Chapter 03: Hereditary Influences on Health Promotion of the Child and Family

MULTIPLE CHOICE

1. Which genetic term refers to a person who possesses one copy of an affected gene and one copy of an unaffected gene and is clinically unaffected?
   a. Allele
   b. Carrier
   c. Pedigree
   d. Multifactorial

   ANS: B
   An individual who is a carrier is asymptomatic but possesses a genetic alteration, either in the form of a gene or chromosome change. Alleles are alternative expressions of genes at a different locus. A pedigree is a diagram that describes family relationships, gender, disease, status, or other relevant information about a family. Multifactorial describes a complex interaction of both genetic and environmental factors that produce an effect on the individual.

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   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

2. Which genetic term refers to the transfer of all or part of a chromosome to a different chromosome after chromosome breakage?
   a. Trisomy
   b. Monosomy
   c. Translocation
   d. Nondisjunction

   ANS: C
   Translocation is the transfer of all or part of a chromosome to a different chromosome after chromosome breakage. It can be balanced, producing no phenotypic effects, or unbalanced, producing severe or lethal effects. Trisomy is an abnormal number of chromosomes caused by the presence of an extra chromosome, which is added to a given chromosome pair and results in a total of 47 chromosomes per cell. Monosomy is an abnormal number of chromosomes whereby the chromosome is represented by a single copy in a somatic cell. Nondisjunction is the failure of homologous chromosomes or chromatids to separate during mitosis or meiosis.

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   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

3. Which is a birth defect or disorder that occurs as a new case in a family and is not inherited?
   a. Sporadic
   b. Polygenic
   c. Monosomy
   d. Association

   ANS: A

Sporadic describes a birth defect previously unidentified in a family. It is not inherited. Polygenic inheritance involves the inheritance of many genes at separate loci whose combined effects produce a given phenotype. Monosomy is an abnormal number of chromosomes whereby the chromosome is represented by a single copy in a somatic cell. A nonrandom cluster of malformations without a specific cause is an association.

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4. The nurse is assessing a neonate who was born 1 hour ago to healthy white parents in their early forties. Which finding should be most suggestive of Down syndrome?
   a. Hypertonia
   b. Low-set ears
   c. Micrognathia
   d. Long, thin fingers and toes

   ANS: B
   Children with Down syndrome have low-set ears. Infants with Down syndrome have hypotonia, not hypertonia. Micrognathia is common in trisomy 16, not Down syndrome. Children with Down syndrome have short hands with broad fingers.

   DIF: Cognitive Level: Understanding REF: p. 82
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

5. Which abnormality is a common sex chromosome defect?
   a. Down syndrome
   b. Turner syndrome
   c. Marfan syndrome
   d. Hemophilia

   ANS: B
   Turner syndrome is caused by an absence of one of the X chromosomes. Down syndrome is caused by trisomy 21 (three copies rather than two copies of chromosome 21). Marfan syndrome is a connective tissue disorder inherited in an autosomal dominant pattern. Hemophilia is a disorder of blood coagulation inherited in an X-linked recessive pattern.

   DIF: Cognitive Level: Understanding REF: p. 53
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

6. Turner syndrome is suspected in an adolescent girl with short stature. What causes this?
   a. Absence of one of the X chromosomes
   b. Presence of an incomplete Y chromosome
   c. Precocious puberty in an otherwise healthy child
   d. Excess production of both androgens and estrogens

   ANS: A
   Turner syndrome is caused by an absence of one of the X chromosomes. Most girls who have this disorder have one X chromosome missing from all cells. No Y chromosome is present in individuals with Turner syndrome. These young women have 45 rather than 46 chromosomes.

   DIF: Cognitive Level: Understanding REF: p. 53
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

7. Which is a sex chromosome abnormality that is caused by the presence of one or more additional X chromosomes in a male?
   a. Turner
   b. Triple X
   c. Klinefelter
   d. Trisomy 13

   ANS: C
   Klinefelter syndrome is characterized by one or more additional X chromosomes. These individuals are tall with male secondary sexual characteristics that may be deficient, and they may be learning disabled. An absence of an X chromosome results in Turner syndrome. Triple X and trisomy 13 are not abnormalities that involve one or more additional X chromosomes in a male (Klinefelter syndrome).

