Student Name ______________________________

Date ___________________ Group ____________

Laboratory 1 - Sex-Linked Inheritance and Human Genetics

Pre-Lab Report

Write a one- or two-sentence summary of the purpose, methods, and most importantly, expected results, for each investigation.

INVESTIGATION 1:

PURPOSE:

MATERIALS & METHODS:

EXPECTED RESULTS:

INVESTIGATION 2:

PURPOSE:

MATERIALS & METHODS:

EXPECTED RESULTS:

PRE-LAB REPORT

INVESTIGATION 3:

PURPOSE:

MATERIALS & METHODS:

EXPECTED RESULTS:

INVESTIGATION 4:

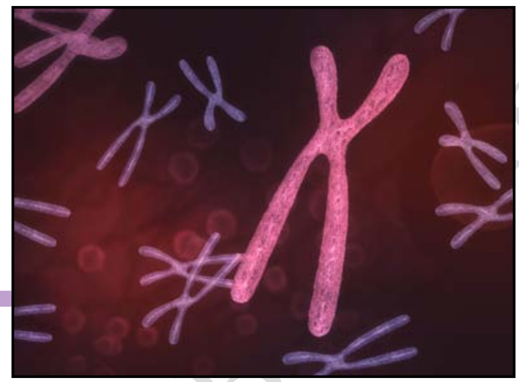
PURPOSE:

MATERIALS & METHODS:

EXPECTED RESULTS:

LABORATORY 1

Sex-Linked Inheritance and Human Genetics



SCENARIO: THE ROYAL DISEASE

Hemophilia, a sex-linked disease, has played an important role in Europe's history, for it suddenly cropped up in the children of Great Britain's Queen Victoria. It became known as the "Royal disease" because it spread to the royal families of Europe through Victoria's descendants. (See Figure 1-1.) Queen Victoria had been worried about the quality of the blood of the British royal family. Her evaluation was "that constant fair hair and blue eyes makes the blood so lymphatic."

It is doubtful that the Queen knew exactly what was wrong with her family's blood. Hemophilia first appeared in Victoria's family in her eighth child, Prince Leopold, Duke of Albany. Throughout his short life, Leopold suffered severe hemorrhages, and always was described as "very delicate." Leading the life of a normal youngster was impossible for Leopold because any cut or bump could lead to death, and it had been necessary to keep him always under strict surveillance. However, in spite of all protection, Prince Leopold died at the age of 31, as a result of a minor fall.

Leopold did marry and have children, however, and his daughter also had a hemophiliac son. Even more significant was the fact that two of Victoria's daughters carried the gene into the royal families of Spain, Prussia, and Russia. Victoria's granddaughter, Alexandra (Alix), was also a carrier. Had she accepted the offer of marriage from Prince Eddy, or his brother George, hemophilia would have been reintroduced into the reigning branch of the British royal family. But Alix married Tsar Nikolas II instead, and carried the disease into the Russian imperial family. She had four daughters before giving birth to the long-awaited son, Alexis, heir to the Russian throne.

Within a few months of his birth, Aexis's parents realized that their precious and only son had hemophilia. The first sign had been some unexpected bleeding from the naval, which had stopped after a few days. Much more serious, however, were the dark swellings that appeared each time the child bumped an arm or a leg. And worst of all was the bleeding into the joints. This meant, as well as excruciating pain, a crippling of the affected limbs. As the boy grew older, he was obliged to spend weeks in bed, and after he was up, to wear a heavy iron brace.

Neither well-experienced doctors nor numerous prayers to God by desperate parents seemed to help the suffering child. Distressed over their son's condition, the parents, the Tsar and Tsarina, turned to the monk, Rasputin, a spiritualist who claimed he could help Alexis. Rasputin received an unlimited trust from Alexandra because he was the only person who was able to relieve her son's sufferings. How he managed to do this is uncertain. A likely explanation is that Rasputin, with his hypnotic eyes and his self-confident presence, was able to create the aura of tranquillity necessary to slow the flow of blood through the boys veins. Where the demented mother and the dithering doctors merely increased the tenseness of the atmosphere around the suffering child, Rasputin calmed him and sent him to sleep. While Tsar and Tsarina were preoccupied with the health of their son, the affairs of state deteriorated, culminating in the Russian revolution. Alexis did not die from hemophilia. At the age of 14 he was executed with the rest of the family.

