

Human Inheritance and Pedigree Analysis

Background: With an understanding of heredity and probability, biologists have learned about the genetics of many human traits. In many of these traits, several pairs of genes are involved and the pattern of inheritance is complex. For this activity we will assume that the traits we are studying are regulated by the alleles of only one gene, with one allele coming from the father and the other allele from the mother, as described in Mendel's Law of Segregation.

Objectives: In this activity you will:

- Determine your phenotype for several traits.
- Determine (as far as possible) your genotype for the same traits.
- Interpret the genotypes of individuals in a pedigree.

Procedures and Observations:

--- Part One: Phenotype and Genotypes of Common Traits ---

You will determine your phenotype and try to determine your genotype for the traits listed in Table 1. Remember, if you show a dominant trait you may be homozygous or heterozygous for that trait. Suppose, however, that one of your parents shows the recessive trait. In that case, the parent would have passed on a gene for that recessive trait and you would be heterozygous. If neither of your parents shows the recessive trait, you may not know if you are heterozygous or homozygous for that trait. In that case, put a blank () for the unknown allele. If you show the recessive trait, record the genotype and the phenotype, with two recessive alleles.

1. Free earlobes, L , are dominant. People whose earlobes are attached directly to the head have the recessive genotype ll .

Have your partner check your earlobes.

- Record Your phenotype and genotype in Table 1. on the next page. (If you cannot determine whether your genotype is homozygous or heterozygous, record $L_$ in the table.)



Free



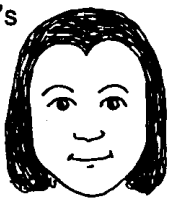
Attached

2. As we have discussed in class, multiple genes control inheritance of eye color but people having the homozygous recessive genotype bb have blue (or green) eyes. People who have the dominant allele, B , may have different shades of brown or hazel eyes.

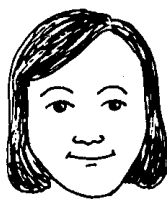
Check your eye color.

- Record your phenotype and genotype in Table 1.

Widow's
Peak



Peak



No Peak

3. A widow's peak is a hairline that forms a downward point in the middle of the forehead. This is caused by the dominant allele, W . A smooth hairline is caused by the recessive genotype ww .

Have your partner check your hairline.

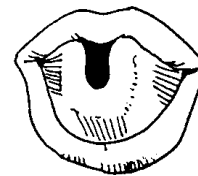
- Record your phenotype and genotype in Table 1

4. A dominant allele, R , gives some people the ability to roll their tongues into a “U” shape when it is extended. People with the recessive alleles, rr , cannot roll their tongues.

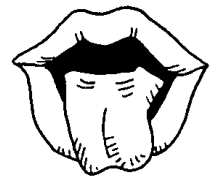
Check to see if you can roll your tongue.

→ Record your phenotype and genotype in Table 1.

Tongue
Rolling

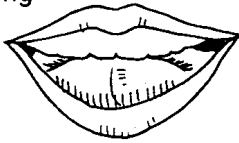


Rolled

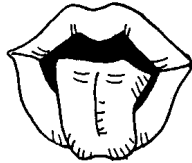


Not Rolled

Tongue
Folding



Folded



Not Folded

5. A dominant allele, T , gives some people the ability to fold their tongues over without using their teeth. People with the recessive genotype, tt , cannot.

Check to see if you can fold your tongue.

→ Record your phenotype and genotype in Table 1.

6. A dominant allele, F , results in the end joint of the little finger bending inward. Straight little fingers are a result of recessive genotype ff .

Place your hands on a flat surface, palms down, and relax. Check to see if the first joints on your little fingers are bent or straight.

→ Record your phenotype and genotype in Table 1.

7. People who can taste the bitter chemical phenylthiocarbamide, PTC, have at least one dominant allele, A . Those with the recessive genotype, aa , cannot taste it.

Taste the PTC paper to find out if it tastes bitter to you.

→ Record your phenotype and genotype in Table 1.

8. Individuals who have hair on the middle joints of their fingers have at least one dominant allele, H . Those with two recessive alleles, hh , do not have hair on that joint.

Check to see if you have hair on the middle joints of your fingers.

→ Record your phenotype and genotype in Table 1.

9. Individuals with red hair have the recessive genotype nn . Those with any other hair have at least one dominant allele, N .

Check your hair color.