   DIF: Cognitive Level: Understanding REF: p. 53
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

8. Parents ask the nurse about the characteristics of autosomal dominant inheritance. Which statement is characteristic of autosomal dominant inheritance?
   a. Females are affected with greater frequency than males.
   b. Unaffected children of affected individuals will have affected children.
   c. Each child of a heterozygous affected parent has a 50% chance of being affected.
   d. Any child of two unaffected heterozygous parents has a 25% chance of being affected.

   ANS: C
   In autosomal dominant inheritance, only one copy of the mutant gene is necessary to cause the disorder. When a parent is affected, there is a 50% chance that the chromosome with the gene for the disorder will be contributed to each pregnancy. Males and females are equally affected. The disorder does not "skip" a generation. If the child is not affected, then most likely he or she is not a carrier of the gene for the disorder. In autosomal recessive inheritance, any child of two unaffected heterozygous parents has a 25% chance of being affected.

   DIF: Cognitive Level: Applying REF: p. 57
   TOP: Integrated Process: Teaching/Learning
   MSC: Client Needs: Health Promotion and Maintenance

9. Parents ask the nurse about the characteristics of autosomal recessive inheritance. Which is characteristic of autosomal recessive inheritance?
   a. Affected individuals have unaffected parents.
   b. Affected individuals have one affected parent.
   c. Affected parents have a 50% chance of having an affected child.
   d. Affected parents will have unaffected children.

   ANS: A
   Parents who are carriers of a recessive gene are asymptomatic. For a child to be affected, both parents must have a copy of the gene, which is passed to the child. Both parents are asymptomatic but can have affected children. In autosomal recessive inheritance, there is a 25% chance that each pregnancy will result in an affected child. In autosomal dominant inheritance, affected parents can have unaffected children.

DIF: Cognitive Level: Applying REF: p. 62
TOP: Integrated Process: Teaching/Learning
MSC: Client Needs: Health Promotion and Maintenance

10. Which is characteristic of X-linked recessive inheritance?
    a. There are no carriers.
    b. Affected individuals are principally males.
    c. Affected individuals are principally females.
    d. Affected individuals will always have affected parents.

    ANS: B
    In X-linked recessive disorders, the affected individuals are usually male. With recessive traits, usually two copies of the gene are needed to produce the effect. Because the male only has one X chromosome, the effect is visible with only one copy of the gene. Females are usually only carriers of X-linked recessive disorders. The X chromosome that does not have the recessive gene will produce the "normal" protein, so the woman will not show evidence of the disorder. The transmission is from mother to son. Usually the mother and father are unaffected.

    DIF: Cognitive Level: Understanding REF: p. 64
    TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

11. A father with an X-linked recessive disorder asks the nurse what the probability is that his sons will have the disorder. Which response should the nurse make?
    a. "Male children will be carriers."
    b. "All male children will be affected."
    c. "None of the sons will have the disorder."
    d. "It cannot be determined without more data."

    ANS: C
    When a male has an X-linked recessive disorder, he has one copy of the allele on his X chromosome. The father passes only his Y chromosome (not the X chromosome) to his sons. Therefore, none of his sons will have the X-linked recessive gene. They will not be carriers or be affected by the disorder. No additional data are needed to answer this question.

    DIF: Cognitive Level: Applying REF: p. 64
    TOP: Integrated Process: Teaching/Learning
    MSC: Client Needs: Health Promotion and Maintenance

12. The inheritance of which is X-linked recessive?
    a. Hemophilia A
    b. Marfan syndrome
    c. Neurofibromatosis
    d. Fragile X syndrome

    ANS: A
    Hemophilia A is inherited as an X-linked recessive trait. Marfan syndrome and neurofibromatosis are inherited as autosomal dominant disorders. Fragile X is inherited as an X-linked trait.

