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Chapter 2. Genomic Assessment: Interpreting Findings and Formulating Differential Diagnosis

MULTIPLE CHOICE

1. The first step in the genomic assessment of a patient is obtaining information regarding:
   A. Family history
   B. Environmental exposures
   C. Lifestyle and behaviors
   D. Current medications

   ANS: A
   A critical first step in any health assessment is collecting and interpreting the family health history (FHH). FHH is key because it reflects shared genetic susceptibilities, shared environment, and common behaviors (Yoon, Scheuner, & Khoury, 2003).

   PTS: 1

2. An affected individual who manifests symptoms of a particular condition through whom a family with a genetic disorder is ascertained is called a(n):
   A. Consultand
   B. Consulband
   C. Index patient
   D. Proband

   ANS: D
   A proband is a family member who manifests symptoms of a particular condition and/or brings the family to medical attention (Bennett, 2010).

   PTS: 1

3. An autosomal dominant (AD) disorder involves the:
   A. X chromosome
   B. Y chromosome
   C. Mitochondrial DNA
   D. Non-sex chromosomes

   ANS: D
   AD inheritance is a result of a gene mutation in one of the 22 autosomes.

   PTS: 1

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4. To illustrate a union between two second cousin family members in a pedigree:
   A. Arrows are drawn pointing to the male and female
   B. Brackets are drawn around the male and female
   C. Double horizontal lines are drawn between the male and female
   D. Circles are drawn around the male and female

   ANS: C
   A consanguineous family is related by descent from a common ancestry and is defined as a "union between two individuals who are related as second cousins or closer" (Hamamy, 2012). Consanguinity, if present in the family history, is portrayed using two horizontal lines to establish the relationship between the male and female partners.

   PTS: 1

5. To illustrate two family members in an adoptive relationship in a pedigree:
   A. Arrows are drawn pointing to the male and female
   B. Brackets are drawn around the male and female
   C. Double horizontal lines are drawn between the male and female
   D. Circles are drawn around the male and female

   ANS: B
   For adopted members of the family, use brackets as the appropriate standardized pedigree symbol.

   PTS: 1

6. When analyzing the pedigree for autosomal dominant (AD) disorders, it is common to see:
   A. Several generations of affected members
   B. Many consanguineous relationships
   C. More members of the maternal lineage affected than the paternal
   D. More members of the paternal lineage affected than the maternal

   ANS: A
   Pedigrees associated with AD disorders typically reveal multiple affected family members with the disease or syndrome. When analyzing the pedigree for AD disorders or syndromes, it is common to see a vertical pattern denoting several generations of affected members.

   PTS: 1

7. In autosomal recessive (AR) disorders, individuals need:
   A. Only one mutated gene on the sex chromosomes to acquire the disease

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B. Only one mutated gene to acquire the disease
C. Two mutated genes to acquire the disease
D. Two mutated genes to become carriers

ANS: C
In AR disorders, the offspring inherits the condition by receiving one copy of the gene mutation from each of the parents. AR disorders must be inherited through both parents (Nussbaum et al., 2016). Individuals who have an AR disorder have two mutated genes, one on each locus of the chromosome. Parents of an affected person are called carriers because each carries one copy of the mutation on one chromosome and a normal gene on the other chromosome. Carriers typically are not affected by the disease.

PTS: 1

8. In autosomal recessive (AR) disorders, carriers have:
A. Two mutated genes: one from each parent that causes disease
B. A mutation on a sex chromosome that causes a disease
C. A single gene mutation that causes the disease
D. One copy of a gene mutation but not the disease

ANS: D
Individuals who have an AR disorder have two mutated genes, one on each allele of the chromosome. Parents of an affected person are called carriers because each parent carries one copy of the mutation on one chromosome and a normal gene on the other chromosome. Carriers typically are not affected by the disease. In pedigrees with an AR inheritance pattern, males and females will be equally affected because the gene mutation is on an autosome.

PTS: 1

9. With an autosomal recessive (AR) disorder, it is important that parents understand that if they both carry a mutation, the following are the risks to each of their offspring (each pregnancy):
A. 50% chance that offspring will carry the disease
B. 10% chance that offspring will be affected by the disease
C. 25% chance that offspring will carry the disease
D. 10% chance that offspring will be disease free

ANS: A
It is important that parents understand that if they both carry a mutation, the risk to each of their offspring (each pregnancy) is an independent event: 25% disease free, 25% affected, and 50% carrier.

PTS: 1

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10. A woman with an X-linked dominant disorder will:
    A. Not be affected by the disorder herself
    B. Transmit the disorder to 50% of her offspring (male or female)
    C. Not transmit the disorder to her daughters
    D. Transmit the disorder to only her daughters

    ANS: B
    Everyone born with an X-linked dominant disorder will be affected by the disease. Transmission of the disorder to the next generation varies by gender, however. A woman will transmit the mutation to 50% of all her offspring (male or female).

