Huether and McCances Understanding Pathophysiology (Canadian) 2nd edition Power Kean Test Bank

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Chapter 02: Genes and Genetic Diseases
Power-Kean et al: Huether and McCance's Understanding Pathophysiology, Second Canadian Edition

MULTIPLE CHOICE

1. A nurse recalls that the basic components of DNA are:
   a. pentose sugars and four phosphate bases.
   b. a phosphate molecule, deoxyribose, and four nitrogenous bases.
   c. adenine, guanine, and purine.
   d. codons, oxygen, and cytosine.

   ANS: B
   The three basic components of DNA are deoxyribose; a phosphate molecule; and four types of nitrogenous, not phosphate, bases. DNA does not contain condone.

   DIF: Cognitive Level: Remember REF: Definitions: Composition and Structure of DNA
   TOP: Physiological Integrity

2. Which of the following mutations have the most significant effect on protein synthesis?
   a. Base pair substitutions
   b. Silent mutations
   c. Intron mutations
   d. Frameshift mutations

   ANS: D
   The frameshift mutation involves the insertion or deletion of one or more base pairs of the DNA molecule. This greatly alters the amino acid sequence, which affects protein synthesis. The base pair substitution is a type of mutation in which one base pair replaces another. Silent mutations do not change amino acids or protein synthesis. Intron mutations are part of RNA sequencing.

   DIF: Cognitive Level: Remember REF: Mutation TOP: Physiological Integrity

3. The base components of DNA are:
   a. A, G, C, and U.
   b. P, G, C, and T.
   c. A, G, C, and T.
   d. X, XX, XY, and YY.

   ANS: C
   The four base components of DNA are cytosine, thymine, adenine, and guanine, and they are commonly represented by their first letters (C, T, A, and G). There are no genetic components identified as P or U. The letters X, XX, XY, and YY are components of human chromosomes.

   DIF: Cognitive Level: Remember REF: DNA as the Genetic Code | Figure 2.2
   TOP: Physiological Integrity

4. A DNA strand has a region with the sequence ATCGGAT. Which of the following would be a complementary strand?

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a. CGATACGT
b. TAGCCTAG
c. TUGCCTUG
d. UAGCCUAG

ANS: B
The consistent pairing of adenine with thymine and of guanine with cytosine is known as complementary base pairing; thus, A complements to T, and C to G, and vice versa throughout the strand. A complements to T; thus, the first letter must be a T. U does not represent a complement in the sequence.

DIF: Cognitive Level: Understand REF: Replication of DNA | Figure 2.2
TOP: Physiological Integrity

5. A biologist is explaining how RNA directs the synthesis of protein. Which process is the biologist describing?
a. Termination
b. Transcription
c. Translocation
d. Translation

ANS: D
In translation, RNA directs the synthesis of a polypeptide, interacting with transfer RNA (tRNA), a cloverleaf-shaped strand of about 80 nucleotides. Termination does not involve the synthesis of protein. Transcription is the process by which DNA specifies a sequence of messenger RNA (mRNA). Translocation is the interchange of genetic material between nonhomologous chromosomes.

DIF: Cognitive Level: Understand REF: Translation TOP: Physiological Integrity

6. What is the result of homologous chromosomes failing to separate during meiosis?
a. Neurofibromatosis
b. Aneuploidy
c. Polyploidy
d. Conjoined twins

ANS: B
Nondisjunction is an error in which homologous chromosomes or sister chromatids fail to separate normally during meiosis or mitosis. The result is aneuploidy. Neurofibromatosis is not due to chromosome failure during meiosis. Polyploidy occurs when a euploid cell has more than the diploid number of chromosomes. Conjoined twins are not due to chromosome failure during meiosis.

DIF: Cognitive Level: Remember REF: Aneuploidy TOP: Physiological Integrity

7. A cell that does not contain a multiple of 23 chromosomes is called a _____ cell.
a. diploid
b. euploid
c. polyploid
d. haploid

ANS: C

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A polyploid cell is one in which a euploid cell has more than 23 pairs of chromosomes. A diploid cell is when the somatic cell nucleus has 46 chromosomes in 23 pairs. A euploid cell is a cell with multiples of the normal number of chromosomes. A haploid cell has only one member of each chromosome pair, for a total of 23 chromosomes.