    DIF: Cognitive Level: Understanding REF: p. 64

TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

13. Chromosome analysis of the fetus is usually accomplished through the testing of which?
    a. Fetal serum
    b. Maternal urine
    c. Amniotic fluid
    d. Maternal serum

    ANS: C
    Amniocentesis is the most common method to retrieve fetal cells for chromosome analysis. Viable fetal cells are sloughed off into the amniotic fluid, and when a sample is taken, they can be cultured and analyzed. It is difficult to obtain a sample of the fetal blood. It is a high-risk situation for the fetus. Fetal cells are not present in the maternal urine or blood.

    DIF: Cognitive Level: Analyzing REF: p. 46
    TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

14. A couple asks the nurse about the optimal time for genetic counseling. They do not plan to have children for several years. When should the nurse recommend they begin genetic counseling?
    a. As soon as the woman suspects that she may be pregnant
    b. Whenever they are ready to start their family
    c. Now, if one of them has a family history of congenital heart disease
    d. Now, if they are members of a population at risk for certain diseases

    ANS: D
    Persons who seek genetic evaluation and counseling must first be aware if there is a genetic or potential problem in their families. Genetic testing should be done now if the couple is part of a population at risk. It is not feasible at this time to test for all genetic diseases. The optimal time for genetic counseling is before pregnancy occurs. During the pregnancy, genetic counseling may be indicated if a genetic disorder is suspected. Congenital heart disease is not a single-gene disorder.

    DIF: Cognitive Level: Applying REF: p. 62
    TOP: Integrated Process: Teaching/Learning
    MSC: Client Needs: Health Promotion and Maintenance

15. A woman, age 43 years, is 6 weeks pregnant. It is important that she be informed of which?
    a. The need for a therapeutic abortion
    b. Increased risk for Down syndrome
    c. Increased risk for Turner syndrome
    d. The need for an immediate amniocentesis

    ANS: B
    Women who are older than age 35 years at the birth of a single child or 31 years at the birth of twins are advised to have prenatal diagnosis. The risk of having a child with Down syndrome increases with maternal age. There is no indication of a need for a therapeutic abortion at this stage. Turner syndrome is not associated with advanced maternal age. Amniocentesis cannot be done at a gestational age of 6 weeks.

    DIF: Cognitive Level: Applying REF: p. 51
    TOP: Integrated Process: Teaching/Learning

MSC: Client Needs: Health Promotion and Maintenance

16. A couple has given birth to their first child, a boy with a recessive disorder. The genetic counselor tells them that the risk of recurrence is one in four. Which statement is a correct interpretation of this information?
    a. The risk factor remains the same for each pregnancy.
    b. The risk factor will change when they have a second child.
    c. Because the parents have one affected child, the next three children should be unaffected.
    d. Because the parents have one affected child, the next child is four times more likely to be affected.

    ANS: A
    Each pregnancy has the same risks for an affected child. Because an odds ratio reflects the risk, this does not change over time. The statement by the genetic counselor refers to a probability. This does not change over time. The statement "Because the parents have one affected child, the next child is four times more likely to be affected" does not reflect autosomal recessive inheritance.

    DIF: Cognitive Level: Analyzing REF: p. 57
    TOP: Integrated Process: Teaching/Learning
    MSC: Client Needs: Health Promotion and Maintenance

17. A couple expecting their first child has a positive family history for several congenital defects and disorders. The couple tells the nurse that they are opposed to abortion for religious reasons. Which should the nurse consider when counseling the couple?
    a. The couple should be encouraged to have recommended diagnostic testing.
    b. The couple needs counseling regarding advantages and disadvantages of pregnancy termination.
    c. Diagnostic testing is required by law in this situation.
    d. Diagnostic testing is of limited value if termination of pregnancy is not an option.

