LABORATORY #7 -- BIOL 111
Genetics and Inheritance

You have seen chromosomes in the onion root tip slides we used to examine the cell cycle. What we cannot see are the individual genes on these chromosomes. This lab is an introduction to how one analyzes genes without ever seeing or touching them. The first person to do this was **Gregor Mendel**, also known as the "Father of Genetics" (interesting name for a monk). In 1866, Mendel published a paper that documented a breeding experiment in garden pea plants. Using several different traits (including seed coat color and texture), Mendel was the first to understand that the traits of an organism are determined by bits of genetic information, or **genes**. At the time, Mendel just referred genes as genetic " factors". He assumed the factors came in different forms, which we now call **alleles.**

Mendel had to infer that the genes of an organism determined what the organism would look like, and that the factors would be passed from parents to offspring. The **appearance or traits** of an organism are called its **phenotype**. The specific combination of factors parents pass to offspring is called the **genotype**. A phenotype is what we can see, therefore it is observable. Until recently, a genotype was unobservable and had to be inferred. Based on his principles, Mendel was able to make predictions (hypotheses) about how many different phenotypes should result from crossing one type of parent to another.

Mendel's data collected from breeding pea plants made him propose that all the traits he studied were controlled by **pairs of genes. Another way of saying this is—every trait is controlled by a pair alleles for a gene. He then made the following proposals about how inheritance works. (His proposals were more general at the time he made them).

- Segregation: When an organism makes sex cells in the process of meiosis, the pairs of chromosomes (an thus the pairs of genes/alleles it carries) segregate into separate sex cells. Sex cells carry one of each gene, not pairs).

- Alleles can be recessive or dominant : For the traits Mendel studied, he saw that the alleles for each gene are not blended; the alleles of each gene are dominant or recessive to each other. The dominant trait shows in individuals with one or two dominant alleles. The recessive trait shows only in individuals with two recessive alleles (no dominant allele)

- Alleles for different genes are not physically connected: segregation of one gene controlling one trait is not affected by the segregation of another gene for an different trait. Therefore the presence/absence of one trait in offspring is not affected by the presence/absence of another trait.

- **Although there are notable extensions of Mendel's observations---he was spot on about how inheritance works. And it applies to humans.**

For humans….
Your parents each have 46 chromosomes in all of their cells. In reality humans have 23 kinds of chromosomes, and a pair of each. When your mother and father made their gametes (egg and sperm), they split up the pairs of chromosomes so that each gamete received only 23 chromosomes. Because they each contributed 23 chromosomes, you now have 2 sets of 23 chromosomes =46 chromosomes. (This represents segregation, just like Mendel proposed).

Each chromosome carries only one copy or allele of each gene; its matching chromosome also carries only one allele of the same gene, but the alleles may be different! The simplest difference in alleles was referred to by Mendel as dominant vs. recessive.

We use symbols (usually using letters) to keep track of genetics and inheritance. **Capitalized or uppercase letters refer to dominant alleles while lowercase letters refer to recessive alleles.** Every individual has two alleles and we list them both—as their genotype. For example, "E" or "e" will be used to symbolize the alleles for earlobe phenotype. Earlobes can be attached or unattached to your head. Studying earlobes in many families shows that unattached earlobe allele **E** seems to be dominant to attached **e**. We even say the trait, unattached earlobes is dominant to the attached earlobe trait or attached is recessive to unattached. We can inherit 3 possible genotypes for the earlobe gene:
EE (unattached) **Ee** (unattached) or **ee** (attached). (Which on might you have?)

One of the coolest things Mendel showed us is that recessive traits can skip generations—-- parents who both show the dominant form of the trait can produce children who show the recessive form. An example is when 2 parents with brown eyes have a child with blue eyes. (But sometimes diseases are recessive and parents may not show the disease--- but end up passing along recessive alleles to a child, who will show the disease). Try to work out how this works, using pairs of alleles with letter symbols!