DIF: Cognitive Level: Understand
REF: Chromosome Aberrations and Associated Diseases: Polyploidy
TOP: Physiological Integrity

8. A 20-year-old pregnant individual gives birth to a stillborn child. Autopsy reveals that the fetus has 92 chromosomes. What term may be on the autopsy report to describe this condition?
   a. Biploidy
   b. Triploidy
   c. Tetraploidy
   d. Aneuploidy

   ANS: C
   Tetraploidy is a condition in which euploid cells have 92 chromosomes. Biploidy is a euploid cell with two times more chromosomes, or 46. Triploidy is a zygote that has three copies of each chromosome, rather than the usual two. Aneuploidy is when an aneuploid cell does not contain a multiple of 23 chromosomes.

   DIF: Cognitive Level: Analyze
   REF: Chromosome Aberrations and Associated Diseases: Polyploidy
   TOP: Physiological Integrity

9. The condition in which an extra portion of a chromosome is present in each cell is called:
   a. reciprocal translocation.
   b. partial trisomy.
   c. inversion.
   d. Down syndrome.

   ANS: B
   Partial trisomy is a condition in which only an extra portion of a chromosome is present in each cell. A reciprocal translocation occurs when breaks take place in two different chromosomes and the material is exchanged. An inversion occurs when two breaks take place on a chromosome, followed by the reinsertion of the missing fragment at its original site, but in inverted order. Down syndrome is an aneuploidy of the twenty-first chromosome.

   DIF: Cognitive Level: Understand
   REF: Chromosome Aberrations and Associated Diseases: Autosomal aneuploidy
   TOP: Physiological Integrity

10. After a geneticist talks to a patient about being a chromosomal mosaic, the patient asks the nurse what that means. What should the nurse say?
    a. “You are only be a carrier of the genetic disease.”
    b. “You have a mild form of the genetic disease.”
    c. “You have two genetic diseases.”
    d. “The expression of your DNA has been altered, causing a genetic disease.”

    ANS: B

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A chromosomal mosaic means the body has two or more different cell lines, each of which has a different karyotype; thus, the person is a carrier and has a mild form of the disease. People with mosaicism have two different lines but not two different diseases. Alterations in DNA expression is due to epigenetic factors, and not mosaicism.

DIF: Cognitive Level: Understand REF: Table 2.1 TOP: Physiological Integrity

11. What is the most common cause of Down syndrome?
    a. Paternal nondisjunction
    b. Maternal translocations
    c. Maternal nondisjunction
    d. Paternal translocations

    ANS: C
    The most common cause of Down syndrome is maternal, not paternal, nondisjunction. Translocation is not a cause of this syndrome.

    DIF: Cognitive Level: Understand REF: Table 2.1 TOP: Physiological Integrity

12. What is a risk factor for Down syndrome?
    a. Fetal exposure to mutagens in the uterus
    b. Increased paternal age
    c. Family history of Down syndrome
    d. Pregnancy in childbearing parents over age 35

    ANS: D
    The primary risk for Down syndrome is pregnancy in childbearing parents over 35. Down syndrome is a trisomy and not due to fetal exposure or paternal age. Down syndrome is a chromosomal abnormality and is not related to family history.

    DIF: Cognitive Level: Understand
    REF: Chromosome Aberrations and Associated Diseases: Autosomal aneuploidy
    TOP: Physiological Integrity

13. A 13-year-old patient has a karyotype that reveals an absent homologous X chromosome with only a single X chromosome present. What medical diagnosis will the nurse observe on the chart?
    a. Down syndrome
    b. cri du chat syndrome
    c. Turner's syndrome
    d. Fragile X syndrome

    ANS: C
    A condition with the presence of a single X chromosome and no homologous X or Y chromosome, so the individual has a total of 45 chromosomes, is known as Turner's syndrome. Down syndrome is a change in one arm of a chromosome. cri du chat syndrome is due to a chromosome deletion. Fragile X syndrome is due to a break or a gap in a chromosome.