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DISCUSSION:

1. What is the genetic defect involved in hemophilia?
2. How did Victoria inherit the gene?
3. Why did Victoria's daughters not realize they carried the gene?
4. Why is the current royal family of England not affected by the disease?
5. What are the probabilities that Alexis's sisters were carriers of the hemophilia allele?
6. Suppose Alexis had lived and married a normal woman.
    a. What are the chances that his daughter would be hemophilic?
    b. What are the chances his daughters would be carriers?
    c. What are the chances that his sons would be hemophiliacs?

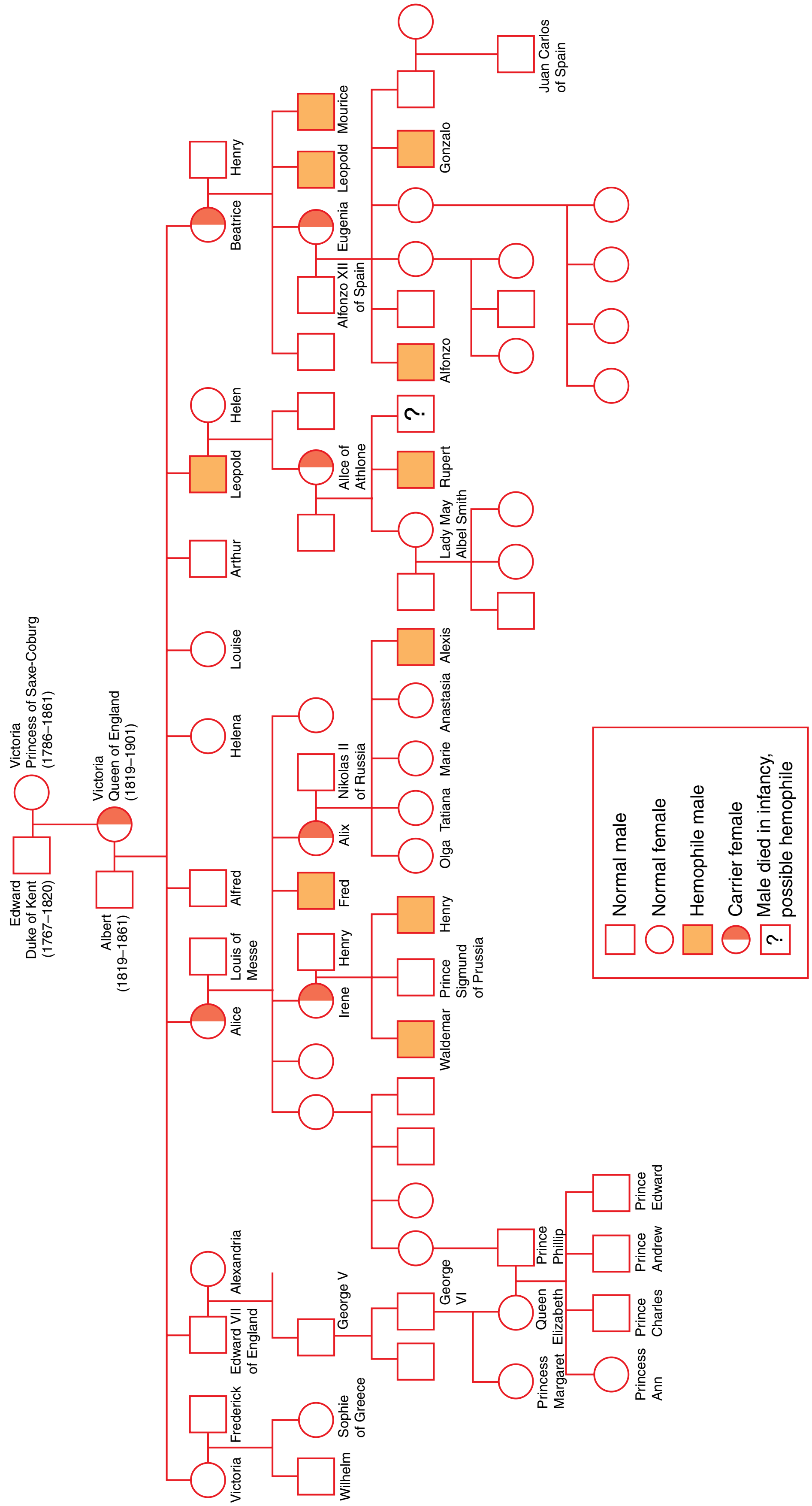

Figure 1-1. Royal family pedigree.

