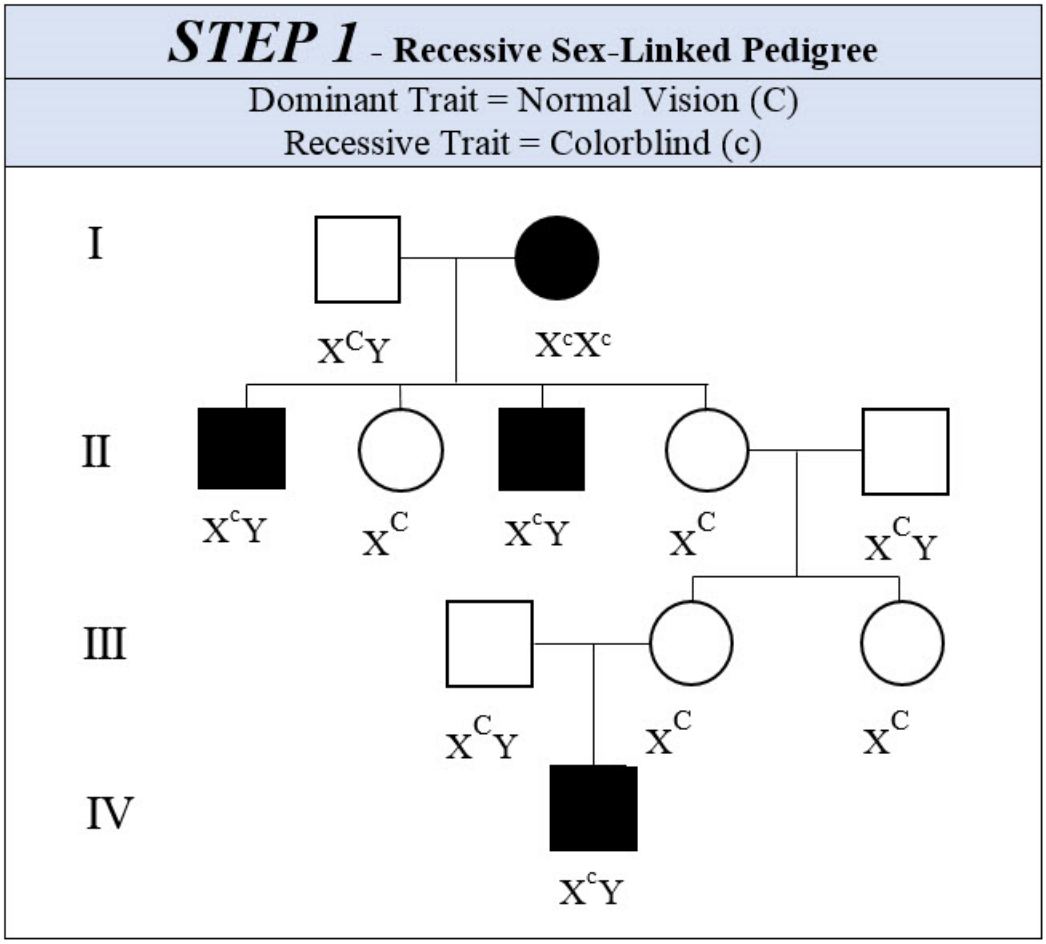


Now we will attempt to determine every individual's genotype and phenotype. Step 1, we will determine the genotype of every individual that we are certain of by how the symbols are drawn for the recessive trait, remembering to use the sex chromosomes. Therefore, we need to break down each symbol by gender.

Every shaded square will be of the genotype "X^cY" because it is a male with the colorblind recessive trait (this trait is only carried on the X's). Every other square that is not shaded will have the genotype "X^CY", because the Y chromosome carries nothing there is no other option for the male.

Every shaded circle will be of the genotype "X^cX^c" because it is a female with the colorblind recessive trait. Every other circle that is not shaded will have the genotype of either "X^CX^C" or "X^CX^c". Notice how this pedigree does not use the half-shaded circle for carrier females so we need to distinguish the empty circle for either "X^CX^C" or "X^CX^c". For now, all we know is that each empty circle has a "X^C" in common.