    DIF: Cognitive Level: Remember
    REF: Chromosome Aberrations and Associated Diseases: Sex chromosome aneuploidy | Table 2.1
    TOP: Physiological Integrity

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14. What genetic disorder is the result if an individual possesses an XXY chromosome configuration?
    a. Turner's syndrome
    b. Klinefelter's syndrome
    c. Down syndrome
    d. Fragile X syndrome

    ANS: B
    Individuals with at least two X chromosomes and one Y chromosome in each cell (47 XXY karyotype) have a disorder known as Klinefelter's syndrome. A condition with the presence of a single X chromosome and no homologous X or Y chromosome, so the individual has a total of 45 chromosomes, is known as Turner's syndrome. Down syndrome is a trisomy. Fragile X syndrome is due to a break or a gap in a chromosome, not an extra chromosome.

    DIF: Cognitive Level: Remember
    REF: Chromosome Aberrations and Associated Diseases | Table 2.1
    TOP: Physiological Integrity

15. A patient demonstrates severe intellectual disability caused by a deletion of part of chromosome 5. What genetic disorder will the nurse see documented in the chart?
    a. Prader-Willi syndrome
    b. Down syndrome
    c. cri du chat syndrome
    d. Trisomy X

    ANS: C
    cri du chat syndrome means "cry of the cat" and describes the characteristic cry of the affected child. Another symptom of the disorder is intellectual disability. The disease is caused by a deletion of part of the short arm of chromosome 5. Prader-Willi syndrome is characterized by short stature, obesity, and hypogonadism. Down syndrome does cause mental retardation but is due to chromosome 21, not chromosome 5. Trisomy X can result in mental retardation but is due to an extra X chromosome.

    DIF: Cognitive Level: Understand REF: Abnormalities of Chromosome Structure
    TOP: Physiological Integrity

16. A support worker asks the nurse why people who have neurofibromatosis show varying degrees of the disease. Which genetic principle should the nurse explain to the support worker?
    a. Penetrance
    b. Expressivity
    c. Dominance
    d. Recessiveness

    ANS: B
    Expressivity is the extent of variation in phenotype associated with a particular genotype. For neurofibromatosis, a variety of manifestations occur among individuals. The penetrance of a trait is the percentage of individuals with a specific genotype who also exhibit the expected phenotype. Dominance refers to observable traits and risk of transmission. Recessiveness refers to silent strains with reduced risk of occurrence.

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DIF: Cognitive Level: Remember REF: Penetrance and Expressivity
TOP: Physiological Integrity

17. What gene abnormality causes cystic fibrosis?
    a. X-linked dominant
    b. X-linked recessive
    c. Autosomal dominant
    d. Autosomal recessive

    ANS: D
    Cystic fibrosis is an autosomal recessive disorder. It is not a result of X links or dominant pathology.

    DIF: Cognitive Level: Remember REF: Autosomal Recessive Inheritance
    TOP: Physiological Integrity

18. A 15-year-old patient with female genitalia is diagnosed with Prader-Willi syndrome. What is this condition an example of?
    a. Genomic imprinting
    b. An autosomal recessive trait
    c. An autosomal dominant trait
    d. A sex-linked trait

    ANS: A
    Prader-Willi, an example of gene imprinting, is not associated with any autosomal sex-linked abnormality.

    DIF: Cognitive Level: Understand REF: Epigenetics and Genomic Imprinting
    TOP: Physiological Integrity

19. A patient, age 9, is admitted to a pediatric unit with Duchenne muscular dystrophy. When planning care, the nurse recalls that the patient inherited this condition through a trait that is:
    a. X-linked dominant.
    b. X-influenced.
    c. X-limited.
    d. X-linked recessive.

    ANS: D
    Duchenne muscular dystrophy is a relatively common X-linked recessive, not dominant, disorder. While it is sex linked, it is not X-limited or X-influenced.