Laboratory Investigations

OBJECTIVES:

From this experience, you should be able to

- observe the principles of inheritance in *Zea mays* by analyzing a genetic cross
- describe how sex-linked traits are inherited
- recognize common dominant and recessive human traits
- demonstrate how human traits are inherited
- analyze a human pedigree

INTRODUCTION:

Genes that are located on the same chromosome are usually inherited together and are referred to as being linked. In contrast, those that reside on different chromosomes are inherited separately, and thus follow the Mendel's law of **independent assortment.** One can demonstrate a predictable pattern of the inheritance of two traits by performing a **dihybrid** cross (i.e., parents are heterozygous for two traits), and can determine phenotypic probabilities by a Punnett square.

The inheritance of genes located on sex chromosomes results in unique patterns according to the gender of the offspring. Since males and females differ in their sex chromosomes, inheritance patterns for genes located on the X-chromosome vary between the sexes. Although these patterns were first identified in fruit flies, the same genetic principles apply to humans. If the offspring is male (XY), that offspring will be essentially haploid for the genes located on the X chromosome sites. If he inherits a recessive allele for a gene on the X chromosome, it will be expressed, because there is no other allele to "counteract" it. The absence of an alternative allele on the Y chromosome leads to a phenotype ratio that can be predicted using Punnett squares. If the offspring is female (XX), she could be heterozygous, and the dominant trait would be expressed. For example, the inability to distinguish between the colors red and green (color-blindness) is an X-linked trait in humans. It is expressed in a male if he received the recessive gene from his mother, but can only be expressed in a female whose father is color-blind and whose mother passes on the recessive gene.

As in other living things, human traits are inherited according to the laws of Mendelian genetics. Many traits involve the interaction of several genes, which results in the variation we see even among brothers and sisters. However, some traits are controlled by single genes that have noticeable phenotypic effects.

INVESTIGATION 1: INHERITANCE OF TWO TRAITS IN ZEA MAYS

OBJECTIVE:

To observe and score the results of genetic crosses in corn which involve two traits, and determine whether a trait is dominant or recessive.

MATERIALS:

- 3 generations of *Zea* corn, exhibiting traits for kernel color ("R" or "r") and texture ("Su" or "su")

PREDICT:

For the two traits you will be scoring, predict which of the two alleles is dominant:
Kernel color: Purple or Yellow?
Kernel texture: Smooth or Wrinkled?

PROTOCOL:

1. You will examine 3 generations of *Zea*. The P_1 (parent generation) and F_1 (first offspring) will be shown to the class by the instructor. The F_2 (resulting from crossbreeding two members of the F_1 generation) will be given to the class for scoring. Each kernel represents a genetically unique individual.

2. Recognition of *Zea* traits.
    1. Kernel Color: An outer layer of the kernel may be pigmented or unpigmented. In this cross, if the layer is pigmented, it is purple. If the layer is unpigmented, the inner portion shows through, and the kernel is yellow. <u>Mottled kernels are scored as purple.</u>

    2. Kernel Texture: How rapidly a corn kernel dries is determined genetically. If water loss is slow, the kernel texture is even or smooth. If water loss is rapid, the kernel texture is wrinkled. Kernels must be uniformly wrinkled to be scored as wrinkled (one indentation does not qualify).

3. Work in pairs. One member will call off the phenotypes of the kernels; the other will record the numbers. Determine the categories of phenotypes before you begin to tally the number in each category.

4. To keep track of the count, put a beginning marker pin (colored pin) in the wide end of the cob at the end of a row of kernels. Leave the beginning marker in place. From the beginning marker pin count all the kernels that are both yellow and wrinkled. Count for 2 minutes, or until you've counted all the yellow-wrinkled kernels on the corn cob. If you did not count the entire cob, mark how far you progressed by inserting a T-pin just after the last kernel you counted. For the remaining 3 phenotypes, you will count the same amount of corn as you did for yellow-wrinkled, either the entire cob, or all the kernels between your beginning pin and your T-pin. Write your counts in Table 1-1.

