SUCCESS ACADEMY EDUCATION INSTITUTE

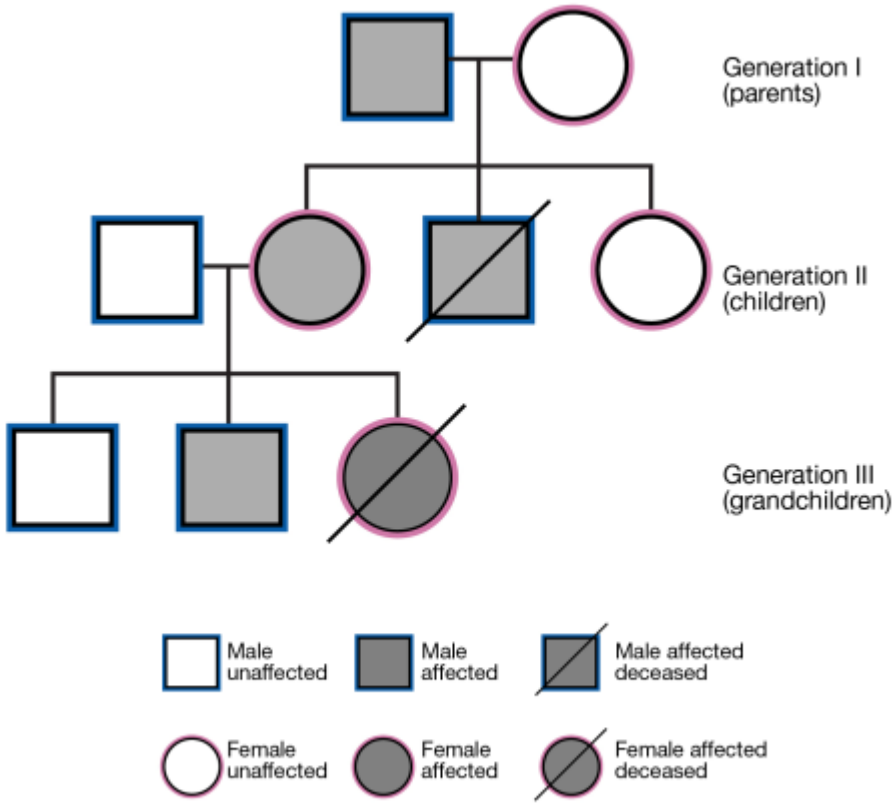
Patterns in Pedigrees Work Packet

Life Science (Lower Middle)
Unit 2

Patterns in Pedigrees Work Packet

Directions: Review the following information on pedigree charts. Then, go through each pedigree example and answer the questions that follow.

A pedigree is a genetic representation of a family tree that diagrams the inheritance of a trait or disease though several generations. The pedigree shows the relationships between family members and indicates which individuals express or silently carry the trait in question.



Patterns in Pedigrees Work Packet

A pedigree is a representation of our family tree. It shows how individuals within a family are related to each other. We can also indicate which individuals have a particular trait or genetic condition. If we take a pedigree, which we usually try to include at least three generations, we might be able to determine how a particular trait is inherited. Using that information, we might be able to tell the chance that a given individual will have the trait themselves or could pass it on to their children.

There are standard ways to draw pedigrees so that we can all look at a pedigree and understand it. We use squares to represent males and circles to represent females. We then can number our generations with roman numerals, so the top generation would be generation one, or Roman numeral I. Along this line, we'd indicate males and females. We would indicate marriages between individuals with a horizontal line connecting the two individuals. If an individual has a genetic trait, we would blacken those individuals in or shade them so that it would be understood that they had a particular trait. We would then draw a line, a vertical line, off the horizontal line where we would indicate any of their children that they had, and we would then indicate if any of their children were infected. And we can do this for as many generations as we have.

It's important when we draw a pedigree that we try to put in as much information as possible. So for example, if there have been children that died in early infancy or were stillborn, we also want to include those individuals. And those are typically shown as very small blackened-in symbols to indicate there was a loss of a child, either in pregnancy or early in life.

