Genetic Counseling

Objective
Students will learn about the field of genetic counseling and will analyze genetic counseling scenarios involving Huntington’s disease and Cystic fibrosis.

Standard
Essential Knowledge:
3A3: The chromosomal basis of inheritance provides an understanding of the pattern of passage (transmission) of genes from parent to offspring.

Materials
Every student needs: Activity worksheet

Procedure
1. Read the worksheet.
2. Answer the Analysis and Application questions.
**Genetic Counseling**

Genetic counseling is a career field in which counselors advise patients about the probabilities of developing or passing on diseases to the next generation. They aid couples with their options for family planning. Genetic counselors work with genetics, family trees, and probability on a daily basis.

One of the probability rules genetic counselors often work with is the multiplication rule. This rule is used when determining the probability that two separate events will occur together. For example, if a green marble, a blue marble, a yellow marble, and a red marble are all put into a bag together, the probability that you will pull out the red marble is \( \frac{1}{4} \). The probability that you will pull out a yellow marble is \( \frac{1}{4} \). The probability that you will pull out a red marble AND a yellow marble together is \( \frac{1}{16} \) (\( \frac{1}{4} \times \frac{1}{4} \)).

In genetics problems, you can use the multiplication rule instead of Punnett squares. For example, what is the probability that a pea plant homozygous recessive for the wrinkled gene (rr) crossed with a pea plant heterozygous for the wrinkled gene (Rr) will produce a wrinkled offspring?

**Method #1: Punnett square**

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Probability that offspring will be wrinkled (rr): \( \frac{1}{2} = 50\% \)

**Method #2: Multiplication rule**

In order to produce a wrinkled pea plant, both parents must provide a recessive gene, r.

The probability that pea plant #1 (rr) will provide r: 1
The probability that pea plant #2 (Rr) will provide r: \( \frac{1}{2} \)

The probability that pea plant offspring will be rr: \( 1 \times \frac{1}{2} = \frac{1}{2} = 50\% \)
Huntington’s Disease

Huntington’s disease is a disease that affects muscle coordination. Although the disease can present at any age, the most common age range is 35-44. Symptoms include coordination issues, jerky movements, and cognitive and behavioral problems.

Huntington’s disease is caused by mutations in the Huntington gene (htt). The gene includes several repeated sections, which vary in length from person to person. When the length of the repeated sections reaches a threshold, the Huntington protein that is transcribed is different from the normal Huntington protein. The mutant protein is the cause of the disease. Huntington’s disease is autosomal dominant, meaning that the disease occurs from either a homozygous dominant gene or a heterozygous gene.

Analysis: Huntington’s disease
A couple comes to your office for counseling. While both the male and female are healthy, the male has a family history of Huntington’s disease. He gives you a family tree, showing that his great great grandfather, grandmother, father and great uncle all suffered from the disease. Using this information and the knowledge that Huntington’s disease is autosomal dominant answer the following questions.
1. What is the probability that their child will have the disease?

2. Is his great great grandfather homozygous or heterozygous for the gene?
Cystic Fibrosis

Cystic Fibrosis is a disease that affects several organs in the body, particularly the lungs, pancreas, liver and the intestines. The disease is caused by a mutation in the CFTR gene. This gene is responsible for the protein that regulates mucus secretions. It is an autosomal recessive disease, meaning that a person must be homozygous recessive for the gene in order to get the disease.

Analysis: Cystic Fibrosis

A man comes in for genetic counseling concerning cystic fibrosis. While he does not have the disease, it runs in his family. He is currently in a serious relationship with a woman and, before he proposes to her, he wants to know whether or not he is a carrier for the disease. He feels that this information has the potential to affect the relationship due to the fact that the woman has cystic fibrosis. Upon your recommendation, he agrees to undergo genetic testing for the disease. The results of the testing show that he is indeed a carrier for cystic fibrosis.

1. If the man and the woman with cystic fibrosis have a child, what is the probability that their child will NOT have cystic fibrosis?

2. If the man and woman have two children, what is the probability that both children will not have the disease?
1. In a plant, axial flowers (A) are dominant to terminal flowers (a), tall flowers (L) are dominant to dwarf flowers (l), and round seeds (R) are dominant to wrinkled seeds (r). From the following cross, what is the probability that the listed offspring will result?

AaLlr x AALlr

a.) AaLlr

b.) AALlRr

c.) AaLlr

2. Tay-Sachs disease is an autosomal recessive disease in which lipids build up in the brain, causing seizures, blindness, and mental issues. Since it is a recessive disease, people who are heterozygous do not get sick. On a molecular level, however, heterozygous individuals have a slight buildup of lipids in the brain, just not enough to cause damage. On the molecular level, heterozygous individuals have a phenotype that is intermediate between the homozygous dominant and homozygous recessive. What type of inheritance pattern is this?

3. Genetic testing is possible for several different genetic diseases. This brings up some ethical issues. Is it ethical for a person to find out information from their genes? At what age is it appropriate for a person to undergo testing? These types of questions usually do not have a strictly right or wrong answer. It often depends on the specific situation.

a.) If your father is diagnosed with Alzheimer’s disease, would you want to get tested for the disease? Explain your answer.

b.) What are the pros and cons of knowing you will get a disease?
Answer Key

Analysis

Huntington’s Disease

1. 0% chance that their children will have the disease. Since neither of them have the disease, they are both recessive for the gene.

2. Heterozygous

Cystic Fibrosis

1. 50% chance that their child will not have the disease.

2. $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4} = 25\%$ chance

Application

1. a. $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{4} = 1/16$
   b. $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{4} = 1/16$
   c. $\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2} = 1/8$

2. Incomplete dominance

3. a. Answers will vary.
   b. Answers will vary. Examples:
      Pro: You know you will get it so you can prepare accordingly.
      Con: You may end up living your life in constant fear of when the disease will strike.