

Name _____ Instructor _____ Lab Section _____

Objectives: To gain an understanding of:

- The statistical analysis of genetic crosses
- The relationship between phenotype and genotype
- The genetic diversity of Biology 100 students
- The analysis of human pedigrees

Background material may be found in

- Chapter 9.1 - 9.9, 9.11 - 9.15, 9.20 - 9.22
- Chapter 11.2

Biology: Concepts & Connections, 8th ed.

Genetics is the study of the way in which different characteristics of an organism (e.g. finger shape, eye color, height, the ability to make a certain digestive enzyme) are determined and passed on from parent to offspring.

The basic unit of heredity is the **gene**. A gene is a **locus** on a **chromosome** containing a portion of **DNA** that codes for a specific **protein**. Each gene is able to govern a particular feature of an organism by determining the type of protein produced. This protein can function as a structural protein, enzyme, or hormone (e.g. insulin) affecting that particular feature. Typically each gene has several different forms (called **alleles**) that result in the variation of a feature one sees in different individuals. For example, one form (allele) of the gene for finger shape (B) may code for bent little fingers, whereas another form of that same gene (b) may code for straight fingers. The difference between the alleles is in the type of protein produced by each, which is due to a difference in the base sequence of DNA making up each allele.

The genetic make-up of an individual for a particular trait is referred to as the **genotype**. Since organisms are typically **diploid** (i.e. with chromosomes occurring in **homologous pairs**), there are a pair of genes governing each trait. For example, the genotype for finger shape may be Bb which actually represents the pair of alleles on a homologous pair of chromosomes.

The actual expression of these alleles is called the **phenotype**. The phenotype of a trait is always the product of the interaction between the individual's genetic make-up for that trait and the individual's environment.

The purpose of today's lab is to consider some basic terminology and concepts of genetics and to determine the genotype of members of the class by looking at your phenotypes for 20 different traits.

IT IS HIGHLY RECOMMENDED THAT YOU USE A PENCIL TO COMPLETE THIS LAB.

FORMATION OF GAMETES

In preparation for the genetics problems below, write the possible **gametes** (sex cells) that can be formed from the following genotypes. ✂ Place a circle around each gamete. Remember that gametes are haploid and that each gamete must contain one of each kind of gene found in the genotype of the diploid parent forming the gamete.

<u>GENOTYPE</u>	<u>POSSIBLE TYPES OF GAMETES</u>
AA	_____
Aa	_____
AABb	_____
AaBb	_____
AaBbCc	_____

GENETICS PROBLEMS-

 Using the information presented, try to solve the following problems.

1. In peas, the allele (T) for tallness is dominant over the allele for short stems (t). If a plant which is homozygous for tallness is crossed with one homozygous for short stems:

a. What are the genotypes of the parents?

b. What are the possible types of gametes they can produce? (Draw a circle around each gamete.)

c. What is/are the possible genotype(s) of the offspring? What is/are the possible phenotype(s)?

Genotypes: _____

Phenotypes: _____

2. In the following matings, write the gametes that can be produced by each parent, and draw a circle around each gamete. Then write the possible genotype(s) of their offspring. Use a Punnett Square to organize the information in part b.

a. TT X Tt

parent 1

parent 2

gametes: _____ offspring genotypes: _____

b Tt X Tt

gametes: _____ offspring genotypes: _____

3. In cattle the allele (A) that produces the polled (hornless) condition is dominant over the allele (a) for the horned condition. A certain polled bull is bred to three cows. With Cow A, which is horned, a polled calf is produced; with Cow B, also horned, a horned calf is produced; and with Cow C, which is polled, a horned calf is produced. Give the Genotypes of each of the parents and the calves produced by each cow.

Bull _____

Cow A _____ calf: _____

Cow B _____ calf: _____

Cow C _____ calf: _____

HUMAN GENETICS

In this part of the lab you will look at 23 different traits in each of yourselves. By observing the phenotype of each trait, you will then try to determine your genotype for each feature. We have selected traits which are easy to determine. Many of them, such as eye color, are governed by more than one pair of alleles. Others are also affected by different exposure in the environment, as in the case of freckles. For the sake of simplicity, we will ignore these shades of difference in the expression of these traits and simply lump dark colored eyes into one category and light colored eyes into another. Record your results in the chart which follows below. Also record these data on the summary chart for the entire section up at the front of the lab. There are photographs and illustrations at the front of the room and on your lab table to assist you.