Your instructor will take you through some examples to show you how to use a handy visual tools called a Punnett Square. Please use this when trying to determine what allele combinations are possible in the offspring of two individuals.

Keep in mind that most traits in multicellular organisms are not controlled by only one gene (e.g., height is controlled by many genes and by environmental factors as well). In this lab we keep things simple by examining traits that are controlled by one gene only (e.g., skin freckles).

Procedures

PART A: Analysis of several single gene characteristic in humans

An important and useful tool provided by Mendel is that one's genotype can often be inferred by knowing the phenotype of the individual's parents, grandparents, children, etc. Furthermore, one can also infer whether the alleles are dominant or recessive.

If individuals are **homozygous dominant** (e.g., AA) or **heterozygous** (e.g., Aa), their phenotype will show the dominant characteristic. If individuals are **homozygous recessive** (aa), their phenotype will show the recessive characteristic.

Work with a partner to determine your phenotype for the traits listed in Table 1. Record your phenotype and possible genotypes (circle the letters on the appropriate line). After the totals for class are tallied, determine the majorities for each characteristic.

Table 1. Genetic analysis of personal characteristics

Your phenotype	Your genotype	Dominant or Recessive?
Earlobes		
-unattached	EE or Ee	
-attached	ee	
Tongue Rolling		
-can	RR or Rr	
-cannot	rr	
Skin pigmentation		
-freckles	FF or Ff	
-no freckles	ff	
Hair on back of hand		
-present	HH or Hh	
-absent	hh	
Thumb hyperextension		
-cannot be bent back	TT or Tt	
-can be bent back 60°	tt	
Bent pinky		
-pinky bends	LL or Ll	
-straight pinky	ll	
Interlacing of fingers		
-left thumb over right	II or Ii	
-right thumb over left	ii	

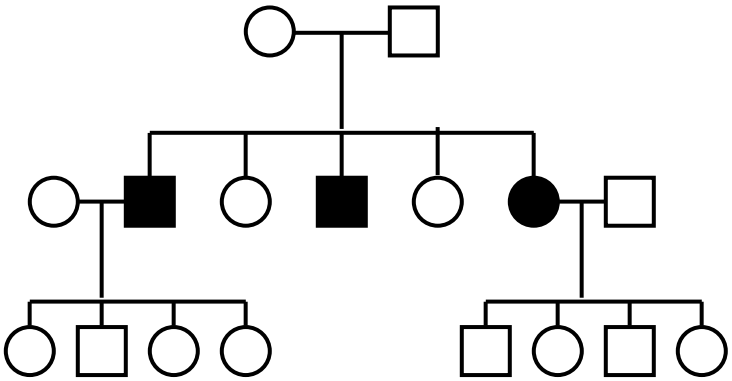
PART B: Human genetics and pedigree analysis

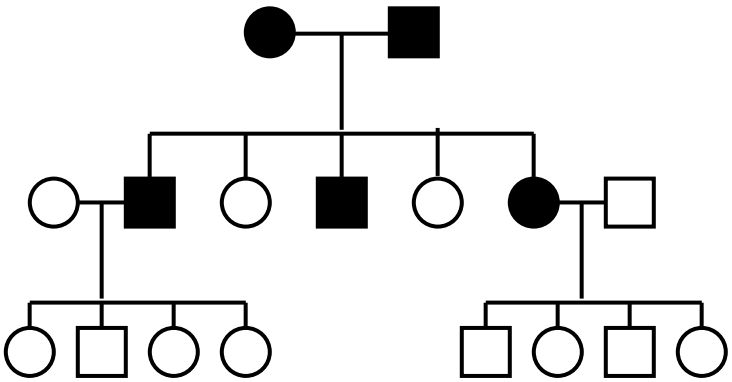
Genetic counselors are sometimes able to identify parents who are likely to produce children with genetic disorders. Fetal cells can then be tested to determine if the newborn does indeed have the disorder. This is called prenatal analysis.