Excerpt Courtesy of National Human Research Institute (NHGRI)

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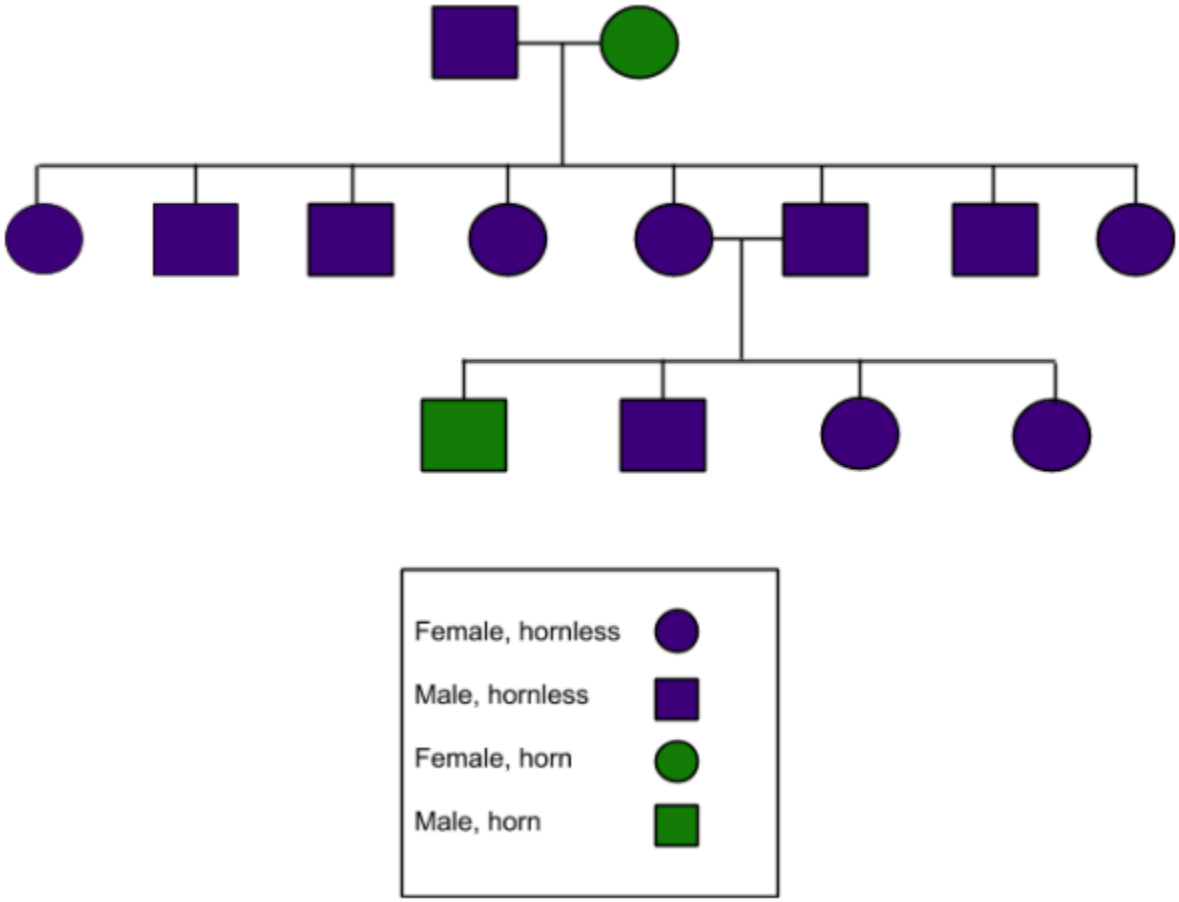
Example #1: Cattle Horns

Pedigree Chart of Horn Presence in Cattle Family



1. How can you determine if horn presence is a dominant or recessive trait?

2. Using alleles “H” and “h,” label each family member’s genotype on the pedigree.

3. In generation 3, how could we test to determine the genotype of the hornless cattle?

Example #2: Human Condition Part I

Cystic fibrosis (CF) is the most common, fatal genetic disease in the United States. About 30,000 people in the United States have the disease. CF causes the body to produce thick, sticky mucus that clogs the lungs, leads to infection, and blocks the pancreas, which stops digestive enzymes from reaching the intestine where they are required in order to digest food.