TRAITS:

PTC Tasting - The allele (T) for tasting PTC is dominant over the non-tasting allele (t).

Sodium Benzoate Tasting - The taster allele (S) for sodium benzoate is dominant over the non-taster allele (s).

Shape of Little Finger - Hold your hands before your face with the palms toward you. Place the little fingers side by side and press them together. Do they run parallel their entire lengths or do the terminal digits flare out away from each other? Each bone in a finger is termed a phalanx. The allele for bent phalanx (B) is dominant over straight phalanx (b).

Mid-Digital Hair - Hair on the back of the middle digits of the fingers (H) is dominant over hairless middle digits (h).

Hitchhiker's Thumb - Hold your hand in the hitchhiker's position and look at the angle that is formed. If the thumb forms an angle with the fist of greater than 50 degrees it is due to the recessive allele (ht). If the thumb cannot hyperextend, it is due to the presence of the dominant allele (HT).

Palmar Muscle - The long palmar muscle lies along the inner aspect of the forearm with a tendon extending through the wrist to the palm of the hand. Count the tendons in the wrist on the palmar side. If you have three tendons the muscle is present; if you can find only two tendons, the muscle is absent. The allele for the absence of the palmar muscle (P) is incompletely dominant over the allele for its presence (P'). If it is found in one arm and not the other, the person is heterozygous (PP') for the trait.

Tongue Rolling - The ability to roll the tongue into a longitudinal U-shaped trough (R) is dominant over the lack of this ability (r).

Tongue folding - Tongue folding (F) is dominant over the lack of the ability (f). A tongue folder must be able to hold the tongue out and without bracing it against the teeth, fold the tip back sharply.

Dimples - The allele for dimples (D) is dominant over the allele for no dimples (d).

Freckles - The allele for some to many freckles (FR) is dominant over the one for the absence of freckles (fr).

Ear Lobe Attachment - Free ear lobe (E) is dominant over attached ear lobe (e).

Darwin's Ear Point - Darwin's ear point (EP) is dominant over no ear point (ep). Check both ears. The size of the ear point may vary.

Eye Color - Assume an allele for various shades of brown eyes (BR) is dominant over the allele for blue or blue-gray eyes (br). (Note: this is an over simplification. There are over 40 shades of eye color, and undoubtedly several pairs of alleles are involved.)

Hair Color, Dark/Light - Assume the allele for dark hair (HC) is dominant over light hair (hc), and that a person heterozygous, HChc, will have dark hair. As in the case with eye color, this is an oversimplification, for there are many shades of hair color. In addition, sunlight and chemicals can alter the expression of the genes for hair color.

Hair Color, Red/Non-red - The color of red is inherited independently of dark/light hair coloration. Assume that the gene for red (rd) is recessive to the gene for non-red (RD), and that a person who is heterozygous (RDrd) will have non-red hair.

Hair Form - Assume the allele for curly hair (U) is not completely dominant over the allele for straight hair (U'). The heterozygous condition (UU') produces wavy hair.

Hair Whorl Pattern - When viewed from above, the whorl pattern of the hair which is clockwise is due to the dominant allele (W). The counterclockwise pattern is due to the allele (w) in the homozygous condition.

Hairline - Examine the hairline on your forehead. The allele for a widow's peak (V) is dominant over the allele for a straight or curved hairline (v).

Cephalic Index - This trait is governed by several pairs of alleles and may be influenced by environmental conditions during development. (See the chart on your desk.) The adaptive value of a particular head shape is not known. Measure the width of the head at its broadest point above the ear with a caliper. Hold the caliper carefully when transferring it from the head to the meter stick so that the measurement will be accurate. Measure the length, placing one point of the caliper midway between the eyebrows and the other point at the point on the back of the head that will give the greatest length. Then divide the width by the length. The cephalic indices are classified:

.75 or less	→	narrow head
.76 - .79	→	median head
.80 or more	→	broad head

INDIVIDUAL SUMMARY CHART – PHENOTYPE AND GENOTYPE OF 23 TRAITS

In the chart below, record your phenotype for each trait (for example, taster or nontaster, straight or bent little finger), and circle your genotype.