Another type of genetic counseling uses **pedigree analysis**. Pedigree charts show the inheritance of a genetic disorder within a family and make it possible to determine whether any particular individual has an allele for that disorder. It allows families to understand where a gentic disease arose, and to predict a probability for passing the disorder on.

In pedigree charts, symbols are used to indicate:
normal (clear) and affected (filled-in),
males (squares) and females (circles),
reproductive partners (linked at midline), and
siblings (linked from above)

For the below practice pedigrees, determine how the characteristic is passed. Is it dominant or recessive? (Hint: use letter symbols to figure this out.) For example, try "dominant". If the trait is dominant everyone who is shaded will have a dominant allele. See whether this can work, in this family. If not, try recessive…





Name_____

Requirements Lab 7 –When applicable, please explain your answers with references to your data. (use Punnett Squares and percentages).

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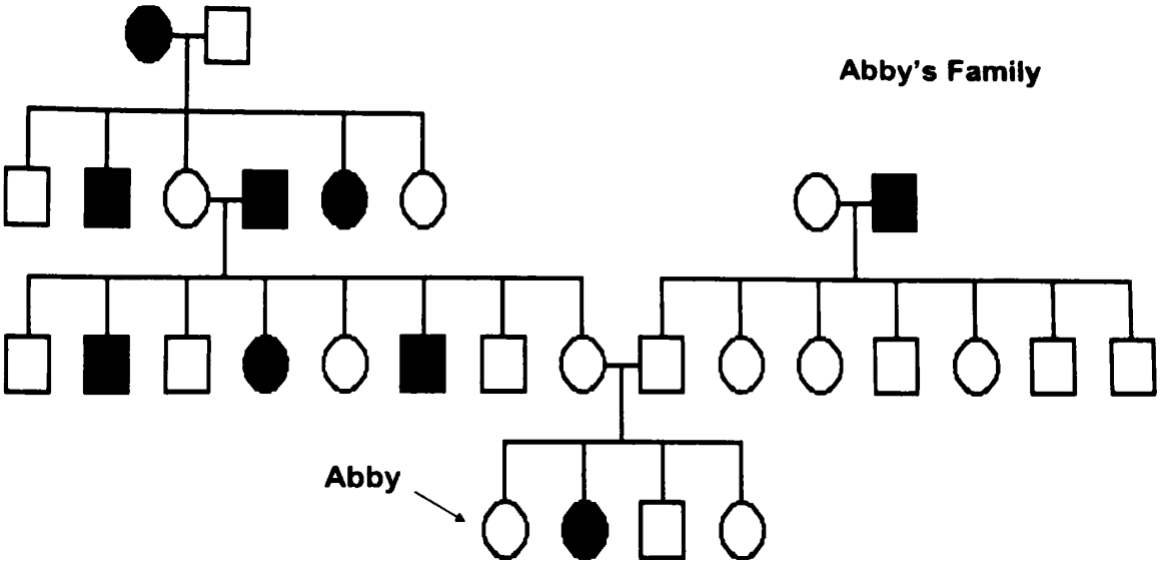
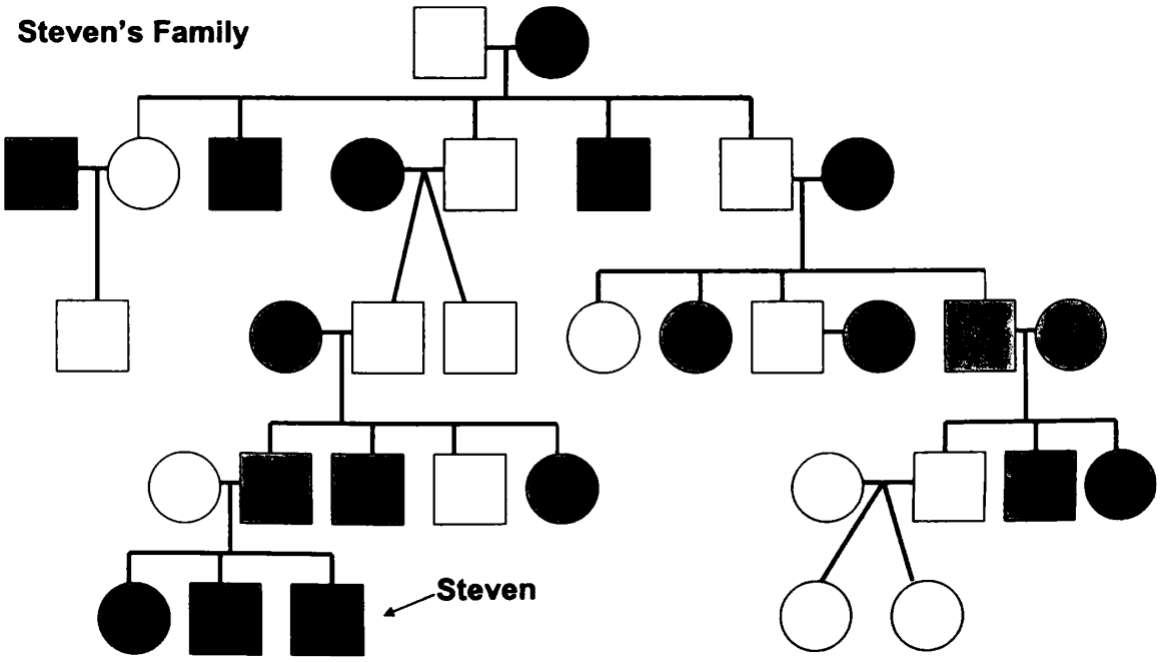
1. In general speech, sometimes the term *dominant* is interpreted as "better" or "more beneficial" than *recessive*. Based on Table 1, do you think the dominant form of the traits is necessarily "more beneficial" than the recessive form of those traits? Does dominant mean "better" in genetics? *(3 pts)*