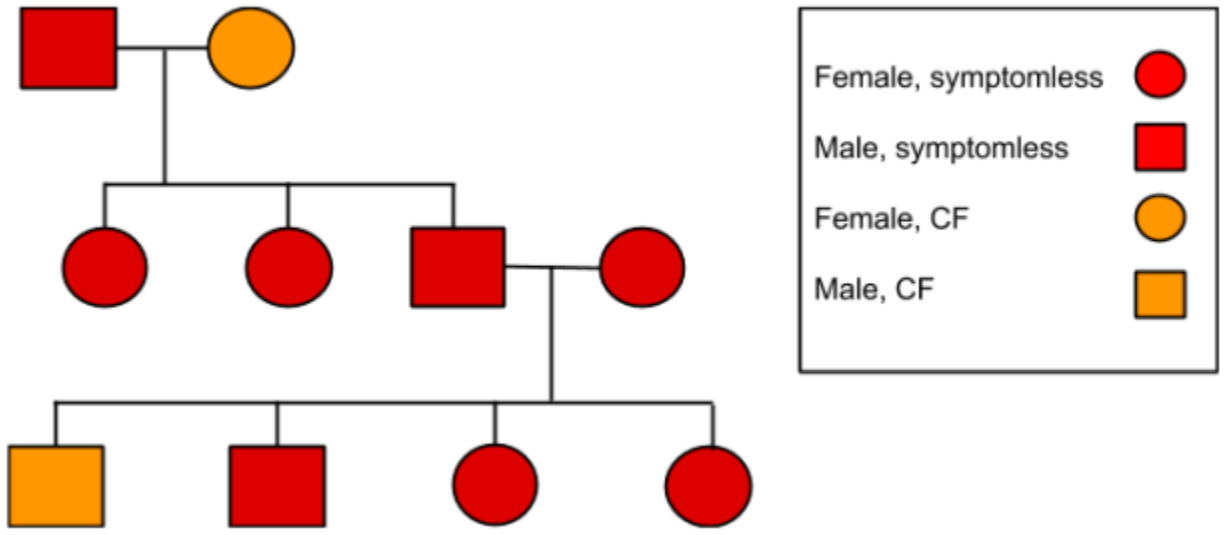
In normal cells, the CFTR protein acts as a channel that allows cells to release chloride and other ions. But in people with CF, this protein is defective and the cells do not release the chloride. The result is an improper salt balance in the cells and thick, sticky mucus. Researchers are focusing on ways to cure CF by correcting the defective gene, or correcting the defective protein.

CF has a variety of symptoms, including very salty-tasting skin, a persistent cough and excessive appetite but poor weight gain. The "sweat test" - which measures the amount of salt in sweat - is the standard diagnostic test for those with symptoms. A high salt level indicates CF.

Gene therapy offers great promise for life-saving treatment for CF patients since it targets the cause of CF rather than just treating symptoms. Gene therapy for CF had its start in 1990, when scientists successfully corrected faulty CFTR genes by adding normal copies of the gene to laboratory cell cultures.

Excerpt Courtesy of National Human Research Institute (NHGRI)

Pedigree Chart of Cystic Fibrosis in a Human Family



4. Based on the pedigree, is CF likely a dominant or recessive trait? Why?

5. Using alleles “F” and “f,” label each family member’s genotype on the pedigree.

6. Scientists use the word “carrier” to describe someone who does not have the genetic disease, but carries the genetic mutation in one allele and is capable of passing down the disease to their offspring. Place a star (★) next to any family members who might be a carrier.

7. Record the possible parent combinations that could produce offspring with the genetic disease of CF.

Example #3: Human Conditions Part II

Marfan syndrome is one of the most common inherited disorders of connective tissue occurring once in every 10,000 to 20,000 individuals. There is a wide variability in clinical symptoms in Marfan syndrome with the most notable occurring in eye, skeleton, connective tissue and cardiovascular systems. Marfan syndrome is caused by mutations in the FBN1 gene.

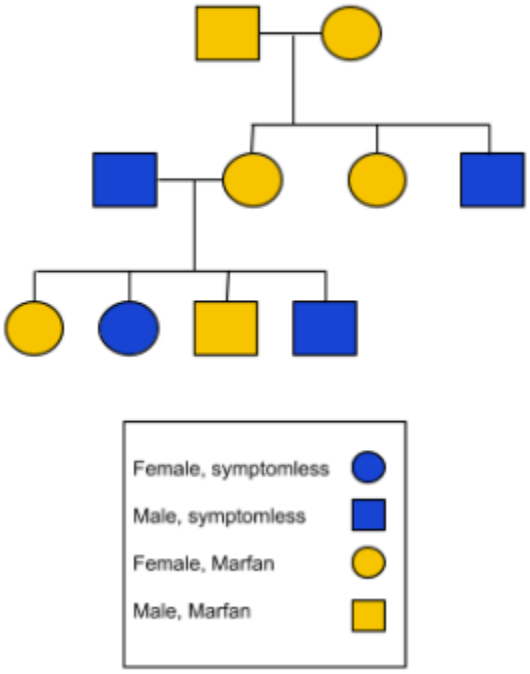
The most common symptom of Marfan syndrome is myopia (nearsightedness from the increased curve of the retina due to connective tissue changes in the globe of the eye). About 60 percent of individuals who have Marfan syndrome have lens displacement from the center of the pupil (ectopia lentis).

