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## Genetic Risk Factors

- Which are true statements about chromosomes in the human body? (*Select all that apply.*)
  - The autosomes represent the 22 chromosome pairs that do not determine gender.
  - Each parent contributes half of each pair to the offspring, including a sex chromosome.
  - Hereditary cancers are the primary reason for a patient to try to understand genetics.
  - Chromosomes have genes located at specific positions; each gene contains genetic information.
  - Three growth codons are associated with the growth of amino acid chains in cancer.
- Which is the true statement about DNA?
  - DNA contains two types of bases, adenine (A) and thymine (T).
  - The base pairs are complementary on the double strand.
  - Messenger DNA contains the information about the order of amino acids in a protein.
  - There are two nucleotide chains running in the same direction, held together by hydrogen bonds.
- Which statements are true about ribonucleic acid (RNA)? (*Select all that apply.*)
  - RNA is a single chain that represents a complementary copy of a strand of DNA.
  - Transcription refers to the process of making proteins from RNA.
  - Translation refers to the process of making RNA from DNA.
  - The sequences of the amino acid chains determine the function of the proteins.
  - In RNA, bases are the same as DNA except the base uracil (U) replaces thymine (T).
- Messenger RNA (mRNA)
  - is created through a process called translation.
  - is a template created by transcription.
  - contains information about the order of the proteins in amino acids.
  - strands with nucleotide changes frequently cause amino acid changes.
- Messenger ribonucleic acid (mRNA) codons
  - are sets of four nucleotides that act as a template for protein synthesis, providing structure for genetic material.
  - consist of four common bases that correspond with matching bases on a tRNA anticodon attached to a specific amino acid.
  - may function as “stop” codons. Examples include transfer RNA (tRNA), ribosomal RNA (rRNA), and several small silencing RNAs.
  - are each coded for a specific amino acid in the transcription process for DNA to RNA.
- Which statement accurately defines a gene?
  - A threadlike structure that contains genetic information on exons and introns
  - An individual unit of hereditary information that codes for a specific protein
  - A specific sequence of amino acids that have a predetermined function
  - Two nucleotide chains coiled around one another to form a double helix
- Understanding the role of the basic mechanisms of carcinogenesis, mutations and heredity in cancer builds on knowing that
  - somatic mutations are usually considered acquired in body cells after a relatively brief period of carcinogenesis.
  - inherited mutations play a stronger role in cancer development than genetic mutations and instability.
  - acquired genetic mutations are associated with exogenous and indigenous factors
  - germline reproductive cells are often involved in someone who develops cancer without a predisposition to cancer.
- Which type of mutation results in the absence of RNA transcribed from a gene copy?
  - Frameshift mutations
  - Splicing mutations
  - Nonsense mutations
  - RNA-negative mutations

9. In someone with a genetic predisposition to cancer, a mutation
- was inherited in the germline, reproductive cells.
  - occurred sometime during the life of the individual.
  - resulted from a nonsense change in an amino acid signal.
  - occurred through polymorphisms involving DNA sequence of a gene.
10. Chromosomal abnormalities include (*Select all that apply.*)
- translocation, which is the loss of a segment of both copies of a chromosome.
  - aneuploidy, which is an abnormal number of chromosomes.
  - loss of heterozygosity in which segments of one chromosome break off.
  - microsatellite instability segments, repetitive pieces of DNA scattered throughout the genome in noncoding regions.
  - polymorphisms leading to segments of one chromosome breaking off and attaching to other chromosomes.
11. Many of the genes responsible for familial cancer syndromes are
- proto-oncogenes.
  - oncogenes.
  - tumor suppressor genes.
  - mismatch repair genes.
12. What is the difference between proto-oncogenes and tumor suppressor genes? (*Select all that apply.*)
- Proto-oncogenes function as regulators of cell growth.
  - Proto-oncogenes have a role in DNA repair.
  - Proto-oncogenes are normal genes essential for normal cell growth.
  - Tumor suppressor genes convert to oncogene activation and can cause uncontrolled growth.
  - Tumor suppressor genes function as regulators of cell growth; some have a role in cell cycle.
  - Tumor suppressor genes are a type of repair genes associated with microsatellite instability in some syndromes.
13. What is a change in a gene's DNA pattern called?
- Tumor suppressor gene.
  - Proto-oncogene.
  - Mismatch repair gene.
  - Mutation
14. If a parent has a hereditary cancer that is autosomal dominant inheritance, what is the percent chance of transmitting that gene mutation to a child?
- 75%
  - 50%
  - 25%
  - 10%
15. A significant safety risk during genetic testing is ⚠
- being certain the family has a hereditary cancer.
  - confirming the presence of a risk that cannot be modified.
  - mislabeling of the specimens prior to sending to the laboratory.
  - failing to remember the volume of blood to collect.
16. In genetic testing for inherited cancer risk, informed consent is very important. One critical component of informed consent is
- confirmation of the family history for cancer.
  - overview of the risks, benefits, and limitations of predisposition genetic testing.
  - recommending an individualized cancer risk management plan before testing.
  - completion of a full history and physical examination to rule out any suspicion of cancer.
17. The primary role of the nurse in predisposition genetic testing includes
- establishing a cancer risk management plan.
  - determining which members of a family should be tested for a genetic alteration.
  - facilitating informed decision making without being directive.
  - selecting the laboratory to perform the test.
18. What role does apoptosis play in cancer?
- It is the activation of a program causing abnormal mutations in genes.
  - As cells age, apoptosis is repressed and gradually lost at the cell level.
  - In cancer, apoptosis is reactivated, triggering cell death.
  - Malfunctions result in uncontrolled cell proliferation of malignant cells.
19. What role does telomerase play in cancer?
- It is reactivated in cancer, which keeps telomeres intact, facilitating cell immortalization.
  - It plays a role in cellular aging through the telomeres, which are at the ends of the chromosomes.
  - Malfunction in telomerase leads to uncontrolled cell proliferation of malignant or damaged cells.
  - It determines the sensitivity of the malignant cells to the chemotherapy and radiation therapy.

