Pedigree Practice Activity

**Instructions:**
1. Draw a pedigree showing all the individuals described in the problem. (Include their names if given.)
2. Label the genotypes of as many individuals in the pedigree as possible.
3. Shade in half of the symbol if you know that the individual is heterozygous or a carrier.

**Draw your own Pedigree - Case study #1:** Condition of Interest: Albinism

Albinism is a condition in which there is a mutation in one of several possible genes, each of which helps to code for the protein melanin. This gene is normally active in cells called melanocytes, which are found in the skin and eyes. Albinism involves a significant reduction or absence of the production of melanin, giving affected individuals a lack of normal coloration to their skin/eyes. Inheritance Pattern: normal melanin protein is produced by an autosomal dominant allele; albinism results from a lack of melanin and is caused by an autosomal recessive allele. Use the letter A or a to represent dominant/recessive forms of albinism.

1. Two normally-pigmented parents have 3 children. The first child (a girl) and their second child (a boy) have normal pigmentation. Their third child (a girl) has albinism. That girl marries a normally pigmented male and they have four children. The first three (two girls and a boy) have normal pigmentation. Their fourth child (a girl) has albinism like her mother.

**Draw your own Pedigree - Case Study #2:** Condition of Interest: Huntington's Disease (also known as HD or Huntington's chorea)

Huntington's disease is a neurodegenerative genetic disorder that affects muscle coordination and leads to cognitive decline and dementia. Inheritance Pattern: the allele for the normal "Huntingtin" protein is autosomal recessive; Huntington's disease is caused by an autosomal dominant allele which codes for an abnormal form of the "Huntingtin" protein. Symptoms are more severe in homozygous individuals. Use H or h to represent the alleles.

2. A normal man (Joseph) marries a woman (Rebecca) who is heterozygous for HD and they have four children. Two of their sons (Adam and Charles) are born healthy without HD. Charles marries a woman without HD and they have a normal daughter. Joseph and Rebecca's daughter Tasha and their last son (James) both have HD. James marries a non-HD woman whose sister and parents also do not suffer from HD. James and his wife have three children - a normal boy, a normal girl, and a son with HD.

**Draw Your Own Pedigree - Case Study #3: Trait: blood type**

Blood type is determined by the presence of several different proteins found on the surface of red blood cells. Blood type “A” has the A protein; blood type “B” has the B protein; blood type AB has both; blood type O has neither. The +/- indicates another protein called Rh. **Forms of the trait:** inheritance via autosomal multiple allelism (A, B, or O) results in the blood types A, B, AB or O. The alleles for blood protein A and B are codominant, the "O" allele is recessive to both the A and B alleles. Use AA, AO, AB, BB, BO or OO to represent the genotypes.

3. As a 9th-grade school project, Maureen decides to trace the inheritance of blood types through her extended family, all the way back to her great-grandmother Katherine. Here's what Maureen found out….

Maureen's great-grandmother Katherine, has A type blood. Katherine and her husband John had four children – two sons, Michael (who has blood type AB) and David (who has type O blood); a daughter (Jessica) with type O blood and another daughter (Jennifer) with type A blood. Jessica never married; her sister Jennifer did get married and had three sons (one with type A blood, one with type AB blood and one with type O blood). Both of Katherine's sons also get married – Michael marries a woman with type O blood and together they have two daughters (Anna – type A; Leanne – type B); David marries a woman with type A blood, and they have three children (daughter Fran and son Albert who both have type A blood, and a son Matthew with type O blood). Matthew marries Janine and together they have one daughter, Maureen. Maureen knows that her parents both have the same blood type, but she has never yet had a blood test to determine her own blood type.
Pedigree Review

For Questions 4-10, use the pedigree chart shown to the right. Some of the labels may be used more than once.

4. A male
5. A female
6. A marriage
7. A person who expresses the trait
8. A person who does not express the trait
9. A connection between parents and offspring
10. How many generations are shown on this chart?

Assuming the chart above is tracing the dominant trait of "White Forelock (F)" through the family. F is a tuft of white hair on the forehead.

11. What is the most likely genotype of individual “A”? (FF, Ff or ff?)
12. What is the most likely genotype of individual “C”? (FF, Ff or ff?)

Practice tracing the path of an autosomal recessive trait

Trait: Falconi anemia

Forms of the trait:

- The dominant form is normal bone marrow function - in other words, no anemia.
- The recessive form is Falconi anemia. Individuals affected show slow growth, heart defects, possible bone marrow failure and a high rate of leukemia.

The pedigree to the right shows a family that carries Falconi anemia. Note that carriers are not indicated with half-colored shapes in this chart.

Analysis Questions:

To answer questions #13-17, use the letter "f" to indicate the recessive Falconi anemia allele, and the letter "F" for the normal allele.

13. What is Arlene's genotype?
14. What is George's genotype?
15. What are Ann & Michael's genotypes?
16. Most likely, Sandra's genotype is?
17. List three people from the chart (other than George) who are most likely carriers of Falconi anemia.

Tracing the path of an autosomal dominant trait

Trait: Neurofibromatosis

Forms of the trait:

- The dominant form is neurofibromatosis, caused by the production of an abnormal form of the protein neurofibromin. Affected individuals show spots of abnormal skin pigmentation and non-cancerous tumors that can interfere with the nervous system and cause blindness. Some tumors can convert to a cancerous form.
- The recessive form is a normal protein - in other words, no neurofibromatosis.

A typical pedigree for a family that carries neurofibromatosis is shown above. Note that carriers are not indicated with half-colored shapes in this chart. Use the letter "N" to indicate the dominant neurofibromatosis allele, and the letter "n" for the normal allele.

Analysis Questions:

18. Is individual #1 most likely homozygous dominant or heterozygous? Explain how you can tell.
19. What is the genotype of individual #3?
20. Can you be sure of the genotypes of the affected siblings of individual #3? Explain.