<u>TRAIT</u>	<u>YOUR PHENOTYPE:</u>	<u>CIRCLE YOUR GENOTYPE:</u>
PTC Tasting	_____	T- tt
Sodium Benzoate Tasting	_____	S- ss
Bent Little Finger	_____	B- bb
Mid-Digital Hair	_____	H- hh
Hitchhiker's Thumb	_____	HT- htht
Palmar Muscle	_____	PP PP' P'P'
Tongue Rolling	_____	R- rr
Tongue Folding	_____	F- ff
Dimples	_____	D- dd
Freckles	_____	FR- frfr
Ear Lobe Attachment	_____	E- ee
Darwin's Ear Point	_____	EP- epep
Eye Color	_____	BR- brbr
Hair Color, Dark/Light	_____	HC- hchc
Hair Color, Red/Non-red	_____	RD- rdrd

Hair Form	_____	UU UU' U'U'
Hair Whorl Pattern	_____	W- ww
Hairline	_____	V- vv
Cephalic Index	_____	NARROW MEDIUM BROAD

QUESTIONS

1. Because an allele is dominant, does this necessarily mean that it will occur at a high frequency in a group?

What does the term "dominant" mean?

2. By knowing the phenotype for a feature (e.g. eye color) can we always determine the genotype exactly? Explain.

Asparagus Trait (Excretion of methyl mercaptan)

Asparagus contains the organic sulfur compound dimethylthetin [HO-S-(CH₃)₂-CH₂-COOH]. About 50% of the American population possess an enzyme which catalyses the conversion of dimethylthetin to methyl mercaptan (CH₃SH). It is this latter substance which gives urine its characteristic odor after asparagus is eaten. The presence of the enzyme is a dominant trait. Many people display this trait later in life - so, if you are negative now, things may change and you may begin to display the trait ("smelly" urine) later in life.

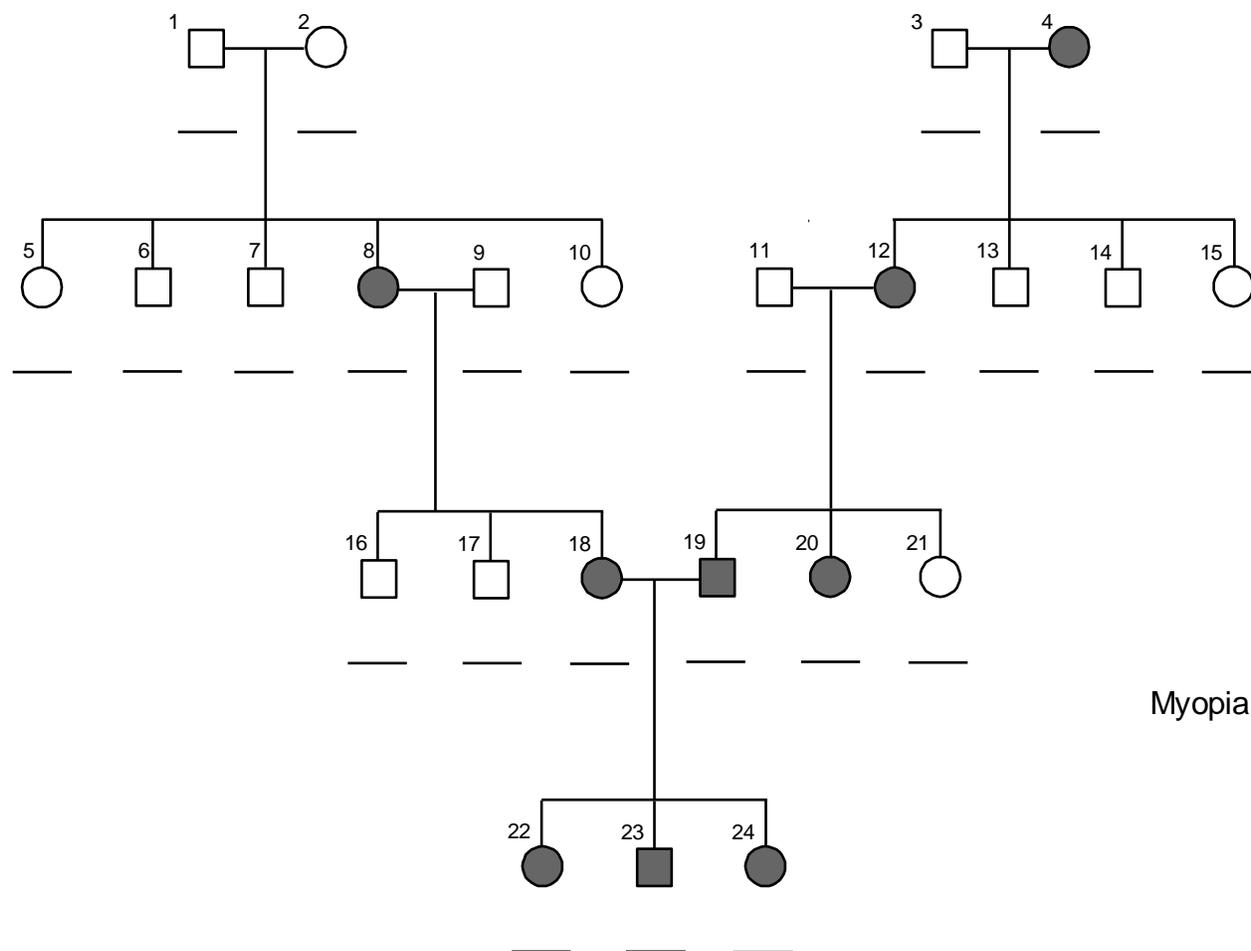
PEDIGREES

A **pedigree** is a representation of the genetic relationship among a group of individuals. Pedigrees can be used for a number of different purposes in the study of genetics including such things as determining the genotype of an individual in the group, predicting the chances of birth defects caused by particular alleles, and determining whether a trait is controlled by a recessive or dominant allele.

MYOPIA (NEARSIGHTEDNESS)

This is a pedigree for myopia (nearsightedness) which we shall assume is transmitted by a single pair of genes. In this and the other pedigrees which follow, those **individuals exhibiting the trait are indicated with shaded square or circle symbols**. Males are indicated by squares and females are indicated by circles.

 Go through the pedigree and **write the genotype for each individual on the line beneath it**. Use **A** and **a** for dominant and recessive. If the individual is dominant for a trait and you cannot determine whether or not (s)he is homozygous dominant or heterozygous, indicate the genotype as **A -**.



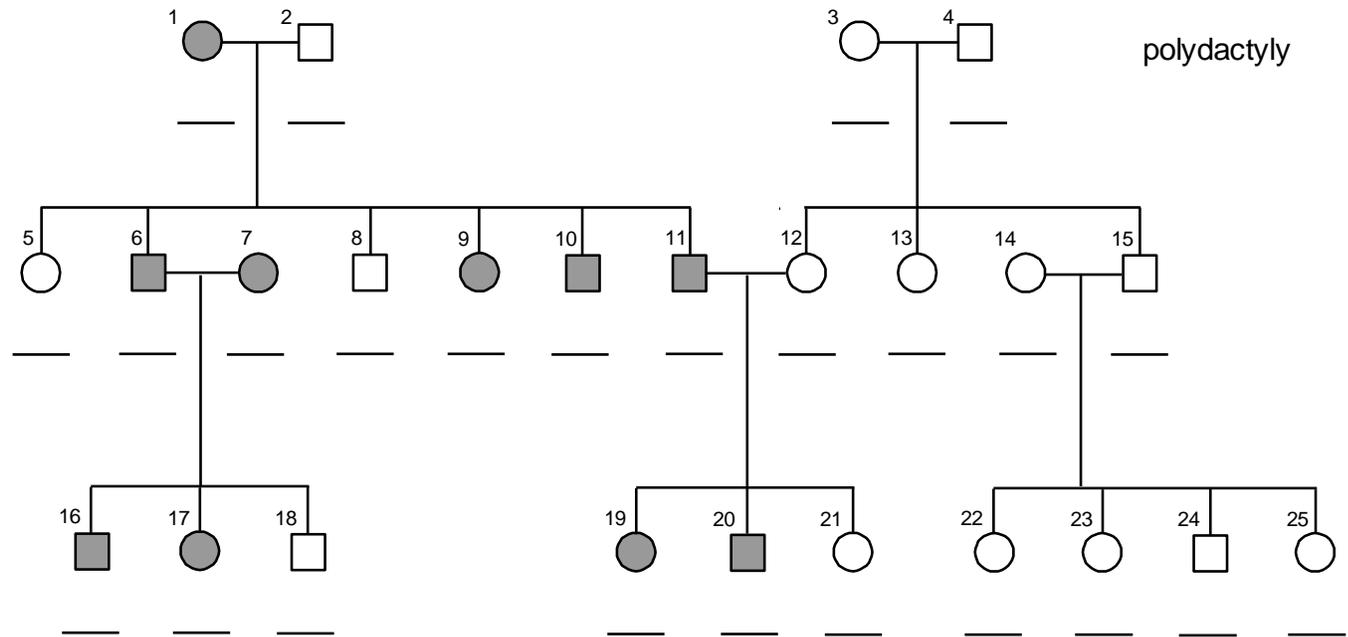
In this pedigree there are certain crosses which indicate whether the gene for the trait myopia is dominant or recessive. If the gene is dominant, any child showing the trait must have at least one parent showing the trait. (We neglect the possibility of a mutation.) If the trait appears in an offspring but not in either of its parents then it must be due to a recessive allele.