5. Repeat your counting for the three remaining kernel phenotypes.

6. Determine the relative percentages for each phenotype, and enter these percents in Table 1-1.

7. Your instructor will put a table on the board that contains composite data from the class for each phenotype. Record these results in Table 1-1.

8. Complete the Punnett square (Figure 1-2) for a dihybrid cross on the response sheet.

QUESTIONS:

1. Which alleles were dominant and which recessive?
2. How many different phenotypes did you see?
3. How many genotypes result from a dihybrid cross?

SHARE:

Compare your results with the total results from the class and with the expected percentages from the Punnett square.

MAKE CONNECTIONS:

How does inheritance of two traits (dihybrid) differ from the monohybrid cross in Lab 7?

INVESTIGATION 2: SEX-LINKED INHERITANCE

OBJECTIVE:

To investigate the patterns of inheritance for sex-linked traits.

MATERIALS:

http://biology.clc.uc.edu/courses/bio105/geneprob.htm

PREDICT:

What is the likelihood that a color-blind man married to a woman who does not carry the recessive gene will have a color-blind son?

PROTOCOL:

1. Go to the section of the website that contains the exercise for sex-linked genes (approximately halfway down the page).
2. Work through the example, and check your answers using the "Am I right?" buttons.

QUESTIONS:

1. What percentage of sons born during her first marriage will be color-blind?
2. What percentage of sons born during her second marriage will be color-blind?
3. What percentage of daughters from her second marriage will be color-blind?

SHARE:

Check with your instructor regarding the results of your exercise. Using the white boards available in class, share the results of your investigation with your classmates.

REVIEW:

For a review of sex-linked inheritance, visit
http://biology.clc.uc.edu/courses/bio105/sex-link.htm or
http://www.biology.arizona.edu/mendelian_genetics/problem_sets/sex_linked_inheritance/07t.html

MAKE CONNECTIONS:

1. How is color-blindness and the disease hemophilia related?

2. What kind of genetic counseling advice would you provide for a hemophiliac father, who learned his wife was about to give birth to a son?

INVESTIGATION 3: HUMAN GENETIC TRAITS

OBJECTIVE:

To illustrate the variety possible in the human genome, using only a dozen of the many thousands of human traits.

MATERIALS:

- a coin
- crayons
- paper

PREDICT:

You and your partner will be "parents" of a hypothetical baby, who will be a sibling of all the other babies in the class.

1. What percentage of the babies will be identical?

2. What percentage will be males?

3. What percentage will be blue-eyed blondes?

PROTOCOL:

1. You and your lab partner will produce a unique offspring by randomly determining the genotype and phenotype of twelve human traits, by flipping a coin.

2. Both parents are heterozygous for every trait, that is, they both possess a dominant and a recessive gene for every trait.

3. Since it is the father who determines the sex of a baby, have the "father" of your pair flip the coin to determine the sex of your baby, heads = Y chromosome and tails = X chromosome.

4. On the response sheet, record your names, the name of your baby, and its gender.

5. Determine your baby's additional traits (as listed on the data sheet) by having **each** parent flip the coin to determine whether the dominant or recessive gene will be passed on to the baby. Since parents are heterozygous for each trait, there is an equal chance for each of them to pass on either allele.

6. In flipping the coin for each parent, heads indicates the dominant allele is passed on, and tails indicates the recessive allele. Record the allele contributed by each parent and the resulting genotype and phenotype of the baby for each trait in Table 1-2 on the response sheet.

7. For traits that are polygenetic, such as hair and eye color, each parent must flip the coin for as many genes as are required for the trait (e.g., hair color is produced by three pairs of alleles, so the coin must be flipped 3 times by each parent to determine hair color).

8. After you have determined the phenotype of your baby, draw a picture to show how he/she will look.

QUESTIONS:

1. Did most of the offspring in the class have brown hair?

2. Can two black-haired individuals have a blonde child?

3. In order for each of the above traits to be inherited independently, they must reside on separate chromosomes (law of independent assortment). How many chromosomes are necessary for these traits to be inherited independently?

HYPOTHESIZE:

From the answers to your questions, generate an hypothesis regarding the probability for two blue-gray-eyed parents to have a brown-eyed child. Complete the Punnett square (Table 1-3) on your response sheet that will test the hypothesis listed above.