→ Record your phenotype and genotype in Table 1.

10. Individuals with curly hair have at least one dominant allele, C . Those with straight or wavy hair have the recessive genotype cc .

Check your Hair form.

→ Record your phenotype and genotype in Table 1.

Hair
Form



Curly

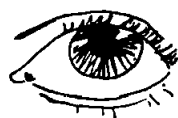


Straight

Eyelashes



Long



Short

11. Long eyelashes are the result of the dominant allele S . Short eyelashes are the result of the recessive genotype ss .

Check the length of your eyelashes.

→ Record your phenotype and genotype in Table 1.

Table 1. Phenotypes and Genotypes of Some Human Traits

Trait and Symbols for Genes	Phenotype	Genotype
Shape of ear lobe L,l		
Eye color B,b		
Shape of hairline W,w		
Ability to roll tongue R,r		
Ability to fold tongue T,t		
Shape of little finger F,f		
Ability to taste PTC A,a		
Hair on middle joint of finger H,h		
Hair color N,n		
Hair curliness C,c		
Eyelash length S,s		

--- Part Two: Human Pedigree Analysis ----

A diagram showing the transmission of a trait through several generations of a family is called a pedigree. In Figure 1, generation I is made up of grandparents, generation II is their children, and generation III is their children.

1. Study the pedigree diagram and the key in figure 1 to learn the symbols.

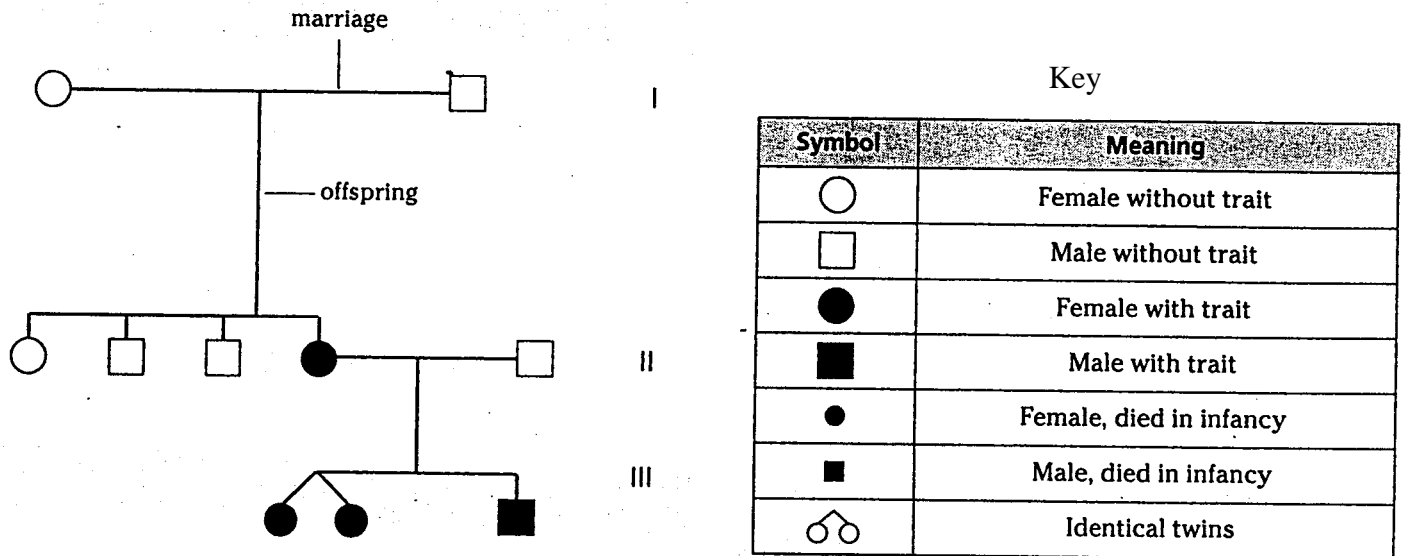


Figure 1

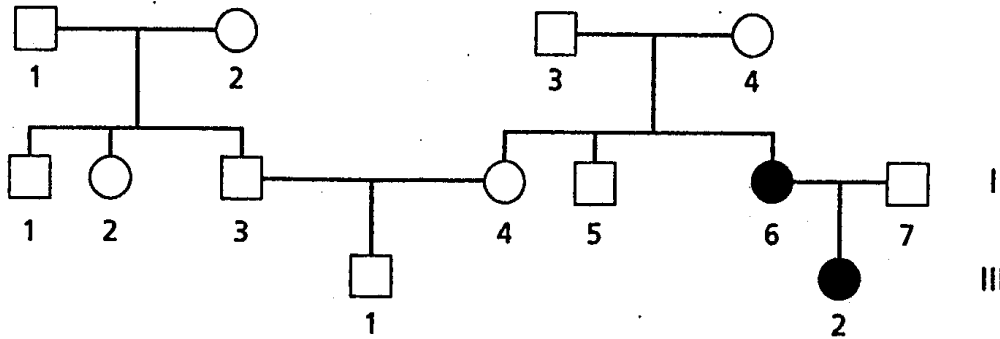
Case one: Albinism Individuals that lack an enzyme needed to form the skin pigment melanin are called albinos. Normal skin pigmentation is dominant.

2. Use D to represent the allele for normal skin and dd to represent the genotype for albinism. Where you cannot be sure whether an individual with the dominant trait is homozygous or heterozygous, show the genotype as $D_$.