Notice that individuals 8, 12, 18, 19, 20, 22, 23, and 24 have the trait. In every case except number 8, each has one parent with the trait. Therefore they do not reveal whether the trait is dominant or recessive. Individual number 8 shows us that the trait is actually recessive because, though she has the trait, neither of her parents has it.

Now that we know the trait myopia is recessive, we can determine whether it is due to a gene on the X chromosome (sex-linked) or due to a gene on an autosome (not sex-linked). Females have two X chromosomes while males have only one. If the gene is on the X chromosome, males with the trait will always show it, while females may be heterozygous and not show the trait they carry. In order to show a sex-linked recessive trait, females must have the gene on both X chromosomes. In the pedigree above, we know that the trait is recessive. Now note that individuals 8, 12, and 18 are females showing the trait. **If the trait were sex-linked, then their fathers should show the trait. Why?** Since they don't, we know the trait is not sex-linked.

POLYDACTYLY (EXTRA FINGER ON EACH HAND)

Examine the following pedigree. **Give the genotypes of each of the individuals in the pedigree.** (Use **A** and **a** for dominant and recessive. For individuals who might be either **AA** or **Aa**, use **A-**). Answer the questions below.

**QUESTION**

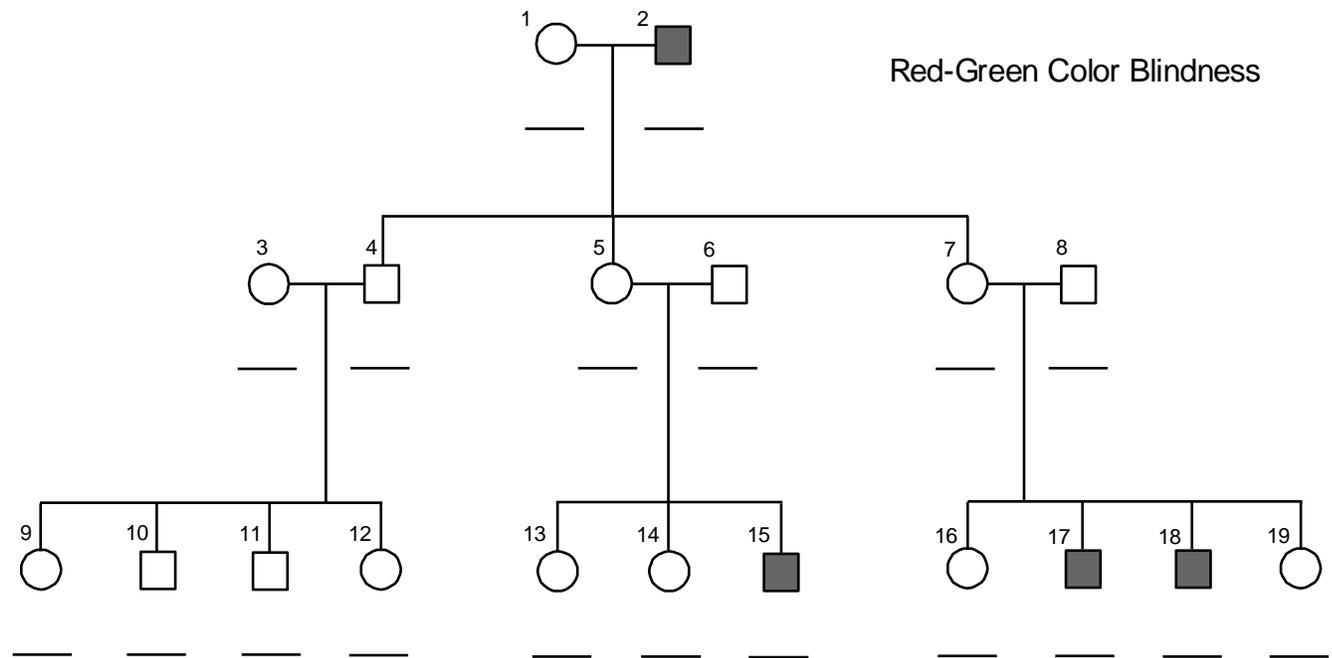
1. Is the trait for polydactyly dominant or recessive? Notice that the trait occurs only in the direct descendants of one of the parents and that it appears every generation.

2. Notice further that both numbers 6 and 7 have the trait, but their son, number 18 lacks the trait. If the trait were recessive, then individuals 6 and 7 would have to be homozygous recessive, and they would not be able to have a normal child. Since they do have a normal child, the trait for polydactyly must be dominant. Both 6 and 7 must be heterozygous and carry the gene for normal fingers.

3. Is the gene for polydactyly located on the X chromosome? Notice that individual 11 has a daughter lacking the trait. Males have only one X chromosome and daughters, having two X chromosomes, must carry that one from the father as well as one of the two X chromosomes of the mother. Therefore, if the dominant gene for polydactyly were on the father's X chromosome, then daughter number 21 would have to have the trait. Since she does not, we know that the trait is an autosomal dominant and is not located on the X chromosome.

RED-GREEN COLOR BLINDNESS

 Write the genotype for each individual on the line beneath its symbol. Answer the following questions using the information concerning color blindness in the pedigree below.



QUESTIONS

- Is the trait for red-green color blindness dominant or recessive?
- Which of the following individuals indicates that the trait is dominant or recessive? Circle one.
 - 2
 - 4
 - 15