    ANS: A
    The benefits of prenatal diagnostic testing extend beyond decisions concerning abortion. If the child has congenital disorders, decisions can be made about fetal surgery if indicated. In addition, if the child is expected to require neonatal intensive care at birth, the mother is encouraged to deliver at a level III neonatal center. The couple is counseled about the advantages and disadvantages of prenatal diagnosis, not pregnancy termination, although the family cannot be forced to have prenatal testing. The information gives the parents time to grieve and plan for their child if congenital disorders are present. If the child is free of defects, then the parents are relieved of a major worry.

    DIF: Cognitive Level: Applying REF: p. 71
    TOP: Integrated Process: Teaching/Learning
    MSC: Client Needs: Health Promotion and Maintenance

18. Parents ask the nurse if there was something that should have been done during the pregnancy to prevent their child's cleft lip. Which statement should the nurse give as a response?
    a. "This is a type of deformation and can sometimes be prevented."
    b. "Studies show that taking folic acid during pregnancy can prevent this defect."
    c. "This is a genetic disorder and has a 25% chance of happening with each

pregnancy."

d. "The malformation occurs at approximately 5 weeks of gestation; there is no known way to prevent this."

ANS: D

Cleft lip, an example of a malformation, occurs at approximately 5 weeks of gestation when the developing embryo naturally has two clefts in the area. There is no known way to prevent this defect. Deformations are often caused by extrinsic mechanical forces on normally developing tissue. Club foot is an example of a deformation often caused by uterine constraint. Cleft lip is not a genetic disorder; the reasons for this occurring are still unknown. Taking folic acid during pregnancy can help to prevent neural tube disorders but not cleft lip defects.

DIF: Cognitive Level: Applying REF: p. 49
TOP: Integrated Process: Teaching/Learning
MSC: Client Needs: Health Promotion and Maintenance

19. The nurse is teaching parents of a child with cri du chat syndrome about this disorder. The nurse understands parents understand the teaching if they make which statement?
    a. "This disorder is very common."
    b. "This is an autosomal recessive disorder."
    c. "The crying pattern is abnormal and catlike."
    d. "The child will always have a moon-shaped face."

ANS: C

Typical of this disease is a crying pattern that is abnormal and catlike. Cri du chat, or cat's cry, syndrome is a rare (one in 50,000 live births) chromosome deletion syndrome, not autosomal recessive, resulting from loss of the small arm of chromosome 5. In early infancy this syndrome manifests with a typical but nondistinctive facial appearance, often a "moon-shaped" face with wide-spaced eyes (hypertelorism). As the child grows, this feature is progressively diluted, and by age 2 years, the child is indistinguishable from age-matched control participants.

DIF: Cognitive Level: Applying REF: pp. 54-55
TOP: Integrated Process: Teaching/Learning
MSC: Client Needs: Health Promotion and Maintenance

20. The nurse is reviewing a client's prenatal history. Which prescribed medication does the nurse understand is not considered a teratogen and prescribed during pregnancy?
    a. Phenytoin (Dilantin)
    b. Warfarin (Coumadin)
    c. Isotretinoin (Accutane)
    d. Heparin sodium (Heparin)

ANS: D

Teratogens, agents that cause birth defects when present in the prenatal environment, account for the majority of adverse intrauterine effects not attributable to genetic factors. Types of teratogens include drugs (phenytoin [Dilantin], warfarin [Coumadin], isotretinoin [Accutane]). Heparin is the anticoagulant used during pregnancy and is not a teratogen. It does not cross the placenta.

DIF: Cognitive Level: Analyzing REF: p. 68 TOP: Nursing Process: Evaluation

MSC: Integrated Process: Physiological Integrity

21. The nurse is teaching student nurses about newborn screening. Which statement made by the student indicates understanding of the teaching?
    a. "The newborn screening is not mandatory but voluntary."
    b. "It is acceptable to 'layer' the blood on the Guthrie paper."
    c. "The initial specimen should be collected as close to discharge as possible."
    d. "It is best to collect the specimen before the newborn takes the first feeding."

