CAREER CONNECTION - Genetic Counselor

Given the intricate orchestration of gene expression, cell migration, and cell differentiation during prenatal development, it is amazing that the vast majority of newborns are healthy and free of major birth defects. When a woman over 35 is pregnant or intends to become pregnant, or her partner is over 55, or if there is a family history of a genetic disorder, she and her partner may want to speak to a genetic counselor to discuss the likelihood that their child may be affected by a genetic or chromosomal disorder. A genetic counselor can interpret a couple's family history and estimate the risks to their future offspring.

For many genetic diseases, a DNA test can determine whether a person is a carrier. For instance, carrier status for Fragile X, an X-linked disorder associated with mental retardation, or for cystic fibrosis can be determined with a simple blood draw to obtain DNA for testing. A genetic counselor can educate a couple about the implications of such a test and help them decide whether to undergo testing. For chromosomal disorders, the available testing options include a blood test, amniocentesis (in which amniotic fluid is tested), and chorionic villus sampling (in which tissue from the placenta is tested). Each of these has advantages and drawbacks. A genetic counselor can also help a couple cope with the news that either one or both partners are a carrier of a genetic illness, or that their unborn child has been diagnosed with a chromosomal disorder or other birth defects.

To become a genetic counselor, one needs to complete a 4-year undergraduate program and then obtain a Master of Science in Genetic Counseling from an accredited university. Board certification is attained after passing examinations by the American Board of Genetic Counseling. Genetic counselors are essential professionals in many branches of medicine, but there is a particular demand for preconception and prenatal genetic counselors.

CONCEPTS IN ACTION- Visit the National Society of Genetic Counselors website for more information about genetic counselors. Visit the American Board of Genetic Counselors, Inc. website for more information about genetic counselors.

Section Summary

Human genetics focuses on identifying different alleles and understanding how they express themselves. Medical researchers are especially interested in the identification of inheritance patterns for genetic disorders, which provides the means to estimate the risk that a given couple's offspring will inherit a genetic disease or disorder. Patterns of inheritance in humans include autosomal dominance, autosomal recessive, X-linked dominance, and X-linked recessive.

Exercises

1. Hemophilia is a X-linked recessive disorder. A woman who has hemophilia and an unaffected (healthy) male have a son; what is the probability that their son will have hemophilia?
    a. 25%
    b. 50%
    c. 75%
    d. 100%
2. Cystic fibrosis is an autosomal recessive disorder. Two heterozygous carriers have an offspring; what is the probability that they will have an offspring with cystic fibrosis?
    a. 25%
    b. 50%
    c. 75%
    d. 100%
3. Marfan syndrome is inherited in an autosomal dominant pattern. Which of the following is true?
    a. Female offspring are more likely to be carriers of the disease.
    b. Male offspring are more likely to inherit the disease.
    c. An affected offspring must have at least one affected parent.
    d. Female offspring are more likely to inherit the disease.
4. Can a male be a carrier of red-green color blindness?

Answers

1. (d)
2. (a)
3. (c)
4. No, males can only express color blindness and cannot carry it because an individual needs two X chromosomes to be a carrier.

Glossary

allosomes: chromosome pair twenty-three in humans and plays a role in a person's sex

autosomal dominant inheritance: pattern of dominant inheritance that corresponds to a gene on one of the 22 autosomal chromosomes

autosomal recessive inheritance: pattern of recessive inheritance that corresponds to a gene on one of the 22 autosomal chromosomes

autosome: chromosome pairs one through twenty-two and does not determine a person's sex

carriers: a heterozygous individual who does not display symptoms of a recessive genetic disorder but can transmit the disorder to his or her offspring

X (sex)-linked: pattern of inheritance in which an allele is carried on the X chromosome of the 23rd pair

X-linked dominant inheritance: pattern of dominant inheritance that corresponds to a gene on the X chromosome of the 23rd pair

X-linked recessive inheritance: pattern of recessive inheritance that corresponds to a gene on the X chromosome of the 23rd pair