    DIF: Cognitive Level: Understand REF: Characteristics of Pedigrees
    TOP: Physiological Integrity

20. A child is diagnosed with cystic fibrosis. History reveals that the child's parents are siblings. Cystic fibrosis was most likely the result of:
    a. X-inactivation.
    b. genomic imprinting.
    c. consanguinity.
    d. obligate carriers.

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ANS: C
Consanguinity refers to the mating of two related individuals, and the offspring of such matings are said to be *inbred*. Consanguineous matings produce a significant increase in recessive disorders and are seen most often in pedigrees for rare recessive disorders. X-inactivation occurs when one X chromosome in the somatic cells of patients with female genitalia is permanently inactivated. Genomic imprinting is related to methylation and other changes. Obligate carriers are those who have an affected parent and affected children and, therefore, must themselves carry the mutation.

DIF: Cognitive Level: Understand REF: Consanguinity
TOP: Physiological Integrity

21. A 12-year-old patient is diagnosed with Klinefelter's syndrome. The patient's karyotype would reveal which of the following?
    a. XY
    b. XX
    c. XYY
    d. XXY

ANS: D
A person with Klinefelter's syndrome has an XXY karyotype. An XY is a normal male. An XX is a normal female. An XYY is an aneuploid karyotype.

DIF: Cognitive Level: Understand REF: X Inactivation
TOP: Physiological Integrity

22. What is needed for a polygenic trait to be expressed?
    a. Genes must interact with the environment.
    b. Several genes must act together.
    c. Multiple mutations must occur in the same family.
    d. Penetrance must occur.

ANS: B
Polygenic traits are those that result from several genes acting together. When environmental factors influence the expression of the trait, the term "multifactorial inheritance" is used. When multiple mutations occur in the same family, the mechanism most likely responsible is termed germline mosaicism. Penetrance of a trait is the percentage of individuals with a specific genotype who also exhibit the expected phenotype.

DIF: Cognitive Level: Understand REF: Multifactorial Inheritance
TOP: Physiological Integrity

23. A 13-year-old patient has a karyotype that reveals an absent homologous X chromosome with only a single X chromosome present. The patient's features include a short stature, widely spaced nipples, and a reduced carrying angle at the elbow. What is the patient's diagnosis?
    a. Down syndrome
    b. cri du chat syndrome
    c. Turner's syndrome
    d. Klinefelter's syndrome

ANS: C

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Turner's syndrome is characterized by short stature, webbed neck, shield-like chest with underdeveloped breasts and widely spaced nipples, and imperfectly developed ovaries. Down syndrome is characterized by distinctive characteristics: low nasal bridge, epicanthal folds, protruding tongue, and low-set ears. Cri du chat syndrome is characterized by low birth weight, severe mental retardation, microcephaly (smaller than normal head size), and heart defects. Klinefelter's syndrome is characterized by small testes, some development of the breasts, sparse body hair, and long limbs.

DIF: Cognitive Level: Understand REF: Table 2.1 TOP: Physiological Integrity

24. The gradual increase in height among the human population over the past 100 years is an example of:
    a. a polygenic trait.
    b. a multifactorial trait.
    c. crossing over.
    d. recombination.

    ANS: B
    The gradual increase in height is an example of multifactorial traits influenced by genes and also by environment. Polygenic traits result from several genes acting together. Crossing over is an abnormal chromosome structure. Recombination results from new arrangements of alleles.

    DIF: Cognitive Level: Understand REF: Multifactorial Inheritance
    TOP: Physiological Integrity

25. When discussing DNA replication, which enzyme is most important?
    a. RNA polymerase
    b. Transfer RNA
    c. Messenger RNA
    d. DNA polymerase

    ANS: D
    DNA polymerase, not RNA polymerase, is the primary enzyme involved in replication. It adds bases to the new DNA strand and performs "proofreading" functions. Neither messenger RNA nor transfer RNA is as important to DNA replication.

    DIF: Cognitive Level: Understand REF: Replication of DNA
    TOP: Physiological Integrity

26. What regions of RNA must be spliced out for functional mRNA to be formed?
    a. Promoter sites
    b. Introns
    c. Exons
    d. Anticodons

    ANS: B
    In eukaryotes, many RNA sequences are removed by nuclear enzymes, and the remaining sequences are spliced together to form the functional mRNA that migrates to the cytoplasm. The excised sequences are called introns (intervening sequences), and the sequences that are left to code for proteins are called exons.