    ANS: C
    Because of early discharge of newborns, recommendations for screening include collecting the initial specimen as close as possible to discharge. Newborn screening tests are mandatory in all 50 U.S. states. When collecting the specimen, avoid "layering" the blood specimen on the special Guthrie paper. Layering is placing one drop of blood on top of the other or overlapping the specimen. Best results are obtained by collecting the specimen with a pipette from the heel stick and spreading the blood uniformly over the blot paper. The screening test is most reliable if the blood sample is taken after the infant has ingested a source of protein.

    DIF: Cognitive Level: Applying REF: p. 71
    TOP: Integrated Process: Teaching/Learning
    MSC: Client Needs: Health Promotion and Maintenance

22. A hospitalized school-age child with phenylketonuria (PKU) is choosing foods from the hospital's menu. Which food choice should the nurse discourage the child from choosing?
    a. Banana
    b. Milkshake
    c. Fruit juice
    d. Corn on the cob

    ANS: B
    Foods with low phenylalanine levels (e.g., some vegetables [except legumes]; fruits; juices; and some cereals, breads, and starches) must be measured to provide the prescribed amount of phenylalanine. Most high-protein foods, such as meat and dairy products, are either eliminated or restricted to small amounts.

    DIF: Cognitive Level: Applying REF: pp. 71-72
    TOP: Nursing Process: Implementation MSC: Client Needs: Health Promotion and Maintenance

23. The nurse understands that which occurring soon after birth can indicate cystic fibrosis?
    a. Murmur
    b. Hypoglycemia
    c. Meconium ileus
    d. Muscle weakness

    ANS: C
    A symptom of cystic fibrosis is a meconium ileus soon after birth. A murmur can be a sign of a congenital heart disease. Hypoglycemia can be a sign of Beckwith-Wiedemann syndrome. Muscle weakness can be a sign of myotonic dystrophy.

    DIF: Cognitive Level: Understanding REF: p. 59
    TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

24. A newborn has been diagnosed with congenital adrenal hyperplasia. Which assessment finding should the nurse expect?
    a. Ambiguous genitalia
    b. Prenatal growth retardation
    c. An abnormally large tongue
    d. Legs and arms significantly shorter than torso

    ANS: A
    A newborn diagnosed with congenital adrenal hyperplasia can have ambiguous genitalia or virilization of female external genitalia caused by elevated androgen levels. Prenatal growth retardation is present with Bloom syndrome. An abnormally large tongue is seen with Beckwith-Wiedemann syndrome. Legs and arms significantly shorter than torso are seen with achondroplasia.

    DIF: Cognitive Level: Analyzing REF: p. 59
    TOP: Nursing Process: Assessment MSC: Integrated Process: Physiological Integrity

25. Parents of a child with hemophilia A ask the nurse, "What is the deficiency with this disorder?" Which correct response should the nurse make?
    a. "Hemophilia A has a deficiency in red blood cells."
    b. "Hemophilia A has a deficiency in platelets."
    c. "Hemophilia A has a deficiency in factor IX."
    d. "Hemophilia A has a deficiency in factor VIII."

    ANS: D
    Hemophilia A is deficient in factor VIII. Glucose-6-phosphate dehydrogenase (G6PD) deficiency shows low red blood cells (hemolytic anemia). Immunosuppression may be the cause of a deficient number of platelets. Hemophilia B is deficient in factor IX.

    DIF: Cognitive Level: Applying REF: p. 60
    TOP: Integrated Process: Teaching/Learning
    MSC: Client Needs: Health Promotion and Maintenance

26. A child with Prader-Willi syndrome has been hospitalized. Which assessment findings does the nurse expect with this syndrome?
    a. Nonverbal
    b. Insatiable hunger
    c. Abnormal, puppetlike gait
    d. Paroxysms of inappropriate laughter

    ANS: B
    Prader-Willi syndrome is characterized by insatiable hunger that can lead to morbid obesity in childhood. Abnormal, puppetlike gait, paroxysms of inappropriate laughter, and nonverbal are characteristics seen in Angelman syndrome.

