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Autosomal Recessive Inheritance

Phenylketonuria (PKU) is a recessive condition where sufferers lack an enzyme to break down tyrosine in their diet. Because it is recessive, both parents must pass down the gene in order for a child to inherit the disorder. PKU, if left untreated, causes brain damage and retardation.

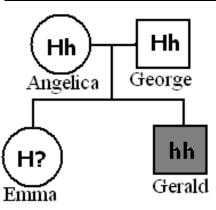
Both Angelica and George are healthy, but they are heterozygous (carriers) for the PKU disorder. This means they each contain one healthy gene (H) but also one recessive gene (h). Angelica and George have two children, Emma (age 12) and Gerald (age 7). Sadly, Gerald has been diagnosed with PKU, but Emma has not.

Example: In the space below, complete a Punnett square in the space below.

Step 1: Set up the Punnett square below Step 2: Fill in the Punnett square below

- 1. According to the Punnett Square, what were the chances that Emma would be heterozygous/carrier?
- 2. According to the Punnett Square, what were the chances that Gerald would inherit PKU?
- 3. According to the Punnett Square, what were the chances that Angelica and George would have a child that was homozygous dominant?

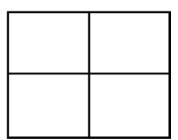
Example: Create a Pedigree in the space below that outlines all four people.



- 4. According to the Pedigree, which symbols show women?
- 5. According to the Pedigree, which symbols show men?
- 6. According to the Pedigree, how are individuals who suffer from the disorder marked?
- 7. According to the Pedigree, what is another word for people who are carriers of the disorder?

8. Elizabeth is a carrier of PKU and is married to Alan, who is homozygous recessive for the PKU disease. They just gave birth to their first child, a girl named Sarah.

Complete their Punnett square below.

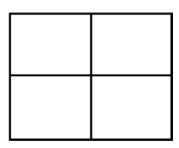


- a. According to the Punnett square, what are the odds that Sarah will have PKU?
- b. According to the Punnett square, what are the odds that Sarah will be homozygous dominant?
- c. According to the Punnett square, what are the odds that Sarah will be heterozygous?
- d. If Elizabeth and Alan have a second child, what are the odds that he/she will be healthy?

<u>Create a Pedigree in the space below that outlines all three people. Be sure to record the genotype next to each circle or square.</u>

9.	Marsha and Peter are trying to start a family. Marsha has no family history (HH) of cystic fibrosis.
	Peter is a carrier (Hh) of the cystic fibrosis gene because his mother and father were also both
	carriers. Cystic fibrosis is a homozygous recessive (hh) disorder where mucus will build up in the
	lungs of sufferers. This leads to excess strain on the heart and respiratory system and usually death
	around mid-30's.

Fill in the Punnett Square for Marsha and Peter below.



- a. What is the probability that Marsha and Peter will have a healthy child?
- b. What is the probability that Marsha and Peter will have a heterozygous child?
- c. What is the probability that Marsha and Peter will have a homozygous recessive child?

<u>Create a Pedigree in the space below that outlines all four people mentioned. Be sure to record the genotype next to each circle or square.</u>

10. Albinism is a recessive disorder where individuals lack melanin pigment in the eyes, skin and	hair, or
more rarely in the eyes alone. A normally pigmented man, whose father was an albino, marrie	s an
albino woman both of whose parents were normally pigmented. They have three children, two	ı
normally pigmented, and one albino. (make the children any gender you wish)	

Fill in the Punnett Square for the man and woman below.



- a. What is the probability that the man and woman will have a normal-pigmented child?
- b. What is the probability that the man and woman will have an albino child?
- c. What is the probability that the man and woman will have a homozygous dominant child?

<u>Create a Pedigree in the space below that outlines all eight people mentioned. Be sure to record the genotype next to each circle or square.</u>