Choose **two of the four cases** below, **fill in the applicable pedigrees** and answer the associated questions. Show Punnett squares and likelihoods of passing a trait whenever possible! *(6 pts each)*

Case #1 Steven & Abby: For all crosses, assume the spouse does not express/show the trait of the other family.



Abby's family has a history of Phenlketonuria (PKU). Individuals with PKU lack the enzyme that converts a nonessential amino acid (phenyalanine) into a useful one (tyrosine). High levels of phenyalanine accumulate in the blood affecting neuronal development—especially in children. Mental retardation can be a result. Since phenylalanine is found in many proteins, patients afflicted with PKU can escape the disease by strictly limiting themselves to low protein diets.



Steven has Huntington disease. Huntington's disease is characterized by degeneration of the nervous system causing uncontrollable movements, dementia, and psychiatric disturbances. Loss of motor skills eventually prevent swallowing and speech. Huntington's generally develops in a person's thirties or forties, leaving the afflicted children and spouse to care for them.

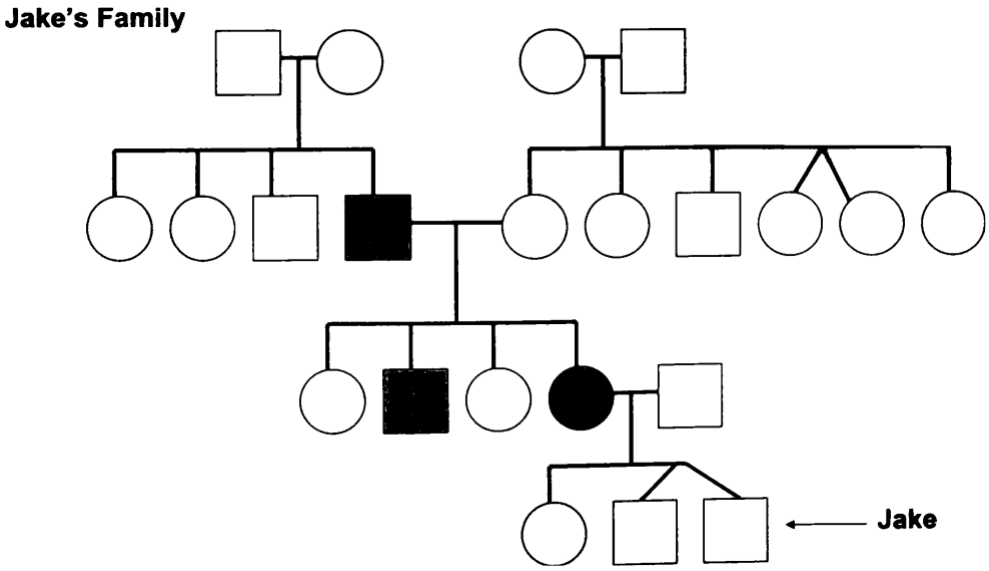
A. Is it possible for Steven & Abby's children to express PKU? Explain using Punnett squares and percentages.