    DIF: Cognitive Level: Analyzing REF: p. 66
    TOP: Nursing Process: Assessment MSC: Integrated Process: Physiological Integrity

27. Which ethnic group is at risk for Tay-Sachs disease?
    a. Black African
    b. Mediterranean
    c. Ashkenazi Jewish

d. Southern and Southeast Asian

ANS: C
The Ashkenazi Jewish ethnic group is at higher risk for Tay-Sachs disease. The black African, Mediterranean, and Southern and Southeast Asian ethnicities are at higher risk for sickle cell anemia disease.

DIF: Cognitive Level: Understanding REF: p. 78
TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

28. A child has been found to have a deficiency in 3-hydroxy-3-methylglutaryl coenzyme A (HMG-CoA) reductase. Which condition is this child at risk for?
a. Increased uric acid
b. Hypercholesterolemia
c. Increased phenylketones
d. Altered oxygen transport

ANS: B
HMG-CoA leads to a disruption of metabolic feedback mechanism and accumulation of end product (cholesterol) with the resulting condition of hypercholesterolemia.

DIF: Cognitive Level: Analyzing REF: p. 48
TOP: Nursing Process: Assessment MSC: Integrated Process: Physiological Integrity

29. Phenylketonuria is a genetic disease that results in the body's inability to correctly metabolize which?
a. Glucose
b. Thyroxine
c. Phenylalanine
d. Phenylketones

ANS: C
Phenylketonuria is an inborn error of metabolism caused by a deficiency or absence of the enzyme needed to metabolize the essential amino acid phenylalanine. Individuals with this disorder can metabolize glucose. Thyroxine is one of the principal hormones secreted by the thyroid gland. Phenylketones are metabolites of phenylalanine excreted in the urine.

DIF: Cognitive Level: Understanding REF: p. 61
TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

30. Early diagnosis of congenital hypothyroidism (CH) and phenylketonuria (PKU) is essential to prevent which?
a. Obesity
b. Diabetes
c. Cognitive impairment
d. Respiratory distress

ANS: C
Untreated, both PKU and CH cause cognitive impairment. With newborn screening and early intervention, cognitive impairment from these two disorders can be prevented. Obesity, diabetes, and respiratory distress do not result from both CH and PKU.

DIF: Cognitive Level: Understanding REF: p. 61
TOP: Nursing Process: Assessment MSC: Client Needs: Physiological Integrity

31. A breastfed infant has just been diagnosed with galactosemia. The therapeutic management of this includes which?
    a. Stop breastfeeding the infant.
    b. Add amino acids to breast milk.
    c. Substitute a lactose-containing formula for breast milk.
    d. Give the appropriate enzyme along with breast milk.

    ANS: A
    The infant with galactosemia is fed a diet free of all milk and lactose-containing foods. This includes breast milk. Soy-protein formula is the formula of choice. Other strategies are being identified.

    DIF: Cognitive Level: Understanding REF: pp. 73-74 TOP: Nursing Process: Planning
    MSC: Client Needs: Physiological Integrity

MULTIPLE RESPONSE

1. Which can be directly attributed to a single-gene disorder? *(Select all that apply.)*
    a. Cleft lip
    b. Cystic fibrosis
    c. Turner syndrome
    d. Klinefelter syndrome
    e. Neurofibromatosis

    ANS: B, E
    Cystic fibrosis is a single-gene disorder inherited as an autosomal recessive trait, and neurofibromatosis is a single-gene disorder inherited as an autosomal dominant trait. Cleft lip is classified as a multifactorial disorder in which a genetic susceptibility and appropriate environment appear to play important roles. Turner and Klinefelter syndromes are disorders of sex chromosome number.

    DIF: Cognitive Level: Analyzing REF: p. 49
    TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

2. The nurse is reviewing the characteristics of autosomal dominant inheritance. Which are true about these characteristics? *(Select all that apply.)*
    a. A carrier state exists.
    b. The phenotype appears in consecutive generations.
    c. Males and females are equally likely to be affected.
    d. Parents who have affected children are usually asymptomatic.
    e. Children of an affected parent have a 50% chance of being affected.

