Lowdermilk: Maternity & Women's Health Care, 10th Edition

Chapter 03: Clinical Genetics

Test Bank

MULTIPLE CHOICE

1. A father and mother are carriers of phenylketonuria (PKU). Their 2-year-old daughter has PKU. The couple tells the nurse that they are planning to have a second baby. Because their daughter has PKU, they are sure that their next baby won't be affected. What response by the nurse is most accurate?

a. "Good planning; you need to take advantage of the odds in your favor."
b. "I think you'd better check with your doctor first."
c. "You are both carriers, so each baby has a 25% chance of being affected."
d. "The ultrasound indicates a boy, and boys are not affected by PKU."

ANS: C
The chance is one in four that each child produced by this couple will be affected by PKU disorder.
This couple still has an increased likelihood of having a child with PKU. Having one child already with PKU does not guarantee that they will not have another.
These parents need to discuss their options with their physician. However, an opportune time has presented itself for the couple to receive correct teaching about inherited genetic risks.
No correlation exists between gender and inheritance of the disorder, because PKU is an autosomal recessive disorder.

DIF: Cognitive Level: Application REF: 53
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Planning

2. A woman is 5 months pregnant. On a routine ultrasound scan, the physician discovers that the fetus has a diaphragmatic hernia. The woman becomes distraught and asks the nurse what she should do. What action by the nurse is most appropriate?

a. Talk to the woman and refer her to a genetic counselor.
b. Suggest that the woman travel to a fetal treatment center for intrauterine surgery.
c. Tell her that everything is going to be fine.
d. Sit with her and calmly suggest that she consider terminating this pregnancy.

ANS: A
Before the woman makes any decisions, she should discuss this newly discovered information with a genetic counselor. Genetic counselors can help with the diagnosis and management of families affected by genetic conditions.
The discussion of potential surgery should be pursuant to genetic counseling.

Telling the woman that everything is going to be fine may give her false hope and is not accurate.
All options should be discussed with the genetic counselor. Furthermore, the guiding principle for genetic counseling is nondirectiveness. This respects the right of the individual or family being counseled to make autonomous decisions.

DIF: Cognitive Level: Application REF: 56
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Planning

3. A woman who is gravida 2 and 16 GW comes in for her prenatal appointment. Her 2-year-old daughter is with her and is wearing a sleeveless top. While interacting with her daughter, you note axillary freckling and several café-au-lait spots (>2 cm). In reviewing her chart, you would assess for documentation of which genetic disease?

a. Tay-Sachs disease
b. Galactosemia
c. Neurofibromatosis (NF)
d. Phenylketonuria

ANS: C
Clinical manifestations of NF may include axillary freckling and café-au-lait spots.
Tay-Sachs disease is not associated with café-au-lait spots. Tay-Sachs is an incurable lipid-storage disorder.
Galactosemia is not associated with café-au-lait spots; rather it is an inborn error of metabolism.
Phenylketonuria is not associated with café-au-lait spots. This child would have difficulty manufacturing the liver enzyme phenylalanine.

DIF: Cognitive Level: Knowledge REF: 52
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

4. A new father has just been told that his child has trisomy 18. He asks the nurse what made her suspect a problem after the birth. The nurse explains that during the infant's newborn assessment, she noted:

a. Microcephaly and capillary hemangiomas
b. Epicanthal folds and a simian crease
c. Oblique palpebral fissures and Cri du chat syndrome
d. Rocker-bottom feet and clenched hands with overlapping fingers

ANS: D
Rocker-bottom feet and clenched hands with overlapping fingers are associated with trisomy 18.
Microcephaly and capillary hemangiomas are associated with trisomy 13.
Epicanthal folds and a simian crease are associated with trisomy 21 (Down syndrome).
Deletion of the short arm of chromosome number 5 is manifested by Cri du chat syndrome.

DIF: Cognitive Level: Comprehension REF: "50, 51"
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

5. A nurse is assessing the knowledge of new parents with a child born with maple syrup urine disease (MSUD). This is an autosomal recessive inherited disorder, which means that:
a. Both genes of a pair must be abnormal for the disorder to be expressed
b. Only one copy of the abnormal gene is required for the disorder to be expressed
c. The disorder occurs in males and heterozygous females
d. The disorder is carried on the X chromosome

ANS: A
MSUD is a type of autosomal recessive inheritance disorder in which both genes of a pair must be abnormal for the disorder to be expressed.
MSUD is an autosomal recessive inheritance disorder that cannot be expressed when only one copy of the abnormal gene is present.
MSUD is not an autosomal dominant inheritance disorder. The disorder would not be present with this genetic presentation.
MSUD is not an X- linked disorder; it is an autosomal recessive inheritance disorder in which both genes of a pair must be abnormal for the disorder to be present.