B. Is it possible for Steven & Abby's children to express Huntington's disease? Explain using Punnett squares and percentages.

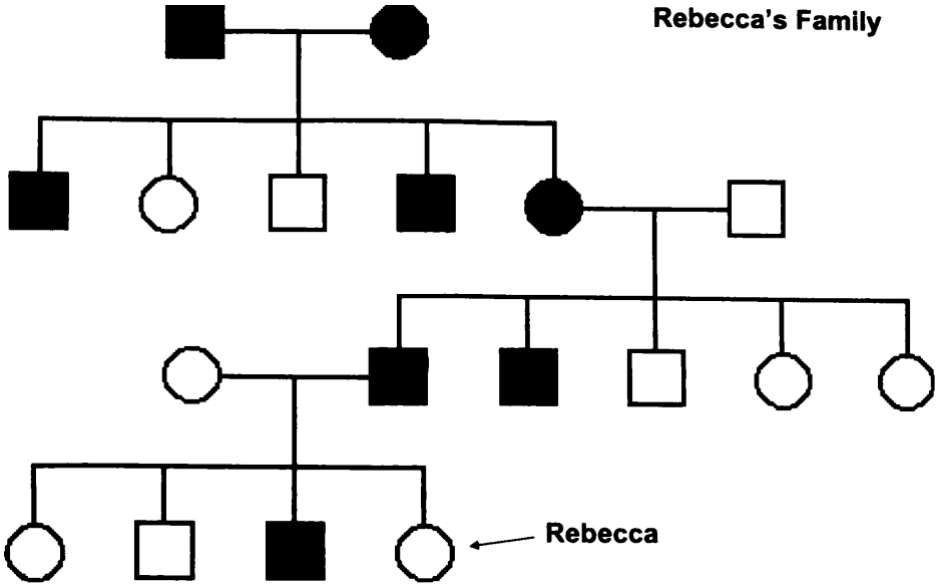
C. Is PKU dominant or recessive? Is Huntington's dominant or recessive?

Be sure you filled in all genotypes in both pedigrees!

Case #2 Jake & Rebecca: For all crosses, assume the spouse does not express/show the trait of the other family.



Jake's family has the most common lethal genetic abnormality in the United States—Cystic Fibrosis. Cystic Fibrosis is characterized by excessive secretion of mucus from the lungs, pancreas, and other organs. The mucus is very thick, causing problems with breathing, digestion and liver function. It also makes the person especially vulnerable to infections like pneumonia. Without a special diet and frequent pounding on the chest and back (to clear the lungs of mucus), most children with CF die by the age of 5.



A rare trait in Rebecca's family causes Marfan's syndrome, in which the connective tissue is not as rigid as it should be. The long bones of the body continue to grow causing an abnormally tall, thin individual, with a narrowed face. The lens of the eyes may be dislocated. The heart valves and arteries stretch and leak, Lung, skin and neurological problem are also common. Rebecca does not have Marfan's, but is concerned she might be a carrier of the disease.

