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Genetics

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1. A permanent change in the normal sequence of DNA that leads to a functional alteration in the protein is defined as a(n)
   a. Allele.
   b. Gene.
   c. Mutation.
   d. Polymorphism.
2. Hereditary cancer syndromes most commonly are transmitted by
   a. Autosomal dominant transmission with complete penetrance.
   b. Autosomal dominant transmission with incomplete penetrance.
   c. Autosomal recessive transmission with complete penetrance.
   d. Autosomal recessive transmission with incomplete penetrance.
3. In autosomal dominant hereditary cancer syndromes, a family member of a known mutation carrier has a 12.5% chance of inheriting a mutation. This relative is a
   a. First-degree relative.
   b. Second-degree relative.
   c. Third-degree relative.
   d. Fourth-degree relative.
4. In a family without a known mutation, first testing an individual who has been diagnosed with cancer is best because
   a. A negative result would eliminate the risk of genetic transmission.
   b. If a mutation exists in a family, affected individuals are more likely to be carriers than unaffected members.
   c. Individuals who have had cancer will not be worried about the results of genetic testing.
   d. Testing is less expensive when performed in an individual already diagnosed with cancer.
5. The impact of the results of genetic testing on family dynamics and relationships should be considered and discussed prior to testing because
   a. The family must decide together whether testing is appropriate.
   b. The mutation status of one family member may reveal the mutation status of others.
   c. Testing an affected individual may be necessary to obtain results for the rest of the family.
   d. All members will need genetic testing once a mutation has been identified in a relative.
6. Post-test counseling provides an opportunity to
   a. Assess psychological reactions to the testing.
   b. Take a detailed family history.
   c. Assess an individual's prior probability of carrying an altered gene.
   d. Disclose results to third parties.
7. A 67-year-old woman develops breast cancer. Her family has no other history of breast cancer. The most likely cause of her breast cancer is
   a. A germline mutation.
   b. A somatic mutation.
   c. Ingestion of at least three alcoholic beverages a day.
   d. Menarche before age 12.
8. During her three-month follow-up visit, a woman who tested positive for a mutation in *BRCA2* states that she has not informed her two sisters or three adult daughters of the test results and their option of genetic counseling and testing. The best way to prevent this problem would have been to
   a. Provide detailed pretest education and counseling.
   b. Have her sign a consent that states that all relatives will be informed.
   c. Refuse to offer testing to this person because of potential family problems.
   d. Inform the patient's physician so he or she can disclose the results to the sisters and daughters.
9. A 27-year-old woman presents for disclosure of her genetic testing results. The laboratory reports a missense mutation of unknown significance in *BRCA2*. The patient should be educated that
   a. The mutation does not really increase risk for developing breast and/or ovarian cancer.
   b. An error occurred in analysis and the specimen will need to be resubmitted.
   c. The mutation does increase the risk for developing breast and/or ovarian cancer.
   d. The risk for cancer is uncertain.
10. When completing a family history to assess for inherited susceptibility risk, for how many generations should the information usually be obtained?
    a. One
    b. Two
    c. Three
    d. Four
11. A 46-year-old woman without breast cancer requests information about her risk for developing the disease. Her paternal grandmother was diagnosed with breast cancer at age 45. Which model would most accurately calculate her risk for developing breast cancer?
    a. The Claus model
    b. The Gail model
    c. The Frank model
    d. The BRCAPRO model
12. A woman has blood sent for *BRCA1* and *BRCA2* genetic tests. The results become available, but she does not keep

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