DIF: Cognitive Level: Comprehension REF: 53
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

6. In presenting to obstetric nurses interested in genetics, the genetic nurse identifies the primary risk(s) associated with genetic testing as:
a. Anxiety and altered family relationships
b. Denial of insurance benefits
c. High false positives associated with genetic testing
d. Ethnic and socioeconomic disparity associated with genetic testing

ANS: B
Decisions about genetic testing are shaped by socioeconomic status and the ability to pay for the testing. Some types of genetic testing are expensive and are not covered by insurance benefits.
Anxiety and altered family relationships are often the result of genetic testing; however, they are not the primary risk.
False-negative results are more likely to affect an individual or family because they will fail to seek necessary follow-up.
Caucasian middle-class families have greater access to genetic screening, so this is less of a risk for this population.
DIF: Cognitive Level: Comprehension REF: 47
OBJ: Client Needs: Health Promotion and Maintenance

TOP: Nursing Process: Implementation

7. A man's wife is pregnant for the third time. One child was born with cystic fibrosis, and the other child is healthy. The man wonders what the chance is that this child will have cystic fibrosis. This type of testing is known as:

a. Occurrence risk
b. Recurrence risk
c. Predictive testing
d. Predisposition testing

ANS: B
The couple already has a child with a genetic disease; therefore, they will be given a recurrence risk test.
If a couple has not yet had children but are known to be at risk for having children with a genetic disease, they are given an occurrence risk test. This couple already has a child with a genetic disorder.
Predictive testing is used to clarify the genetic status of an asymptomatic family member.
Predisposition testing differs from presymptomatic testing in that a positive result does not indicate 100% risk of a condition developing.

DIF: Cognitive Level: Comprehension REF: 56
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Planning

8. Cancer is genetic and begins with one or more genetic mutations. A cancer specifically being investigated in this regard is:

a. Lung cancer
b. Liver cancer
c. Colorectal cancer
d. Oral cancer

ANS: C
Colorectal cancer usually results from one or more predisposing genes and is the third leading cause of cancer deaths in women.
Tobacco smoke is a known causative factor for lung cancer. There may also be an acquired mutation of an oncogene present.
Liver cancer is not being investigated in this regard.
Oral cancer may be caused by an inherited mutation of one or more oncogenes.

DIF: Cognitive Level: Knowledge REF: 55
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

9. In practical terms regarding genetic health care, nurses should be aware that:

a. Genetic disorders affect equally people of all socioeconomic backgrounds, races, and ethnic groups
b. Genetic health care is more concerned with populations than individuals
c. The most important of all nursing functions is providing emotional support to the family during counseling
d. Taking genetic histories is the province of large universities and medical centers

ANS: C
Nurses should be prepared to help with a variety of stress reactions from a couple facing the possibility of a genetic disorder.
Although anyone may have a genetic disorder, certain disorders appear more often in certain ethnic and racial groups.
Genetic health care is highly individualized because treatments are based on the phenotypic responses of the individual.
Individual nurses at any facility can take a genetic history, although larger facilities may have better support services.

DIF: Cognitive Level: Comprehension REF: 57
OBJ: Client Needs: Psychosocial Integrity
TOP: Nursing Process: Planning

10. The Human Genome Project, which began in 1990:
a. Expected to complete a map of the entire human genome by 2010
b. Found that all human beings are 99.9% identical at the DNA level
c. Has not yet been able to translate the accumulating raw research into anything medically practical
d. Put its research relevant to nursing in a document published by the International Society of Nurses in Genetics (ISONG)

ANS: B
The majority of the 0.1% genetic variations are found within and not among populations.
The project completed its map ahead of schedule, in 2000.
The project's research has been very valuable in the identification of genes involved in disease and in the development of genetic tests.
The ISONG document, the "Statement of the Scope and Standards of Genetics Clinical Nursing Practice," is very useful but not directly related to the Human Genome Project.