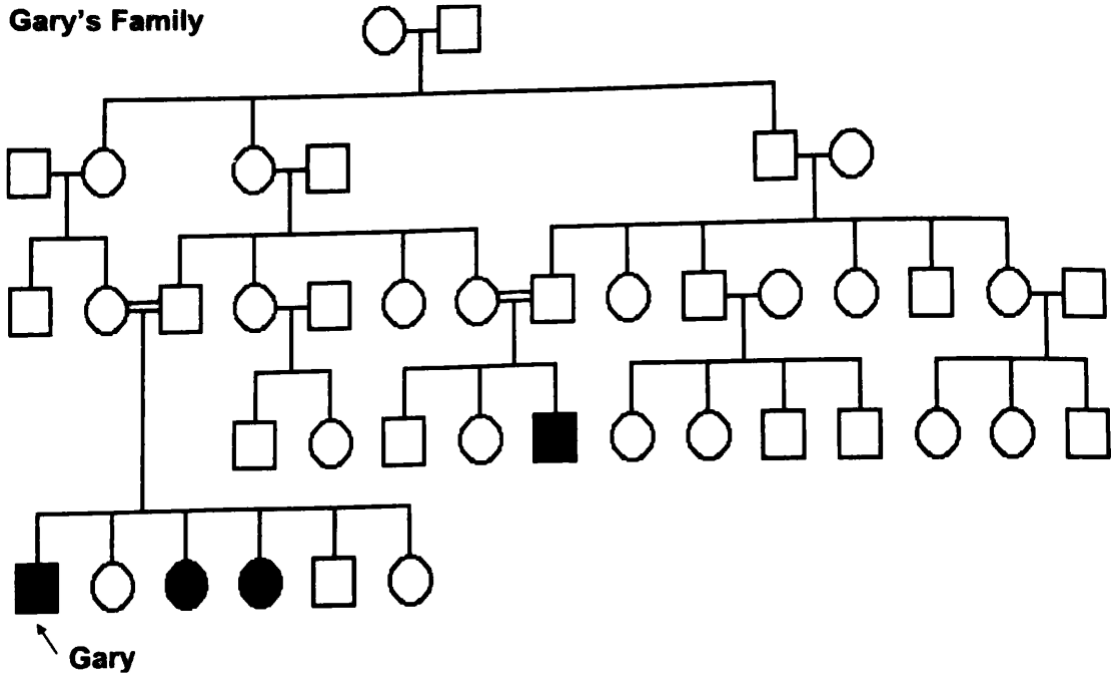
A. Is it possible for Jake & Rebecca's children to express Cystic Fibrosis? Explain using Punnett squares and percentages.

B. Is it possible for Jake & Rebecca's children to express Marfan's Syndrome? Explain using Punnett squares and percentages.