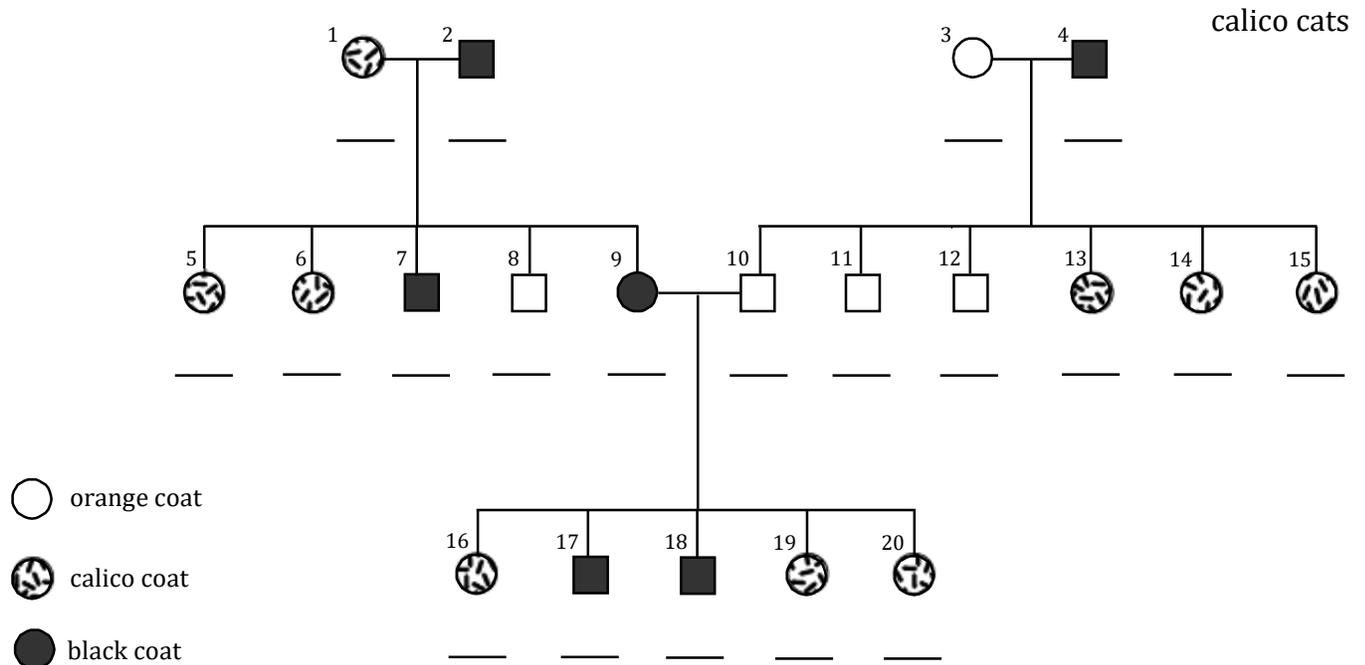
Explain why.

- Is the trait for red-green color blindness sex-linked?

4. Notice that individuals 9, 10, 11, and 12 do not show the trait which grandfather (number 2) has. Is this just due to chance, or is there a reason why they should not have grandfather's trait? Explain.

THE CASE OF THE TORTOISE-SHELL (CALICO) CATS

The following pedigree is of the inheritance of black, orange, and tortoise-shell coat patterns (i.e. black and orange on a background of white) in the domestic cats (*Felis catus*). **Indicate the genotypes of each of the individuals in the pedigree. Use O for orange and B for black. Use Y for the Y chromosome.**



First let us assume that this trait is due to one pair of genes and that it is a case of **co-dominance** in which the heterozygous individuals have calico (tortoise-shell) coat colors. In the development of the embryo, a certain cell which will grow into a large area of skin apparently becomes modified so that the hair which grows from it will only express the allele for orange. Another cell which will grow into an adjacent area of skin apparently becomes modified so that its offspring will only express the allele for black hair. **Are these alleles located on autosomes or on sex chromosomes?** In order to find out, complete the following section.

QUESTIONS

1. How many female calico cats are there in this pedigree?

How many males?

2. Let us assume that orange coat is homozygous, calico coat is heterozygous and black coat is homozygous. Notice that individual 3 would be homozygous for orange and individual 4 would be homozygous for black. If this were the case, then all of their offspring should be calico. However, they are not. The females are heterozygous, but the males have the orange coat color of the mother. There is no expression of the father's black coat color in his sons. (In the determination of sex in mammals, the male has only one X chromosome in his cells, and he received that from his mother. From his father he received the Y chromosome, which does not contain any genes for body traits.) Therefore, we can explain this pedigree by assuming that the genes for orange and black coat color in cats are located on the X chromosome. The first step in understanding this is to work out the genotypes of each individual.

3. Suppose our explanation is acceptable; however a case arises in which an individual like number 5 above is a male, not a female. What rare combination of chromosomes could allow this to happen?

What do we call humans with this phenomenon?

QUESTIONS

1. If a man with the dominant characteristic for tasting PTC marries a woman who is a nontaster and they have a child who is a nontaster, what is the genotype of each parent?

father _____ mother _____

2a. This question considers a lethal, x-linked recessive, allele that results in the death of the embryo prior to birth. If a woman has this lethal recessive allele on one X chromosome, and she marries a normal male, what is the probability that the first child she gives birth to will be a son?

2b. If they give birth to a daughter, what is the probability that she will be a carrier of the lethal allele?

Clean-up:

_____ **Return all supplies to side counter.**

LABORATORY NOTES