Other common symptoms of Marfan syndrome involve the skeleton and connective tissue systems. These include bone overgrowth and loose joints (joint laxity). Individuals who have Marfan syndrome have long thin arms and legs (dolichostenomelia). Overgrowth of the ribs can cause the chest bone (sternum) to bend inward (pectus excavatum or funnel chest) or push outward (pectus carinatum or pigeon breast). Curvature of the spine (scoliosis) is another common skeletal symptom that can be mild or severe and progressively worsen with age. Scoliosis shortens the trunk also contributes to the arms and legs appearing too long.

Individuals who have Marfan syndrome are treated by a multidisciplinary medical team that includes a geneticist, cardiologist, ophthalmologist, orthopedist and cardiothoracic surgeon.

Adapted Excerpt Courtesy of National Human Research Institute (NHGRI)

Pedigree Chart of Marfan Syndrome in a Human Family



8. Based on the pedigree, is Marfan Syndrome likely a dominant or recessive trait? Why?

9. Using alleles “R” and “r,” label each family member’s genotype on the pedigree.

10. Why doesn’t this genetic condition skip every other generation?

Example #4: Human Condition Part III

Codominance is a relationship between two versions of a gene. Individuals receive one version of a gene, called an allele, from each parent. If the alleles are different, the dominant allele usually will be expressed, while the effect of the other allele, called recessive, is masked. In codominance, however, neither allele is recessive and the phenotypes of both alleles are expressed.

Phenotype	Red	Pink	White
Genotype	RR	Rr	rr

Codominance means that neither allele can mask the expression of the other allele. An example in humans would be the ABO blood group, where alleles A and alleles B are both expressed. So if an individual inherits allele A from their mother and allele B from their father, they have blood type AB.

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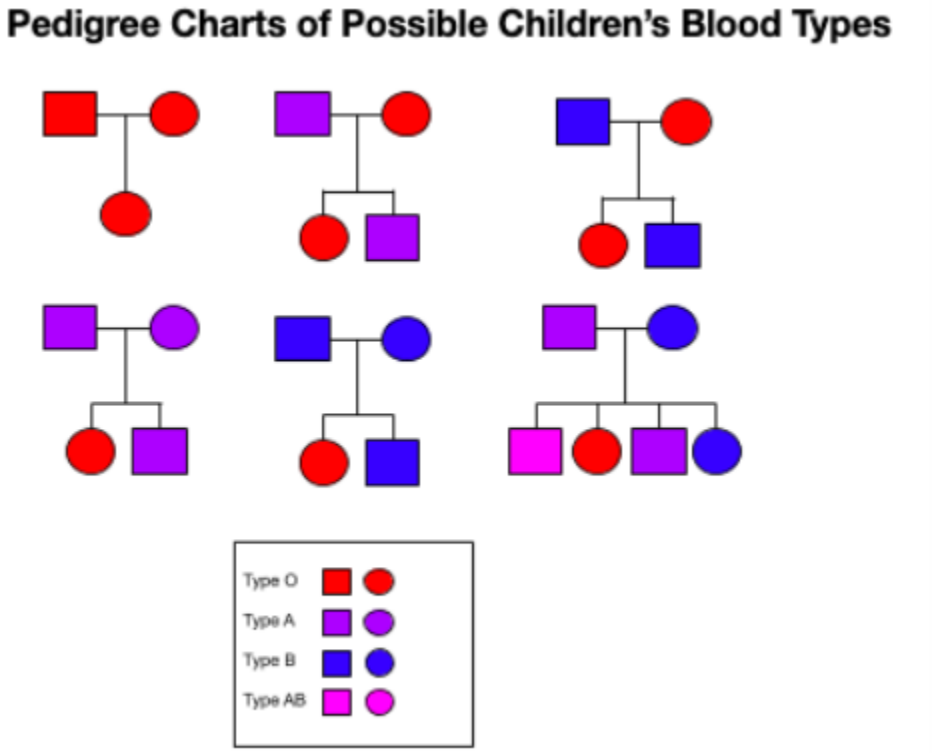

11. Based on the pedigree, how is blood type inherited?

12. Determine the possible genotypes of the four blood types in the table below.

ABO Blood Groups	
Blood Type	**Possible Genotype(s)**
O	
A	
B	
AB	

13. What blood groups are codominant? Which blood groups are recessive?