20. Which statement best describes penetrance of a gene?
- It is the record of an individual's ancestral history, showing inheritance patterns for a given trait.
  - It is whether an individual of a given genotype expresses the corresponding phenotype.
  - It includes the characteristics (appearance and activity) of an organism that result from the interaction between that organism's genotype and the environment.
  - It is the degree to which a single individual with a specific genotype will exhibit a specific trait.
21. Which of the following items represents one application of pharmacogenetics in clinical practice?
- Drugs intended to produce a genetic change or altered protein product
  - Introduction of a functioning gene into cells to replace missing function
  - Introduction of a functioning gene into the egg or sperm to prevent transmission of a gene mutation
  - Measurement of the structure, composition, and function of proteins that are made from genes
22. Key technical characteristics of predisposition genetic testing and tumor profiling include
- direct sequencing that detects one single specific mutation that involves a short sequence of DNA.
  - Genome-wide association studies that detect small mutations to determine association with disease
  - Single-strand confirmation polymorphism analysis designed to offer faster and cheaper ways to obtain genetic data
  - Microarray techniques that detect large rearrangements, deletions, and duplications
23. Considerations for recommending a laboratory for genetic testing include: (*Select all that apply.*)
- Clinical Laboratory Improvement Act (CLIA)–approved laboratory
  - DNA Laboratory Proficiency Certification by genetic testing review association
  - Laboratory Director is certified by the American Board of Medical Genetics
  - Institution laboratory with genetic research and oversight to meet biosafety standards
  - Evidence of meeting NIH Guidelines for Research Involving Recombinant DNA Molecules
24. The use of genetic markers for diagnosis of cancer includes
- cytogenetics, which uses a personalized approach to diagnose cancer and suggest the best treatment regimen for a person's cancer.
  - cytogenetics, which focuses on the structure, function, and abnormalities of the chromosomes to diagnose both solid and hematologic malignancies.
  - karyotyping, which focuses on techniques such as DNA microarray or serial analysis of gene expression.
  - gene expression profiling to identify the genetic basis for differences in the metabolism of cancer cells.
25. Mutations in which gene have been correlated with an increased risk for both breast and ovarian cancer?
- p53
  - PTEN
  - BRCA1*
  - APC
26. Which of the following is a clinical feature of hereditary cancer?
- Older age of cancer onset
  - Telomerase
  - Multiple primary cancers in a single individual
  - The presence of metastasis at the time of diagnosis
27. What type of mutation adds or deletes one or more bases from the normal gene sequence?
- Frameshift
  - Missense
  - Splicing
  - Translocation
28. An individual who has known colon cancer susceptibility in his family has tested negative. His colon cancer risk
- cannot be established.
  - is at least equivalent to the general population.
  - is nil.
  - is still elevated.
29. Mutations in *MSH2* have been associated with an increased risk for which form of cancer?
- Sarcoma
  - Ovary
  - Lung
  - Thyroid

30. Pharmacogenomics identifies genetic differences that influence cancer treatment, including
- how the body absorbs, distributes, metabolizes, and excretes the drug, called pharmacodynamics.
  - the biochemical and physiological effects of drugs on the body, referred to as pharmacokinetics.
  - genetic variations such as the role of decision peptide driver (DPD) protein in the inactivation of active 5 FU for decreased toxicity.
  - genetic testing to identify the extent of disease to assist with determining the cancer diagnosis.
31. Pharmacogenomic testing is required for some cancer therapies, including
- trastuzumab, imatinib, and capecitabine.
  - cetuximab, gemcitabine and vemurafenib.
  - letrozole, fulvestrant, and tyrosine kinase inhibitors.
  - imatinib, busulfan in CML, and 5FU for colon cancers.
32. The son of a patient is happy to see the results of his genetic testing were negative. What is important for him to understand about his level of risk? (*Select all that apply.*)
- A negative test result in the presence of a known genetic mutation indicates that the client is within the general population risk of cancer associated with that branch of the family.
  - Because the results of the test are negative, his risk for cancer is minimal.
  - Without knowing the type of cancer his parent has, you cannot give accurate information about his risk level.
  - You do not know if his parent's cancer is the result of a genetic mutation. You need more information to help him interpret his results and assess his risk.
  - It is still important for him to have the surgery for removal of tissue at risk for the cancer.
33. What role does medical management play for individuals with a mutation in a cancer susceptibility gene? (*Select all that apply.*)
- Surveillance to detect disease early
  - Prophylactic surgery to remove as much of the tissue at risk as possible to reduce the risk of developing a specific cancer
  - Chemoprevention, which is the administration of medicine, vitamins, or other substances to reduce the risk of a cancer
  - Discourage the individual from having children, who might develop the inherited cancer
  - Promotion of healthy behaviors, including diet and exercise
34. Nurses can advocate for individuals with known cancer risk mutations by
- referring them to the NCI Physician Data Query Summaries on Genetics.
  - supporting (Health Insurance Portability and Accountability Act (HIPAA) guidelines for the use of genetic information.
  - discouraging them from sharing their information with others.
  - encouraging them to have their test repeated in 6 months to see if the results are consistent.