C. Is Cystic Fibrosis dominant or recessive? Is Marfan's Syndrome dominant or recessive?

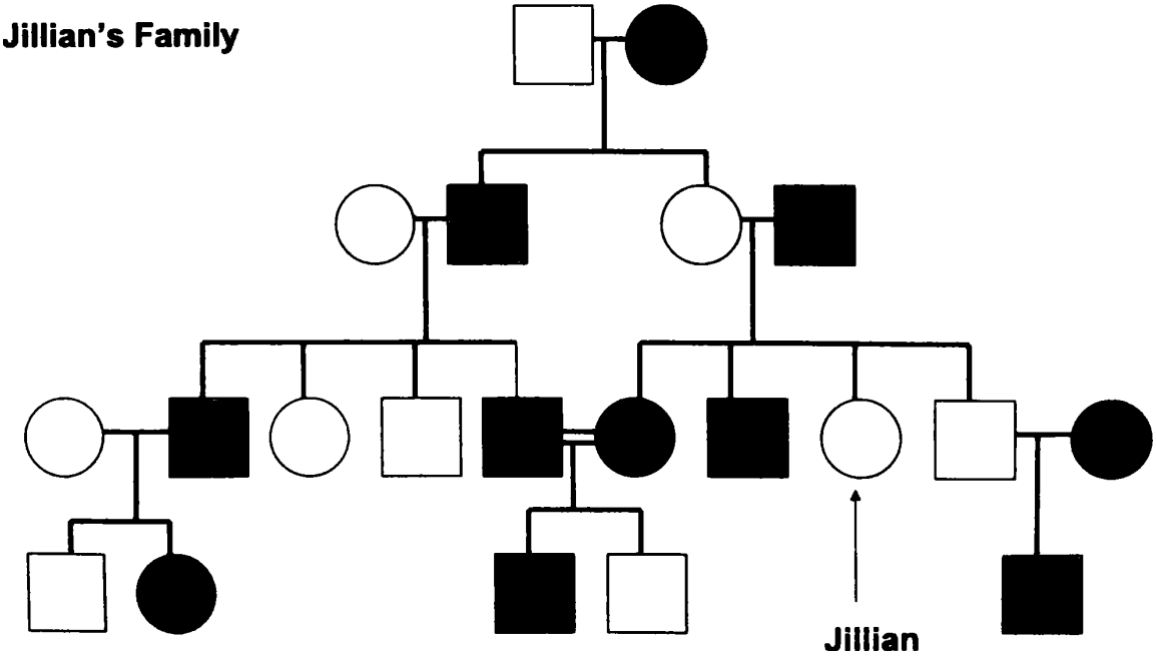
Be sure you filled in all genotypes in both pedigrees!

Case #3 Gary & Jillian: For all crosses, assume the spouse does not express/show the trait of the other family.



Gary has Albinism. Those with this trait lack pigment, causing very pale skin, hair and eyes. They tend to suffer more UV damage than those without Albinism, and are therefore forced to wear long sleeves, pants, sunglasses, hats and gloves whenever exposed to sunlight.

Jillian's Family



Many of Jillian's family have a type of dwarfism called Achondroplasia. In this condition, the head and torso develop normally, but the limbs are short. Only heterozygous individuals have this disorder. The homozygous genotype causes death of the embryo. Jillian does not have this Achondroplasia, but wants to be sure she will not pass on the disorder.

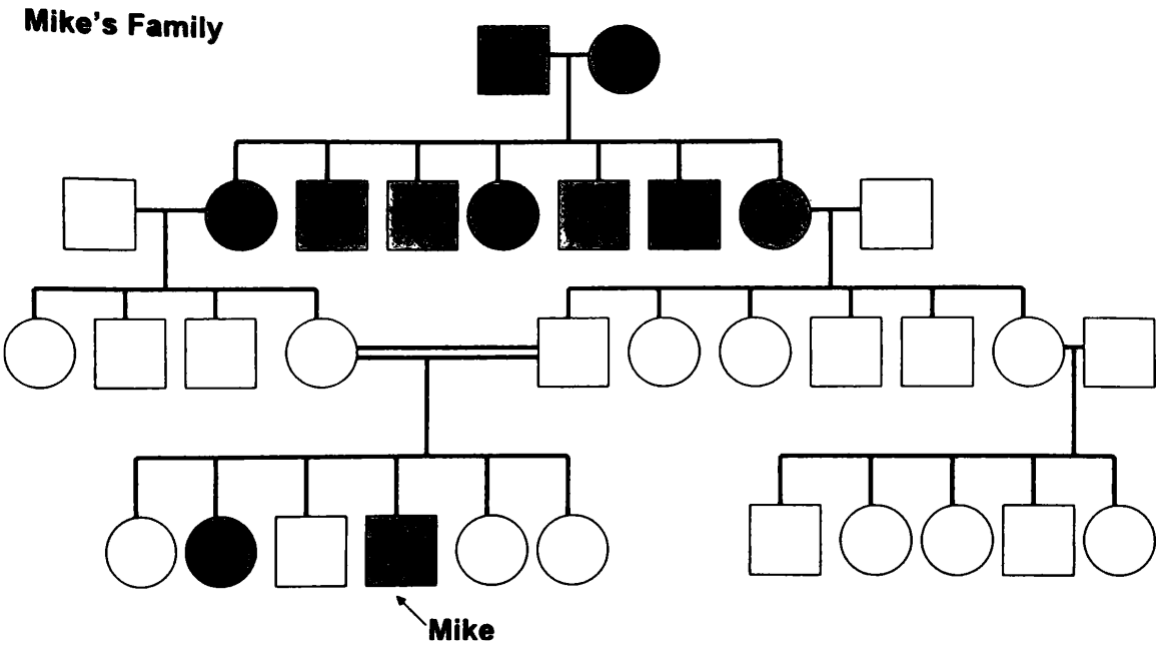
A. Is it possible for Gary & Jillian's children to express Albinism? Explain using Punnett squares and percentages.

