Case Studies – Genetics

A baby girl is admitted to the Neonatal Intensive Care Unit shortly after birth. It was noted at delivery that the child had widely spaced nipples and webbing of the neck. Further evaluation noted a heart murmur, edematous feet, and difficulty palpating or locating the pulse in the groin. An echocardiogram is ordered, and the child is diagnosed with coarctation of the aorta (a narrowing of the aorta that causes the left ventricle of the heart to pump against higher resistance, which can lead to heart failure). As a result of the nature of the symptoms, a geneticist is consulted and the newborn is diagnosed with Turner syndrome. The family meets with the physicians to discuss the ramifications of this diagnosis.

The physicians explain that the child has an abnormal number of chromosomes. This particular syndrome is designated as

1. 45X karyotype
2. 47XXY karyotype
3. Trisomy 21
4. 5p minus

The physicians explain that Turner syndrome is caused by:

1. Aneuploidy
2. Consanguinity
3. Pseudoautosomal region
4. X-linked recessive genes

Responding to an advertisement in a college newsletter, one set of adopted twins (one male twin and one female twin) arrives at a research center in a local hospital. The study involves a generalized health assessment, some basic medical tests, and a psychologic examination. Participants will learn of any predispositions they may have toward a variety of disease processes. The twins have brought a vague family history from their natural parents and their adoptive parents. Twin studies and adoption studies are often used to estimate the relative influence of genes and environmental factors. This particular set of twins is dizygotic, which means they are the result of double ovulation, followed by the fertilization of each egg by a different sperm. Monozygotic twins are an example of natural clones; that is, the developing embryo divides to form two separate but identical embryos. Dizygotic twins are an important because although they may share environmental influences, their genetics are as great as those among siblings.

The female twin has a history of a cleft palate, whereas the male twin does not. This trait would be considered to be:

1. Discordant
2. Concordant

3. Multifactorial
4. Quantitative

Multifactorial inheritance is influenced by which factor?

1. The recurrence risk is higher if the proband is of the less commonly affected sex.
2. The expression of the disease in the mother dictates the recurrence risk.
3. Those with a lower threshold have a lower incidence rate.
4. The highest risk generally falls to male relatives of a female proband.

A 34-year-old woman was recently diagnosed with colon cancer. Although her father has been previously treated for colon cancer, her identical twin sister has not. She meets with her oncologist who begins to explain the process of inherited diseases. Genetics has traditionally focused on the ways in which alteration of DNA sequences may lead to disease. At times, diseases are the result of chemical modification of the DNA sequences that alter the expression of genes, or epigenetics.

Although three major types of epigenetic modifications have been identified, specific environmental or nongenetic factors can cause epigenetic modifications. Identical twins have essentially the same DNA sequences; however, aging and significant lifestyle differences cause changes in:

1. Genomic imprinting
2. Histone modification
3. Chromatin structure
4. DNA methylation

Unlike DNA sequence mutations, epigenetic modifications can be reversed through:
1. Dietary changes
2. Pharmaceutical intervention
3. Lifestyle choices
4. Reversal of the aging process