Huether and McCance: Understanding Pathophysiology, 5th Edition

Chapter 02: Genes and Genetic Diseases

Test Bank

MULTIPLE CHOICE

1. A nurse recalls 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
   DNA has three basic components: the pentose sugar molecule, deoxyribose; a phosphate molecule; and four types of nitrogenous bases.
   DNA contains four nitrogenous bases, not phosphate bases.
   Adenine and quinine are purines and are only a portion of the components of DNA.
   DNA synthesizes body protein, of which a codon is a component.

   REF: p. 35

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. They alter the amino acid sequence.
   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.

   REF: p. 37

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 are commonly represented by their first letters: A, C, T, and G.
The four base components of DNA are cytosine, thymine, adenine, and guanine and are commonly represented by their first letters: A, C, T, and G. U is not included.
The four base components of DNA are cytosine, thymine, adenine, and guanine and are commonly represented by their first letters: A, C, T, and G. P is not included.
X, XX, XY and YY are components of human chromosomes.

REF: p. 35

4. A DNA strand has a region with the sequence ATCGGAT. Which of the following would be a complementary strand?
   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.
   U does not represent a complement in the sequence.

   REF: p. 35

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.
   At a termination signal, translation and polypeptide formation cease. This does not involve 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.

   REF: p. 39

6. When homologous chromosomes fail to separate during meiosis, which of the following occurs?
   a. Neurofibromatosis
   b. Nondisjunction
   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.
   Neurofibromatosis is a dominant disorder. It 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.

   REF: p. 42

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
   A polyploid cell is one in which a euploid cell has more than the diploid number 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.

   REF: p. 39

8. A 20-year-old pregnant female 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 2 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.

REF: p. 40

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.

   REF: p. 40

10. After a geneticist talks to the patient about being a chromosomal mosaic, the patient asks the nurse what that means. How should the nurse respond? You may _____ genetic disease(s).
    a. Only be a carrier of the
    b. Have a mild form of the
    c. Have two
    d. Be sterile as a result of the

    ANS: B
    A chromosomal mosaic means the body has two or more different cell lines, each of which has a different karyotype; thus, the person has a mild form of the disease.
    Mosaics are not only carriers; they have the disease.
    Mosaics have two different lines but not two different diseases.
    Mosaics are not sterile.

    REF: p. 42

11. The nurse is teaching staff about the most common cause of Down syndrome. What is the nurse describing?
    a. Paternal nondisjunction
    b. Maternal translocations
    c. Maternal nondisjunction
    d. Paternal translocations

    ANS: C
    The most common cause of Down syndrome is maternal nondisjunction.
    Down syndrome is not related to paternal nondisjunction.

Down syndrome is related to the maternal side, but not due to translocation.
Down syndrome is not related to paternal abnormalities.

REF: p. 42

12. A patient wants to know the risk factors for Down syndrome. What is the nurse's best response?
    a. Fetal exposure to mutagens in the uterus
    b. Increased paternal age
    c. Family history of Down syndrome
    d. Pregnancy in women over age 35

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

    REF: p. 42

13. A 13-year-old girl 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 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 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.

    REF: p. 43

14. An XXY person asks the nurse what this genetic disorder is called. What is the nurse's best response? This disorder is _____ syndrome.
    a. Turner
    b. Klinefelter
    c. Down
    d. Fragile X

    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 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 syndrome.
Down syndrome is a trisomy.
Fragile X syndrome is due to a break or a gap in a chromosome, not an extra chromosome.

REF: p. 43

15. A patient has severe mental retardation 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 mental retardation.
    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.

    REF: p. 43

16. A couple has three offspring: one child with an autosomal dominant disease trait and two who are normal. The father is affected by the autosomal dominant disease, but the mother does not have the disease gene. What is the recurrence risk of this autosomal dominant disease for their next child?
    a. 50%
    b. 33%
    c. 25%
    d. Impossible to determine

    ANS: A
    For each child with an autosomal dominant disease parent there is a 1 in 2, or 50%, risk.
    The risk is 50%.
    The risk is 50%.
    The risk can be determined and the risk is 50%.

    REF: p. 46

17. An aide asks the nurse why people who have neurofibromatosis will show varying degrees of the disease. Which genetic principle should the nurse explain to the aide?
    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.

REF: p. 48

18. When a patient asks what causes cystic fibrosis, how should the nurse respond? Cystic fibrosis is caused by an _____ gene.
a. X-linked dominant
b. X-linked recessive
c. Autosomal dominant
d. Autosomal recessive

ANS: D
Cystic fibrosis is an autosomal recessive disorder.
Cystic fibrosis is not X linked, but autosomal.
Cystic fibrosis is not X linked, but recessive.
Cystic fibrosis is not dominant.

REF: p. 50

19. A 15-year-old female is diagnosed with Prader-Willi syndrome. This condition is an example of:
a. Gene imprinting
b. An autosomal recessive trait
c. An autosomal dominant trait
d. A sex-linked trait

ANS: A
Prader-Willi is an example of gene imprinting.
Prader-Willi is not an autosomal recessive trait, but due to gene imprinting.
Prader-Willi is not an autosomal dominant train, but due to gene imprinting.
Prader-Willi is not a sex-linked trait, but due to gene imprinting.