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DIF: Cognitive Level: Understand REF: Gene Splicing
TOP: Physiological Integrity

27. A 50-year-old patient was recently diagnosed with Huntington's disease. What is a key feature of this condition?
    a. Penetrance of a trait
    b. Recurrence risk
    c. Expressivity
    d. Delayed age of onset

    ANS: D
    A key feature of Huntington's disease is its delayed age of onset such that symptoms are not seen until 40 years of age or later. The penetrance of a trait is the percentage of individuals with a specific genotype who also exhibit the expected phenotype. Recurrence risk is the percentage of family members who will inherit the disease. Expressivity is the extent of variation in phenotype associated with a particular genotype.

    DIF: Cognitive Level: Remember REF: Delayed Onset of Age
    TOP: Physiological Integrity

28. What type of mutation does not change the amino acid sequence and thus has no observable consequence?
    a. Frameshift
    b. Spontaneous
    c. Silent
    d. Missense

    ANS: C
    Silent mutations do not change the amino acid sequence and therefore have no consequences. Frameshift mutations involve the insertion or deletion of one or more base pairs of the DNA molecule. They alter the amino acid sequence. Spontaneous mutations occur in the absence of exposure to a mutagen and produce changes in the amino acid sequence. Missense mutations, a form of base pair substitution, alter amino acids, which produce a change (i.e., the "sense") in a single amino acid.

    DIF: Cognitive Level: Understand REF: Mutation TOP: Physiological Integrity

29. A nurse is reviewing the pedigree chart. When checking for a proband, what is the nurse looking for?
    a. The person who is first diagnosed with a genetic disease
    b. The individual who has a disease gene but is phenotypically normal
    c. The phenotype of genetic material
    d. The codominance

    ANS: A
    The pedigree chart summarizes family relationships and shows which members of a family are affected by a genetic disease. The pedigree begins with the proband. The person who has a disease gene but is phenotypically normal is a carrier. The phenotype is the result of both genotype and environment; it is not a proband. Codominance is not represented by a proband, but it occurs when the heterozygote is distinguishable from both homozygotes.

    DIF: Cognitive Level: Understand REF: Dominance and Recessiveness

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TOP: Physiological Integrity

30. Which one of the following disorders is manifested primarily in patients with male genitalia?
    a. Cystic fibrosis
    b. Neurofibromatosis
    c. Muscular dystrophy
    d. Down syndrome

    ANS: C
    Muscular dystrophy is manifested primarily in patients with male genitalia. Cystic fibrosis, neurofibromatosis, and Down syndrome are manifested in both patients with male and female genitalia.

    DIF: Cognitive Level: Understand REF: Characteristics of Pedigrees
    TOP: Physiological Integrity

MULTIPLE RESPONSE

1. When the nurse is teaching the staff about X-linked recessive disorders, which information should the nurse include? (*Select all that apply.*)
    a. The trait is seen much more often in patients with female genitalia than in patients with male genitalia.
    b. The trait is never transmitted from a parent with male genitalia to offspring with male genitalia.
    c. The gene can be transmitted through a series of carriers with female genitalia.
    d. The gene is passed from an affected parent with male genitalia to all offspring with female genitalia.
    e. The trait never skips generations.

    ANS: B, C, D
    The principles of X-linked recessive inheritance include the following: the trait is seen much more often in patients with male genitalia than in patients with female genitalia; the trait is never transmitted from a parent with male genitalia to offspring with male genitalia; the gene can be transmitted through a series of carriers with female genitalia; the gene is passed from an affected parent with male genitalia to all offspring with female genitalia, who, as phenotypically normal carriers, transmit it to approximately half their offspring with male genitalia, who are affected. X-linked recessive disorders can skip generations.

    DIF: Cognitive Level: Understand REF: Characteristics of Pedigrees
    TOP: Physiological Integrity

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