→ What does the Law of Segregation say about where each individual gets each one of their alleles

→ Use the data table to list the genotype of each individual in figure 2 on the next page. Keep Mendel's laws in mind.

Albinism Pedigree



Individual	Genotype
I 1	
I 2	
I 3	
I 4	
II 1	
II 2	
II 3	
II 4	
II 5	
II 6	
II 7	
III 1	
III 2	

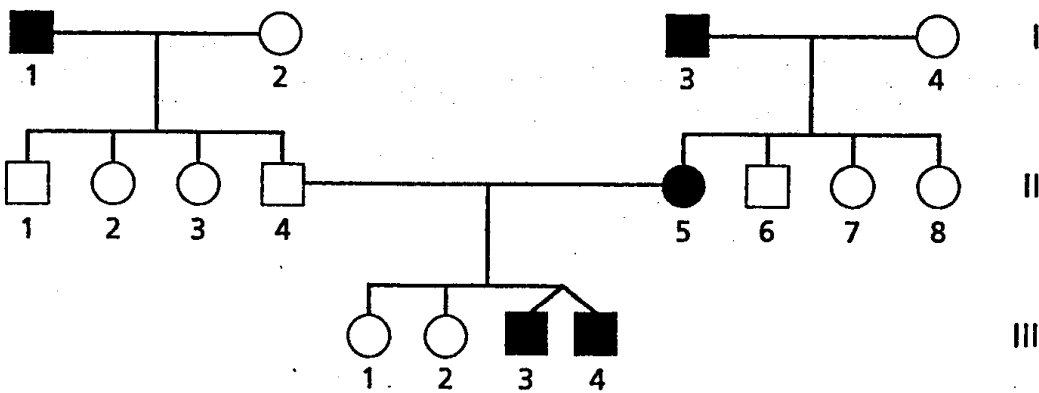
Figure 2

Case Two: PTC Non-Taster

We already discussed these individuals in Part One.

- Use A to represent the allele for the ability to taste PTC, a dominant allele. Use aa for the PTC non-taster, who exhibits the recessive trait. Use $A_$ where the genotype is uncertain. **Individuals that are filled in are the recessive PTC non-tasters**
- Use the data table to list the genotypes of each individual in figure 3 below. Remember to keep Mendel's laws in mind (especially the Law of Segregation).

PTC Taster Pedigree



Individual	Genotype
I 1	
I 2	
I 3	
I 4	
II 1	
II 2	
II 3	
II 4	
II 5	
II 6	
II 7	
II 8	
III 1	
III 2	
III 3	
III 4	

Figure 3

Yes, There ARE questions on the back!!!!!!

Analysis and Conclusions:

1. Do you think anyone in the class has the exact same traits that you have on your list from part one? Why/why not?
2. In the pedigree in Figure 2, if the individuals 6 and 7 have another child, what is the chance that it will be albino. Show the Punnet square.
3. In Figure 2, can you determine the genotypes of individuals 1 and 2 in generation I? Explain your answer.
4. In the pedigree in Figure 3, if individuals 4 and 5 have another child, what is the probability that it will be a taster?
5. In Figure 3, if individual 8 in generation II married a man with genotype AA, what is the probability that she will have a non-taster child? Illustrate your reasoning with a Punnet square.