B. Is it possible for Gary & Jillian's children to express Achondroplasia? Explain using Punnett squares and percentages.

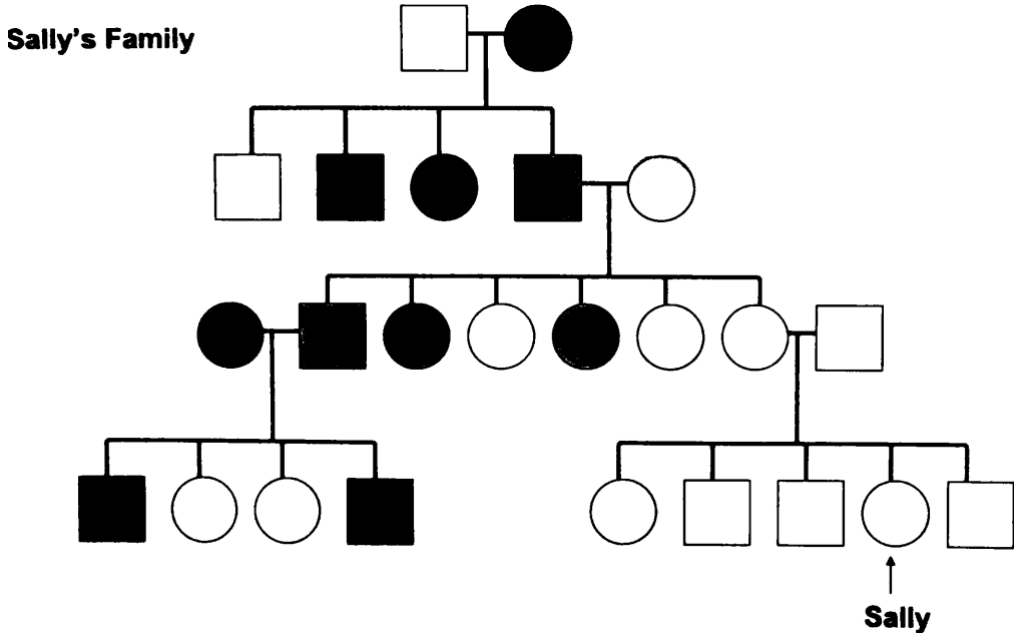
C. Will the cousin marriage in Jillian's family produce Albinos? Explain why or why not.

Be sure you filled in all genotypes in both pedigrees!

Case #4 Mike & Sally: For all crosses, assume the spouse does not express the trait in question.



Leptodigitalia occasionally shows up in Mike's family. Leptodigitalia is a condition where the fingers and toes are excessively long and slender. Such digits are extremely fragile and often suffer frostbite in conditions considered normal to unaffected individuals. Mike has this trait, but does not wish to pass it on to his children.



Sally's family has genetic predisposition to Syndactyly, a condition in which two or more digits are fused together. While she does not exhibit this condition, she is concerned that she might be a carrier of this trait.

A. Is Leptodigitalia a dominant or recessive trait? Does Mike possess the Leptodigitalia allele?

B. Is Syndactyly a dominant or recessive trait? Does Sally possess the Syndactyly allele?

C. What are the odds that Mike & Sally's children will express both Leptodigitalia and Syndactyly? Explain using Punnett squares and percentages.

Be sure you filled in all genotypes in both pedigrees!