    ANS: B, C, E
    Characteristics of autosomal dominant inheritance include the phenotype appears in consecutive generations, males and females are equally affected, and children of an affected parent have a 50% chance of being affected. A carrier state and parents who have affected children are usually asymptomatic are characteristic of autosomal recessive inheritance.

DIF: Cognitive Level: Analyzing REF: p. 55
TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

3. The nurse is reviewing the characteristics of autosomal recessive inheritance. Which are true about these characteristics? *(Select all that apply.)*
   a. Most affected persons are males.
   b. Males and females are equally affected.
   c. All daughters of an affected male are carriers.
   d. Carrier parents have a 25% chance of producing an affected child.
   e. Carrier parents have a 50% chance of producing a carrier child in each pregnancy.

   ANS: B, D, E
   Characteristics of autosomal recessive inheritance include males and females are equally affected, carrier parents have a 25% chance of producing an affected child, and carrier parents have a 50% chance of producing a carrier child in each pregnancy. Most affected persons who are males and all daughters of an affected male are carriers are characteristics of X-linked recessive inheritance.

   DIF: Cognitive Level: Analyzing REF: p. 57
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

4. Which are signs and symptoms the nurse should assess in the newborn that can indicate an inborn error of metabolism? *(Select all that apply.)*
   a. Jaundice
   b. Strabismus
   c. Poor feeding
   d. Acrocyanosis
   e. Metabolic acidosis

   ANS: A, C, E
   Signs of inborn errors of metabolism include jaundice, poor feeding, and metabolic acidosis. Strabismus and acrocyanosis are normal findings in the newborn.

   DIF: Cognitive Level: Applying REF: p. 68
   TOP: Nursing Process: Assessment MSC: Client Needs: Physiological Integrity

5. The nurse is interviewing a prenatal client about specific risk factors that are indications for prenatal testing. Which specific risk factors should the nurse note? *(Select all that apply.)*
   a. Previous twins
   b. Inherited disorder
   c. Previous preterm birth
   d. Cytomegalovirus infection
   e. Previous stillbirth or neonatal death

   ANS: B, D, E
   Specific risk factors that are indications for prenatal testing include inherited disorder, cytomegalovirus infection (teratogenic infection), and previous stillbirth or neonatal death. Previous twins or previous preterm birth are not specific risk factors that are indications for prenatal testing.

   DIF: Cognitive Level: Analyzing REF: p. 82
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance

6. The nurse is teaching nursing students about assessment clues to genetic disorders in the newborn. Which should the nurse include in the teaching session? *(Select all that apply.)*
   a. Low-set ears
   b. Mongolian spots
   c. Epicanthal folds
   d. Cephalohematoma
   e. Forehead prominence

   ANS: A, C, E
   Assessment clues to genetic disorders in the newborn include low-set ears, epicanthal folds, and forehead prominence. Mongolian spots and cephalohematoma are findings in a newborn that are not indicative of a genetic disorder.

   DIF: Cognitive Level: Applying REF: p. 45
   TOP: Integrated Process: Teaching/Learning
   MSC: Client Needs: Health Promotion and Maintenance

MATCHING

Match the key genetic terms to their definitions.
a. Concordant
b. Congenital
c. Cytogenetics
d. Genome
e. Teratogen

1. Study of chromosomes, with special focus on chromosome abnormalities
2. Complete genetic information of an organism
3. A condition in which two individuals have the same genetic trait
4. Present at birth
5. An environmental agent capable of producing a birth defect

1. ANS: C DIF: Cognitive Level: Understanding REF: p. 47
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance
2. ANS: D DIF: Cognitive Level: Understanding REF: p. 46
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance
3. ANS: A DIF: Cognitive Level: Understanding REF: p. 46
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance
4. ANS: B DIF: Cognitive Level: Understanding REF: p. 46
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance
5. ANS: E DIF: Cognitive Level: Understanding REF: p. 48
   TOP: Nursing Process: Assessment MSC: Client Needs: Health Promotion and Maintenance