    PTS: 1

11. In creating your female patient's pedigree, you note that she and both of her sisters were affected by the same genetic disorder. Although neither of her parents had indications of the disorder, her paternal grandmother and her paternal grandmother's two sisters were affected by the same condition. This pattern suggests:
    A. Autosomal dominant disorder
    B. Chromosomal disorder
    C. Mitochondrial DNA disorder
    D. X-linked dominant disorder

    ANS: D
    A man with an X-linked dominant disorder will transmit the mutation to 100% of his daughters (they receive his X chromosome) and none of his sons (they receive his Y chromosome). The pedigree of a family with an X-linked dominant disorder would reveal all the daughters and none of the sons affected by the disorder if the father has an X-linked disorder.

    PTS: 1

12. A woman affected with an X-linked recessive disorder:
    A. Has one X chromosome affected by the mutation
    B. Will transmit the disorder to all of her children
    C. Will transmit the disorder to all of her sons
    D. Will not transmit the mutation to any of her daughters

    ANS: C
    An X-linked recessive disorder means that in a woman, both X chromosomes must have the mutation if she is to be affected. Because males have only one copy of the X chromosome, they will be affected if their X chromosome carries the mutation.

    PTS: 1

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13. Which of the following are found in an individual with aneuploidy?
    A. An abnormal number of chromosomes
    B. An X-linked disorder
    C. Select cells containing abnormal-appearing chromosomes
    D. An autosomal recessive disorder

    ANS: A
    An individual with an abnormal number of chromosomes has a condition called aneuploidy, which is frequently associated with mental problems or physical problems or both (Jorde et al., 2015; Nussbaum et al., 2016).

    PTS: 1

14. The pedigree of a family with a mitochondrial DNA disorder is unique in that:
    A. None of the female offspring will have the disease
    B. All offspring from an affected female will have the disease
    C. None of the offspring of an affected female will have the disease
    D. All offspring from an affected male will have the disease

    ANS: B
    Mitochondrial DNA is inherited from the ovum and, therefore, from the mother. The pedigree of a family with a mitochondrial DNA disorder is unique in that all offspring (regardless of gender) of an affected female will have the disease, and none of the offspring from an affected male will have the disease.

    PTS: 1

15. Which population is at highest risk for the occurrence of aneuploidy in offspring?
    A. Mothers younger than age 18
    B. Fathers younger than age 18
    C. Mothers older than age 35
    D. Fathers older than age 35

    ANS: C
    Some individuals or couples have unique identifiable risks that should be discussed prior to conception whenever possible. For example, women who will be 35 years of age or older at delivery (advanced maternal age) are at increased risk for aneuploidy.

    PTS: 1

16. Approximately what percentage of cancers are due to a single-gene mutation?
    A. 50% to 70%
    B. 30% to 40%

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C. 20% to 25%
D. 5% to 10%

ANS: D
The majority of cancers are sporadic or multifactorial due to a combination of genetic and environmental factors; however, approximately 5% to 10% of all cancers are due to a single-gene mutation (Garber & Offit, 2005).

PTS: 1

17. According to the Genetic Information Nondiscrimination Act (GINA):
A. Nurse practitioners (NPs) should keep all genetic information of patients confidential.
B. NPs must obtain informed consent prior to genetic testing of all patients.
C. Employers cannot inquire about an employee's genetic information.
D. All of the above

ANS: D
On May 21, 2008, President George W. Bush signed the Genetic Information Nondiscrimination Act (GINA) to protect Americans against discrimination based on their genetic information when it comes to health insurance and employment, paving the way for patient-personalized genetic medicine without fear of discrimination (National Human Genome Research Institute, 2012).

PTS: 1

18. The leading causes of death in the United States are due to:
A. Multifactorial inheritance
B. Single gene mutations
C. X-linked disorders
D. Aneuploidy

ANS: A
Most disease-causing conditions are not due to a single-gene disorder, but to multifactorial inheritance, a result of genomics and environmental or behavioral influences. In fact, the leading causes of mortality in the United States—heart disease, cerebrovascular disease, diabetes, and cancer—are all multifactorial. Most congenital malformation, hypertension, arthritis, asthma, obesity, epilepsy, Alzheimer's disease, and mental health disorders are also multifactorial.

PTS: 1

19. Which of the following would be considered a red flag that requires more investigation in a patient assessment?

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A. Colon cancer in family member at age 70
B. Breast cancer in family member at age 75
C. Myocardial infarction in family member at age 35
D. All of the above

ANS: C
Early onset cancer syndromes, heart disease, or dementia are red flags that warrant further investigation regarding hereditary disorders.

PTS: 1

20. When patients express variable forms of the same hereditary disorder this is due to:
A. Penetrance
B. Aneuploidy
C. De novo mutation
D. Sporadic inheritance

ANS: A
Some disorders have a range of expression from mild to severe. This variability is referred to as the penetrance of genetic disease. For example, patients with neurofibromatosis (NF1), an AD disorder of the nervous system, may manifest with many forms of the disease. For instance, some patients with NF1 may have mild symptoms, like café au lait spots or freckling on the axillary or skin, while others may have life-threatening spinal cord tumors or malignancy (Jorde et al., 2015; Nussbaum et al., 2016).

PTS: 1

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