SHARE:

Display your child at the front of the class. Compare his or her appearance with that of his/her siblings.

MAKE CONNECTIONS:

1. Discuss how the variation of the babies might change if one parent were homozygous dominant and the other were homozygous recessive for every trait.

2. The Punnett square from the hypothesis on the response sheet most closely illustrates a
   a. Monohybrid cross
   b. Dihybrid cross

c. Trihybrid cross
d. Test cross

INVESTIGATION 4: DETERMINING YOUR GENETIC TRAITS

OBJECTIVE:

To determine which alleles of genetic traits you express.

MATERIALS:

- test for color-blindness
- PTC paper

PREDICT:

The ability to taste PTC is due to a dominant gene. Predict whether there will be more tasters than non-tasters in the class. Defend your prediction.

PROTOCOL:

For each of the following traits, determine your phenotype and possible genotypes and record in Table 1-4.

1. PTC Tasting. For some people, phenylthiocarbamide (PTC) has a distinctively bitter taste; for others it is tasteless. Test for your ability to taste it by touching a piece of PTC paper to your tongue, or by chewing the paper. (If you are uncertain about whether you can taste the compound, you are a non-taster. Tasters know immediately.)

2. Color-blindness. Test for your ability to distinguish colors by using the chart book available in class.

3. Ear lobes. Examine your and your partner's ear lobes and note whether they are free or attached. The free lobe is the dominant trait.

4. Tongue Rolling. Stick out your tongue at your partner and roll it into a 'U' shape. The ability to roll the tongue is controlled by a dominant gene.

5. Widow's Peak. Check your and your partner's forehead for hairline appearance. The tendency to extend down and form a distinct point is dominant.

6. Mid-digital hair. Individuals with the dominant trait have hair on the middle (second) joint of their fingers.

7. Bent little finger. With both hands flat and relaxed on the table, check your little finger. If you have the dominant gene, the last joint of the little finger will bend in slightly toward the fourth finger.

8. Interlocking fingers. When you interlace your fingers, if you cross your left thumb over your right, this is dominant over the allele for crossing your right thumb over your left.

9. Hitchhiker's thumb. Bend the distal (closest to the tip) joint of your thumb back as far as possible. A recessive gene determines the ability to bend the thumb back to at least a 60 degree angle or more.

10. Index and ring finger lengths. The gene for the index (second) finger being shorter than the ring (fourth) finger is influenced by the sex of the individual. It is dominant in males and recessive in females. Place your hand on a sheet of lined paper with the fingers together. Place the fourth finger so that it barely touches one of the lines. Look at the index finger to see if it touches the line.

QUESTION:

1. How many females in the class would you predict to be color-blind?

SHARE:

Record your findings on the data sheet. Give your results to the instructor, who will tabulate them for the class.

MAKE CONNECTIONS:

1. How can you tell your genotype for each of these traits?

2. Which phenotype has only one genotype, dominant or recessive?

DISCUSSION:

1. If a gene is carried on the same chromosome as another gene (such as hair and eye color), would the results of the baby's phenotype be the same? Why or why not?

2. Huntington's disease is caused by a dominant allele. Yet its incidence in the population is approximately one in 10,000 persons. How do you explain that a dominant allele could be expressed so rarely?

3. Imagine an egg that mistakenly contains no X chromosome.
   a. If it is fertilized by a sperm with an X chromosome, what would be the result?

   b. If it is fertilized by a sperm with a Y chromosome, what would be the result?

   c. Which, if either, would result in a live birth? Why?

4. Return to the scenario and Figure 1-1.
   a. How many sons did Queen Victoria have?
      i. What percentage would have been expected to have hemophilia?

      ii. How many actually did?

   b. How many daughters did Queen Victoria have?
      i. What percentage would have been expected to be carriers of hemophilia?

      ii. How many actually did? Can you be certain?

Student Name ______________________________

Date __________________ Group ___________

Laboratory 1 - Sex-Linked Inheritance and Human Genetics

Response/Data Sheet

SCENARIO DISCUSSION QUESTIONS—SEX-LINKED INHERITANCE:

1.

2.

3.

4.

5.

6.

   a.

   b.

   c.