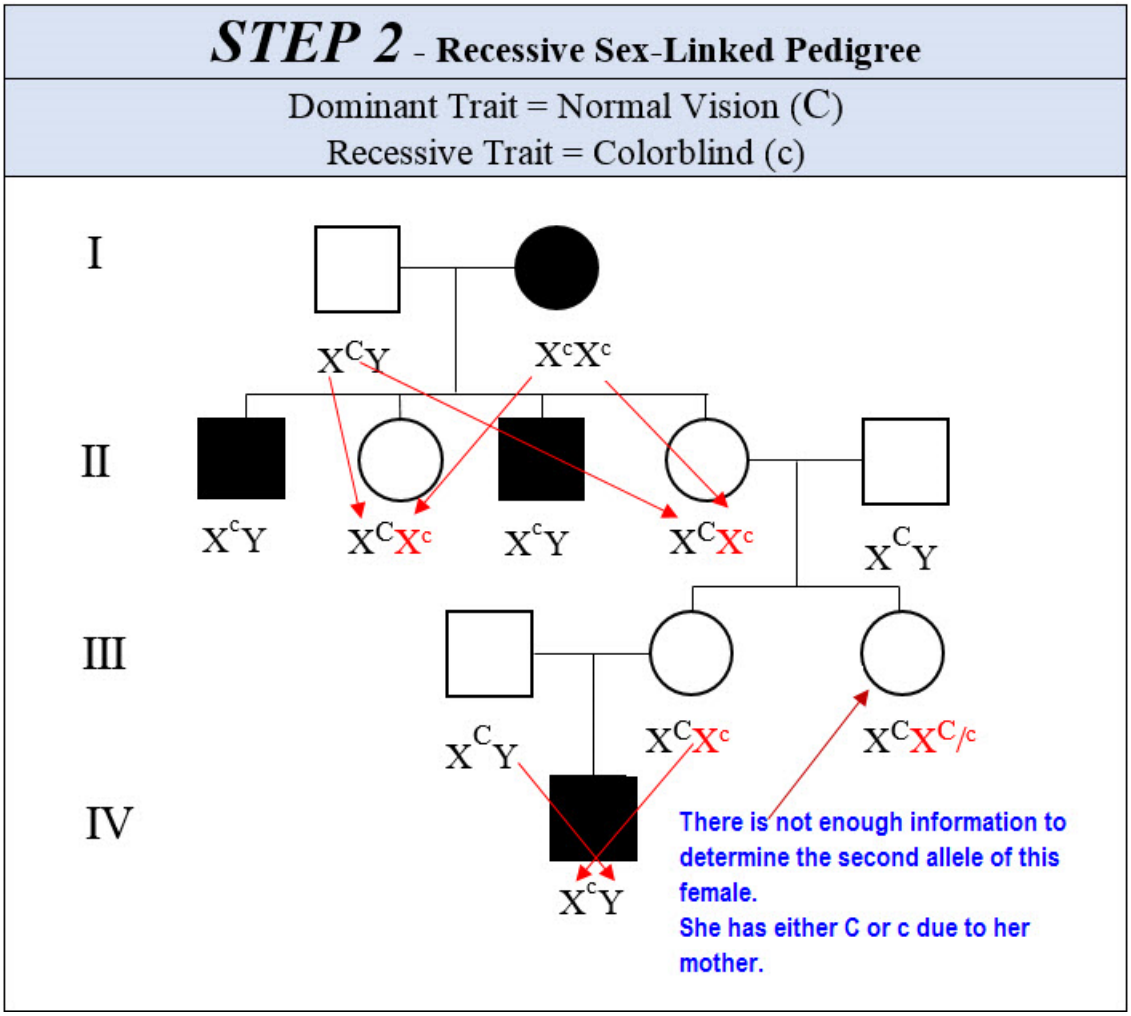
Now, we can fill in the pedigree with the genotypes we know.



Step 2, we need to determine the second allele of every empty female circle. Remember that each individual needs two alleles, one from the mother and one from the father. Each male, in a sex-linked trait, always inherits the "Y" from his father. The "X" the male inherits always comes from his mother.

In females, one "X" comes from her father and one "X" comes from her mother. To determine the second allele we need to examine the offspring and parents together. Follow the arrows in the pedigree below to understand how the second allele is determined.

Again, it is very important to understand that the two alleles each individual has are the result of one allele from the mother and one allele from the father. Once you have determined that a parent gives one specific allele, you can no longer look to that parent to give another allele to the same child, **for example** the first female in generation II. We know she has an "X^C" for two reasons. One reason is because her symbol is empty and she has normal vision. The other reason is because, when looking at her father in generation I, the only "X" he can give her is "X^C". Looking at her mother, we see the only X see can give her daughters is "X^c".



Pedigree Analysis Summary

You should be able to determine and explain all individual genotypes and phenotypes of any pedigree by analyzing parents and offspring, and knowing what type of inheritance the pedigree uses. Remember, when analyzing a pedigree, to first identify what the symbols tell you directly as in who has the trait. Second, determine the second unknown allele by examining which parent gives which allele, remembering that each parent only gives one allele at a time to each offspring.

UNIT VOCABULARY REVIEW

Unit 26 Worksheet Pedigrees

Click on the **Quizlet icon** below to access the quizlet.com vocabulary flash cards. Review the vocabulary before completing your assessment.

Quizlet

Now answer questions 1 through 20.