DIF: Cognitive Level: Knowledge REF: 44
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

11. With regard to prenatal genetic testing, nurses should be aware that:

a. Maternal serum screening can determine whether a pregnant woman is at risk of carrying a fetus with Down syndrome
b. Carrier screening tests look for gene mutations of people already showing symptoms of a disease
c. Predisposition testing predicts with near certainty that symptoms will appear
d. Presymptomatic testing is used to predict the likelihood of breast cancer

ANS: A
Maternal serum screening identifies the risk for the neural tube defect and the specific chromosome abnormality involved in Down syndrome.
Carriers of some diseases, such as sickle cell disease, do not display symptoms.
Predisposition testing determines susceptibility, such as for breast cancer.
Presymptomatic testing indicates that if the gene is present, symptoms are certain to appear.

DIF: Cognitive Level: Knowledge REF: 45
OBJ: Client Needs: Physiologic Integrity
TOP: Nursing Process: Planning

12. A nurse must be cognizant that an individual's genetic makeup is known as his/her:
a. Genotype
b. Phenotype
c. Karyotype
d. Chromotype

ANS: A
The genotype comprises all the genes the individual can pass on to a future generation.
The phenotype is the observable expression of an individual's genotype.
The karyotype is a pictorial analysis of the number, form, and size of an individual's chromosomes.
Genotype refers to an individual's genetic makeup.

DIF: Cognitive Level: Knowledge REF: 47
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

13. The U.S. Department of Health and Human Services has designated Thanksgiving Day as National Family History Day. The Surgeon General encourages family members to discuss important family health information while sharing in holiday gatherings. This initiative is significant to nurses because:
a. There are few genetic tests available to identify this information
b. Only physicians should obtain this detailed information
c. Clients cannot accurately complete these histories on their own
d. This is the single most cost effective source of genetic information

ANS: D

Although there are more than 1000 genetic tests available, the single most cost-effective piece of genetic information is the family history.
Although there are more than 1000 genetic tests available, the single most cost-effective piece of genetic information is the family history.
Nurses are ideally suited to take the lead in ongoing efforts to recognize the significance of the family history as an important source of genetic information.
A computerized tool called *My Family Health Portrait* is available free of charge (www.hhs.gov/familyhistory/download.html). Other tools to aid the lay community in completing their family histories are available to the public.

DIF: Cognitive Level: Comprehension REF: 45
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Assessment

MULTIPLE RESPONSE

1. Which congenital malformations result from multifactorial inheritance? Choose all that apply.

a. Cleft lip
b. Congenital heart disease
c. Cri du chat syndrome
d. Anencephaly
e. Pyloric stenosis

ANS: A, B, D, E
Cleft lip, congenital heart disease, anencephaly, and pyloric stenosis are associated with multifactorial inheritance.
Cri du chat syndrome is related to a chromosomal deletion.

DIF: Cognitive Level: Knowledge REF: 52
OBJ: Client Needs: Psychosocial Integrity
TOP: Nursing Process: Diagnosis

2. Which activities are included in the role of a nurse practicing in the field of genetics? Choose all that apply.

a. Assessing the responses of family members to a genetic disorder
b. Performing genetic testing, such as amniocentesis
c. Constructing a family pedigree of three or more generations
d. Advising a pregnant mother whose fetus has a genetic disorder to have an abortion
e. Offering parents information about genetics

ANS: A, C, E
Assessing the responses of family members, constructing a family pedigree, and offering parents information about genetics are activities that a genetics nurse would carry out in caring for a family undergoing genetic counseling.

Physicians perform amniocentesis; the nurse may assist in this procedure. It is important for nurses to be aware of their own values and beliefs and to refrain from attempting to influence the family. The nurse must respect the right of the individual or family to make autonomous decisions.

DIF: Cognitive Level: Comprehension REF: 44
OBJ: Client Needs: Psychosocial Integrity
TOP: Nursing Process: Planning

COMPLETION

1. The karyotype designated as female is _____.

ANS: 46 XX

DIF: Cognitive Level: Comprehension REF: 48
OBJ: Client Needs: Psychosocial Integrity
TOP: Nursing Process: Diagnosis

TRUE/FALSE

1. BRCA1 and BRCA2 mutations account for approximately 50% of hereditary breast and ovarian cancer. Is this statement true or false?

ANS: F
These mutations account for about 70% to 85% of hereditary breast and ovarian cancers. The mutation is inherited as an autosomal dominant pattern; thus each offspring of an individual found to carry a BRCA mutation has a 50% chance of inheriting the same mutation. According to estimations of lifetime risk, 12% of women in the general population will develop breast cancer compared with about 60% of women who have inherited a deleterious mutation in their BRCA genes.

DIF: Cognitive Level: Knowledge REF: 55
OBJ: Client Needs: Health Promotion and Maintenance
TOP: Nursing Process: Planning