W O R K S H E E T

INVESTIGATION 1: INHERITANCE OF TWO TRAITS IN ZEA MAYS

PREDICT:

Table 1-1. Relative Numbers of Phenotypes.

Kernel Appearance	Tabulations	Total	Percent	Class Total	Class Percent
Purple/ Smooth					
Purple/ Wrinkled					
Yellow/Smooth					
Yellow/Wrinkled					
	Total Kernels Counted		100%		100%

Figure 1-2. Punnet square of a dihybrid cross between two F1 Zea mays individuals: SusuRr x SusuRr.

Put the genotype in the upper left of each cell and the phenotype in the lower right of each cell. Allele Abbreviations: Color: R = purple; r = yellow; Texture: Su = smooth; su = wrinkled.

Percentages of Phenotypes:

Purple/Smooth:
Purple/Wrinkled:
Yellow/Smooth:
Yellow/Wrinkled:

QUESTIONS:

1.

2.

3.

MAKE CONNECTIONS:

INVESTIGATION 2: SEX-LINKED INHERITANCE

PREDICT:

QUESTIONS:

1.

2.

3.

MAKE CONNECTIONS:

1.

2.

INVESTIGATION 3: HUMAN GENETIC TRAITS

PREDICT:

1.

2.

3.

See Table 1-2 on the following page for recording your baby's traits.

QUESTIONS:

1.

2.

3.

HYPOTHESIS:

Test your hypothesis by filling out the Punnett square below. The possible genetic contributions from the mother are filled in for you. Circle the alleles that represent a phenotypically brown-eyed child. What percentage of offspring have brown eyes?

Table 1-3. Offspring of Blue-gray-eyed Parents.

	Bc	bc	Bc	bc

MAKE CONNECTIONS:

1.

2.

Table 1-2. Inheritance of Human Traits.

Baby's Name ____Sex ______________

Inheritance Pattern				Alleles from:		Phenotype
	Trait	Dominant	Recessive	Mother	Father	
Simple Dominance	Widow's Peak	Present (WW, Ww)	Absent (ww)			
	Eyebrows	Bushy (BB, Bb)	Fine (bb)			
	Eyelashes	Long (LL, Ll)	Short (ll)			
	Lips	Thick (TT, Tt)	Thin (tt)			
	Dimples	Present (DD, Dd)	Absent (dd)			
	Freckles	Present (FF, Ff)	Absent (ff)			
	Face Shape	Rounded (RR, Rr)	Squarish (rr)			
Incomplete Dominance	Hair Type	Curly (CC) Wavy (Cc) Straight (cc)				
	Eye size	Large (LL) Medium (Ll) Small (ll)				
	Mouth Length	Long (LL) Medium (Ll) Short (ll)				
	Nose Size	Large (LL) Medium (Ll) Small (ll)				
Polygenic inheritance	Hair Color (3 genes)–Toss the coin three times each, once for each of the following allele pairs: XxYyZz	6 dom: black (XXYYZZ)	5 dom: dark brown			
		4 dom: brown	3 dom: light brown			
		2 dom: blonde	0 dom: almost white (xxyyzz)			
	Eye Color (2 genes)–Toss the coin twice, once for each of the following allele pairs BbCc	BBCC or BBCc: dark brown BbCC or BBcc: brown	bbCC: green BbCc: hazel			
		Bbcc: blue gray bbCc: dark blue	bbcc: light blue			

INVESTIGATION 4: DETERMINING YOUR GENETIC TRAITS

PREDICT:

Table 1-4.

Trait		Alleles	Your Phenotype	Possible Genotypes	Number in Class	
Dominant	Recessive				Dom	Rec
PTC taster	Non taster	Tt				
See Color	Colorblind	Cc				
Free ear lobes	Attached	Ff				
Tongue roller	Non Roller	Rr				
Widow's peak	No Widow's peak	Ww				
Mid-digital hair	No mid digital hair	Hh				
Bent little finger	Straight little finger	Bb				
Left top interlocking	Right top interlocking	$T_I t_I$				
No hitch-hiker thumb	Hitchhiker's thumb	Tt				
Index vs ring finger shorter / longer	Index vs ring finger longer / shorter	Ii /Rr				

QUESTION:

MAKE CONNECTIONS:

1.

2.

DISCUSSION:

1.

2.

3.

4a. i.

ii.

4b. i.

ii.