REF: p. 49

20. A patient, age 9, is admitted to a pediatric unit with Duchenne muscular dystrophy. When planning care the nurse recalls the patient inherited this condition through a _____ trait.

a. Sex-linked dominant
b. Sex-influenced
c. Sex-limited
d. Sex-linked recessive

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

REF: p. 52

21. A child is born with blue eyes (bb). The child's mother has blue eyes and the father has brown eyes. Which of the following represents the father?
a. Bb
b. Bb
c. BB
d. Bbb

ANS: B
The father would need to have one b and one B because blue eyes are recessive and the father has a dominant eye color.
bb is incorrect because, with this description, the father could not have brown eyes.
BB would describe the father's brown eyes, but without a b he could not father a blue-eyed child.
The father would have only two, not three.

REF: p. 52

22. 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

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 females 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.

REF: p. 50

23. A 12-year-old male is diagnosed with Klinefelter syndrome. His karyotype would reveal which of the following?
    a. XY
    b. XX
    c. XYY
    d. XXY

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

    REF: p. 51

24. To express a polygenic trait:
    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.

    REF: p. 54

25. What is the diagnosis of a 13-year-old female who has a karyotype that reveals an absent homologous X chromosome with only a single X chromosome present? Her features include a short stature, widely spaced nipples, reduced carrying angle at the elbow, and sparse body hair.
    a. Down syndrome
    b. Cri du chat syndrome
    c. Turner syndrome
    d. Klinefelter syndrome

    ANS: C
    Turner syndrome is characterized by short stature, female genitalia, webbed neck, shieldlike 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 syndrome is characterized by small testes, some development of the breasts, sparse body hair, and long limbs.

REF: p. 43

26. A normal male and a female carrier for red-green color blindness mate. Given that red-green color blindness is an X-linked recessive trait, what is the likelihood of their children being affected?
    a. 25%
    b. 50%
    c. Females most affected; no males affected
    d. Males most affected; no females affected

    ANS: D
    Because a single copy of an X-linked recessive gene will cause disease in a male, whereas two copies are required for disease expression in females, more males are affected by X-linked recessive diseases than are females.
    Males are more often affected at a greater than 25% rate.
    Males are more often affected at a greater than 50% rate.
    Males are most often affected.

    REF: p. 27

27. 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.

    REF: p. 54

28. 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 is the primary enzyme involved in replication. It adds bases to the new DNA strand and performs "proofreading" functions.
It is DNA polymerase, not RNA polymerase, that is most important for DNA replication.
It is DNA polymerase, not transfer RNA, that is most important for DNA replication.
It is DNA polymerase, not messenger RNA, that is most important for DNA replication.

REF: p. 36

29. The regions of the heterogeneous nuclear RNA that must be spliced out to form functional RNA are called:
a. Promoter sites
b. Introns
c. Exons
d. Anticodon

ANS: B
Introns are spliced from the mRNA before the mRNA leaves the nucleus.
A promoter site is a sequence of DNA that specifies the beginning of a gene.
In eukaryotes, many RNA sequences are removed by nuclear enzymes; the sequences that are left to code for proteins are called exons.
In translation, RNA directs the synthesis of a polypeptide, a cloverleaf-shaped strand of about 80 nucleotides. The tRNA molecule has a site where an amino acid attaches. The three-nucleotide sequence at the opposite side of the cloverleaf is called the anticodon.

REF: p. 39

30. A 5-year-old male presents with mental retardation and is diagnosed with fragile X syndrome. When the parents ask what caused this, how should the geneticist respond? This was most probably caused from:
a. Translocation
b. Inversion
c. Nondisjunction
d. Duplication at fragile sites

ANS: D
Unaffected transmitting males have been shown to have more than about 50 repeated DNA sequences near the beginning of the fragile X gene.
Translocation effects do not result in mental retardation.
Inversions have no physical effects.
Fragile X is not related to nondisjunction.

REF: p. 45

31. A 50-year-old male was recently diagnosed with Huntington disease. Transmission of this disease is associated with:
    a. Penetrance
    b. Recurrence risk
    c. Expressivity
    d. Delayed age of onset

    ANS: D
    A key feature of Huntington 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 that will inherit the disease.
    Expressivity is the extent of variation in phenotype associated with a particular genotype.

    REF: p. 48

32. Mutations that do not change the amino acid sequence and thus have no consequence are termed _____ mutations.
    a. Frameshift
    b. Spontaneous
    c. Silent
    d. Missense

    ANS: C
    Silent mutations do not change the amino acid sequence and have no consequences.
    Frameshift mutation involves 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 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.

    REF: p. 37

33. 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 proband, but it occurs when the heterozygote is distinguishable from both homozygotes.

REF: p. 46

34. Which of the following disorders is manifested primarily in males?
    a. Cystic fibrosis
    b. Neurofibromatosis
    c. Muscular dystrophy
    d. Klinefelter syndrome

    ANS: C
    Muscular dystrophy is manifested primarily in males.
    Cystic fibrosis is manifested in males and females.
    Neurofibromatosis is manifested in males and females.
    Klinefelter syndrome is manifested in males and females.

    REF: p. 52

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 females than in males.
    b. The trait is never transmitted from father to son.
    c. The gene can be transmitted through a series of carrier females.
    d. The gene is passed from an affected father to all his daughters.
    e. The trait never skips generations.

    ANS: B, C, D
    The principles of X-linked recessive inheritance include: the trait is seen much more often in males than in females; the trait is never transmitted from father to son; the gene can be transmitted through a series of carrier females; the gene is passed from an affected father to all his daughters, who, as phenotypically normal carriers, transmit it to approximately half their sons, who are affected. X-linked recessive disorders can skip generations since it is a 1 in 4 chance.

    REF: p. 52