PRIOR KNOWLEDGE:
1. What do you already know about the difference between dominant and recessive traits? Or sex-linked and autosomal traits?

Dominant trait: Recessive
Sex-linked: Autosomal traits

CASE STUDIES: Read each case study and answer the questions.


Cystic fibrosis is a progressive, genetic disease that causes persistent lung infections and limits the ability to breathe over time. In people with CF, a defective gene causes a thick, sticky buildup of mucus in the lungs, pancreas, and other organs. In the lungs, the mucus clogs the airways and traps bacteria leading to infections, extensive lung damage, and eventually respiratory failure. In the pancreas, the mucus prevents the release of digestive enzymes that allow the body to break down food and absorb vital nutrients. Cystic fibrosis is a genetic disease. People with CF have inherited two copies of the defective CF gene -- one copy from each parent. Both parents must have at least one copy of the defective gene, making the gene for CF autosomal recessive. People with only one copy of the defective CF gene are called carriers, but they do not have the disease. Carriers are not affected by cystic fibrosis because the normal gene compensates for the faulty one.

The diagram below is called a pedigree and it illustrates how genes for diseases like Cystic Fibrosis can be passed from parents to their children.

![Pedigree Diagram]

2. Using the letters A and a, what is the genotype for each of the following people?

Person 1: **aa**
The father of person 2: **Aa** → person #2 has **CF** - **aa**
Both parents of person 3: **Aa**
Father would have to be **Aa**
3. Is Cystic Fibrosis dominant or recessive? Use evidence from the reading and pedigree to support your answer.

Recessive: looking at the pedigree you can tell that only individuals that inherit both the genes have it & those individuals that have siblings don't have it.

4. Person 4 has a child with someone who is a carrier of CF. Complete a Punnett Square to determine the chance their offspring will have Cystic Fibrosis.

\[
\begin{array}{cc}
A & a \\
\hline
AA & \frac{25}{25} \\
Aa & \frac{25}{25} \\
aa & \frac{25}{25}
\end{array}
\]

Each box 25% => 25% chance of having a child with CF.

4. Would be \(\frac{Aa}{Aa}\) they don't have CF but would carry the gene. \(\frac{Aa}{Aa}\) mother has CF.

Case Study 2: Huntington's Disease (adapted from: http://hda.org/what-is-hd/) Image from: http://www.openpathways.com/w/page/22377142/Huntingtons-Disease

Huntington's Disease, or Huntington's Chorea, is a rare autosomal dominant disease that results in the slow deterioration of the physical and mental abilities of the affected person. Experts estimate that one in every 10,000 people (about 30,000 Americans) suffer from Huntington disease and another 150,000 are at risk for developing it. Many describe the symptoms of HD as having ALS, Parkinson's and Alzheimer's – simultaneously.

Symptoms usually appear between the ages of 30 to 50, and worsen over a 10 to 25 year period. Ultimately, the weakened individual succumbs to pneumonia, heart failure or other complications. Every person who inherits the HD gene will eventually develop the disease. Over time, HD affects the individual's ability to reason, walk and speak. Since the disease does not manifest until a late age, an affected person will often have children already. You can take genetic tests to see if you are at risk for the disease.

Use this pedigree to answer the questions about Huntington's.

5. Using the letters H and h, what is the genotype for each of the following people?

Person 1: \(hh\)  \(\#\) h
Person 2: \(\#h\)
Person 3: \(\#h\)
6. Is Huntington’s dominant or recessive? Use evidence from the reading and pedigree to support your answer.

**Individuals that have Huntington’s disease only have to inherit one allele to have the genetic disorder**

7. Person 3 has a child with someone who does not have Huntington’s. Complete a Punnett Square to determine the chance their offspring will have Huntington’s.

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+ +  + +  + +  + h
+ h  + h  + h  + h
h h  h h  h h  h h

Person 3: Hh  Person who does not: hh

*50% chance their offspring will have Huntington’s*
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8. Sex linked traits are typically carried on the X chromosome. To denote the genotypes, a superscript is used. A female carrier of hemophilia will have the genotype X^H^X^h. Using the letters X and Y, what is the genotype for each of the following people?

- Person 1: X^H^X^h - female *is a carrier*
- Person 2: X^h^Y - male *has hemophilia*
- Person 3: X^H^Y - male

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**Case Study 3: Hemophilia** (adapted from: https://www.hemophilia.org/Bleeding-Disorders/Types-of-Bleeding-Disorders/Hemophilia-A image from: https://www.geneticseducation.bradford.ac.uk/images-of-inheritance/x-linked-conditions/)

Hemophilia A is a genetic disorder caused by a missing blood clotting protein. People with hemophilia A often bleed longer than other people. Bleeds can occur internally, into joints and muscles, or externally, from minor cuts, dental procedures or trauma. According to the US Centers for Disease Control and Prevention, hemophilia occurs in approximately 1 in 5,000 live births. There are about 20,000 people with hemophilia in the US. All races and ethnic groups are affected.

The X and Y chromosomes are called sex chromosomes. The gene for hemophilia is carried on the X chromosome. Hemophilia is inherited in an X-linked recessive manner. Females inherit two X chromosomes, one from their mother and one from their father (XX). Males inherit an X chromosome from their mother and a Y chromosome from their father (XY). That means if a son inherits an X chromosome carrying hemophilia from his mother, he will have hemophilia. It also means that fathers cannot pass hemophilia on to their sons.

But because daughters have two X chromosomes, even if they inherit the hemophilia gene from their mother, most likely they will inherit a healthy X chromosome from their father and not have hemophilia. A daughter who inherits an X chromosome that contains the gene for hemophilia is called a carrier. She can pass the gene on to her children. Hemophilia can occur in daughters, but is rare. 

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Hemophilia is likened to DMD in the way it is inherited.
9. If you have the gene for hemophilia, will you always have hemophilia? Under what circumstances could you not have hemophilia? Use evidence from the reading and pedigree to support your answer.

- Female has the gene but not hemophilia: $X^h X^h$
- Male has the gene & has hemophilia: $X^h Y$
- Males do not have a back-up gene like females so if they inherit the defect they will have the genetic disorder.

10. Person 3 has a second child, a daughter. Complete a Punnett Square to determine the chance this child will have hemophilia.

<table>
<thead>
<tr>
<th></th>
<th>$X^h$</th>
<th>$X^h$</th>
</tr>
</thead>
<tbody>
<tr>
<td>$Y$</td>
<td>$X^h Y$</td>
<td>$X^h Y$</td>
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</tbody>
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CONCLUSION: