Pathophysiology: Concepts of Human Disease **(Sorenson)**
Chapter 2 Genetics, Genomics, and Epigenomics

1) The community health nurse is planning a program for a local health fair on genetics and health, using Healthy People 2020 goals. Which concepts should guide the nurse's planning for the program?
A) People should know their family health history.
B) All people should have genetic testing done.
C) Genetic testing determines what diseases a person will develop.
D) All diseases have a genetic basis.
Answer: A
Explanation: A) Research substantiates the benefits of taking a family health history and the use of genetic and genomic tests to identify risk factors, diagnose disease, and guide the use of screening tests and treatment options.
B) Research substantiates the benefits of taking a family health history and the use of genetic and genomic tests to identify risk factors, diagnose disease, and guide the use of screening tests and treatment options. If a family history does not suggest the presence of genomic and genomic risk factors, genetic testing is not indicated.
C) Research substantiates the benefits of taking a family health history and the use of genetic and genomic tests to identify risk factors, diagnose disease, and guide the use of screening tests and treatment options. Having a risk factor for a disease does not mean that the person will develop the disease.
D) Although not all diseases have a genetic basis, genomics plays a role in 9 out of 10 of the leading causes of death, including heart disease, cancer, stroke, diabetes, and Alzheimer's disease. Genomics also plays a role in a variety of neuromuscular, immune, vision, and hearing disorders.
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Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Planning | Learning Outcome: 2.1 Describe the impact of genetics, genomics, and epigenomics on personalized healthcare, and explain the concepts related to them. | QSEN Competencies: I.C.10 Value active partnership with patients or designated surrogates in planning, implementation, and evaluation of care | AACN Essential Competencies: VII.1 Assess protective and predictive factors, including genetics, which influence the health of individuals, families, groups, communities and populations
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 1: Recognize the concepts and relationships of human genetics, genomics, and epigenomics.

2) Which statement would be most appropriate for the nurse to make to a patient newly diagnosed with colorectal cancer?
A) "There is no need for genetic testing as you have already been diagnosed with colorectal cancer."
B) "With genetic testing, family members will not need colonoscopies."
C) "Genetic testing will help identify familial colorectal cancer syndromes."
D) "Family members will be able to determine if they will get colon cancer."
Answer: C
Explanation: A) One objective of Healthy People 2020 related to genomics is to "increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify familial colorectal cancer syndromes." One purpose of this objective is to increase the early diagnosis and treatment of asymptomatic individuals with colorectal cancer, thereby improving survival. Another purpose is to identify individuals at risk who need more frequent screening, such as colonoscopy.
B) One objective of Healthy People 2020 related to genomics is to "increase the proportion of persons with newly diagnosed colorectal cancer who receive genetic testing to identify familial colorectal cancer syndromes." One purpose of this objective is to increase the early diagnosis and treatment of asymptomatic individuals with colorectal cancer, thereby improving survival. Another purpose is to identify individuals at risk who need more frequent screening, such as colonoscopy.
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Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.1 Describe the impact of genetics, genomics, and epigenomics on personalized healthcare, and explain the concepts related to them. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

3) Which concept does the nurse use when explaining the inheritance of genetic disorders to a couple beginning to think about starting a family?
A) Humans have 46 pairs of chromosomes.
B) Egg and sperm cells each have 23 pairs of chromosomes.
C) Males have two X chromosomes.
D) A person inherits one chromosome from a chromosome pair from each parent.
Answer: D
Explanation: A) Humans have a total of 46 chromosomes in most cells of the body, which are grouped in 23 pairs. Think of them as 23 pairs of shoes, with a total of 46 shoes.
B) Humans have a total of 46 chromosomes in most cells of the body, which are grouped in 23 pairs. Think of them as 23 pairs of shoes, with a total of 46 shoes. For each pair of chromosomes, an individual inherits one chromosome from each parent.
C) The 23rd pair are the sex chromosomes and are designated either X or Y. Individuals with two X chromosomes are female, and those with an X chromosome and a Y chromosome are male.
D) Humans have a total of 46 chromosomes in most cells of the body, which are grouped in 23 pairs. Think of them as 23 pairs of shoes, with a total of 46 shoes. For each pair of chromosomes, an individual inherits one chromosome from each parent.
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Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.2 Identify components of the genetic code and the organization of genes on chromosomes. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX.7. Provide appropriate patient teaching that reflects developmental stage, age, culture, spirituality, patient preferences, and health literacy considerations to foster patient engagement in their care
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 2: Consider gene replication, transcription, translation and their impact on health.

4) The nurse involved in genomics engages in which of the following activities?
A) Studying the entire genome.
B) Studying individual genes.
C) Studying mutations.
D) Studying chemicals that instruct the genome.
Answer: A
Explanation: A) Genomics is a field of science that examines mechanisms of health and disease by studying the entire genome versus individual genes.
B) Genomics is a field of science that examines mechanisms of health and disease by studying the entire genome versus individual genes.
C) Genomics is a field of science that examines mechanisms of health and disease by studying the entire genome versus individual genes. It is not the study of gene mutations.
D) Genomics is a field of science that examines mechanisms of health and disease by studying the entire genome versus individual genes. It is not the study of the chemical compounds that instruct the genome where and when genes are expressed.
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Cognitive Level: Understanding
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Assessment | Learning Outcome: 2.3 Understand the differences between the human genome and epigenome and the mechanisms by which epigenetic modifications occur and affect gene expression. | QSEN Competencies: III.A.1. Demonstrate knowledge of basic scientific methods and processes | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 1: Recognize the concepts and relationships of human genetics, genomics, and epigenomics.

5) While taking a family history and drawing a genetic pedigree, the nurse explains to the patient that:
A) individuals in the same generation are arranged vertically.
B) offspring are in the row below the parents.
C) individuals who have had children together are connected by a vertical line.
D) circles represent males.
Answer: B
Explanation: A) In clinical practice, it is often useful to draw a genetic pedigree through several generations so that the pattern of genetic disease can be visualized. Individuals of the same generation are on one row, and their offspring are drawn in a row below, connected by a vertical line to their parents.
B) In clinical practice, it is often useful to draw a genetic pedigree through several generations so that the pattern of genetic disease can be visualized. Individuals of the same generation are on one row, and their offspring are drawn in a row below, connected by a vertical line to their parents.
C) Individuals who have had children together are linked by a horizontal line.
D) In a pedigree, circles represent women, and squares represent men.
Page Ref: 35
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

6) The nurse is answering nursing students' questions about the process of meiosis and mitosis. Which statement by a student should the nurse correct?
A) "In mitosis, cells divide to form an identical cell."
B) "Gonadal cells contain 23 chromosomes."
C) "Sperm and egg cells are diploid cells."
D) "Haploid cells have 23 chromosomes."
Answer: C
Explanation: A) The process of cell division used to create identical copies of a cell is called mitosis.
B) Gonadal cells, that is egg and sperm cells, have only half of the genetic information (one of each chromosome pair), or 23 chromosomes total.
C) In meiosis, egg and sperm cells are created for reproduction. Meiosis begins with a cell that has 46 total chromosomes, two of each chromosome type. However, the goal is to place one of every chromosome pair into a cell. This occurs by following DNA replication with two sets of cell divisions, and the end product is four daughter cells, each with a total of 23 chromosomes, one of each chromosome pair. These are called haploid cells.
D) In meiosis, egg and sperm cells are created for reproduction. Meiosis begins with a cell that has 46 total chromosomes, two of each chromosome type. However, the goal is to place one of every chromosome pair into a cell. This occurs by following DNA replication with two sets of cell divisions, and the end product is four daughter cells, each with a total of 23 chromosomes, one of each chromosome pair. These are called haploid cells.
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Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Evaluation | Learning Outcome: 2.4 Explain the function and sequence of events involved in gene replication during mitosis and meiosis, transcription, and translation. | QSEN Competencies: III.A.1. Demonstrate knowledge of basic scientific methods and processes | AACN Essential Competencies: VIII.13. Articulate the value of pursuing practice excellence, lifelong learning and professional engagement to foster professional growth and development
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 2: Consider gene replication, transcription, translation and their impact on health.

7) The nurse explains to the parents of a child with Prader-Willi syndrome that this disease is caused by:
A) genetic imprinting.
B) mosaicism.
C) single-nucleotide polymorphism (SNP).
D) missense mutation.
Answer: A
Explanation: A) In genetic imprinting, some genes have differential expression based on the parent from whom the genes are inherited. For example, a deletion of a portion of chromosome 15 leads to a disorder called Prader-Willi syndrome when inherited from the father, but the same gene deletion leads to a different condition called Angelman syndrome when inherited from the mother.
B) Nondisjunction can occur during meiosis (creation of sperm and egg cells). This can also occur shortly after conception, causing the presence of more than one genetic cell line in a person, a condition called mosaicism. The clinical effects of the mosaicism often depend on how many abnormal cells are present, but this is not the cause of Prader-Willi syndrome.
C) Not all genetic changes among individuals are pathogenic. When present in at least 1% of the population, a change is referred to as a single-nucleotide polymorphism (SNP). Some polymorphisms may just result in human variation with no clinical impact at all; others may contribute to disease, but this is not the cause of Prader-Willi syndrome.
D) A missense mutation causes a change in the amino acid sequence, but this is not the cause of Prader-Willi syndrome.
Page Ref: 33-34
Cognitive Level: Applying
Client Need & Sub: Physiological Integrity: Physiological Adaptation
Standards: Nursing Process: Implementation | Learning Outcome: 2.5 Compare single-nucleotide polymorphisms and the various types of gene mutations, including point mutations, insertions, deletions, and translocations, in regard to their characteristics and possible clinical consequences. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

8) Which statement indicates that a pregnant woman requires more teaching about prenatal screening?
A) "Screening tests assess a woman's risk of having a baby with chromosome problems."
B) "Amniocentesis obtains fetal cells to assess chromosome number and structure."
C) "Chorionic villus sampling can diagnose chromosome problems."
D) "Screening tests can diagnose all chromosome abnormalities."
Answer: D
Explanation: A) Prenatal screening can assess whether the woman is at a higher risk of having a baby with certain chromosomal problems. This can be done with ultrasound and maternal serum screening by blood.
B) Chorionic villus sampling and amniocentesis obtain cells from the growing fetus to assess fetal chromosome number and structure.
C) While screening tests can assess risk of certain chromosome problems, diagnosis can occur only with the use of invasive procedures, such as chorionic villus sampling or amniocentesis.
D) While screening tests can assess risk of certain chromosome problems, diagnosis can occur only with the use of invasive procedures, such as chorionic villus sampling or amniocentesis.
Page Ref: 34
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Evaluation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 4: Recognize diagnostic tests, genetic- and genome-based therapies, and future advances in the provision of personalized health care.

9) Which manifestation would the nurse expect to find in a child with Down syndrome?
A) Downward slanting eyes with an epicanthal fold.
B) A prominent brow and nose
C) Wide, short hands and fingers
D) Increased muscle tone and lack of flexibility
Answer: C
Explanation: A) The physical manifestations of Down syndrome are decreased or poor muscle tone; short neck; flattened facial profile and nose; small head, ears, and mouth; upward slanting eyes, often with epicanthal fold; wide, short hands and fingers; single deep crease across the palm; and deep groove between first and second toes.
B) The physical manifestations of Down syndrome are decreased or poor muscle tone; short neck; flattened facial profile and nose; small head, ears, and mouth; upward slanting eyes, often with epicanthal fold; wide, short hands and fingers; single deep crease across the palm; and deep groove between first and second toes.
C) The physical manifestations of Down syndrome are decreased or poor muscle tone; short neck; flattened facial profile and nose; small head, ears, and mouth; upward slanting eyes, often with epicanthal fold; wide, short hands and fingers; single deep crease across the palm; and deep groove between first and second toes.
D) The physical manifestations of Down syndrome are decreased or poor muscle tone; short neck; flattened facial profile and nose; small head, ears, and mouth; upward slanting eyes, often with epicanthal fold; wide, short hands and fingers; single deep crease across the palm; and deep groove between first and second toes.
Page Ref: 35
Cognitive Level: Applying
Client Need & Sub: Physiological Integrity: Physiological Adaptation
Standards: Nursing Process: Assessment | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: III.A.1. Demonstrate knowledge of basic scientific methods and processes | AACN Essential Competencies: IX. 1. 1. Conduct comprehensive and focused physical, behavioral, psychological, spiritual, socioeconomic, and environmental assessments of health and illness parameters in patients, using developmentally and culturally appropriate approaches
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

10) When providing genetic counseling, the nurse explains that the observable clinical expression of genetic coding is called:
A) phenotype.
B) genotype.
C) variable expressivity.
D) reduced penetrance.
Answer: A
Explanation: A) Genotype is the actual genetic code in a person, whereas phenotype is the clinical expression related to the genotype.
B) Genotype is the actual genetic code in a person, whereas phenotype is the clinical expression related to the genotype.
C) The clinical phenotype in individuals with mutations in the same gene can be different. This is called variable expressivity. Reduced penetrance means that not everyone who inherits a mutation will have clinical symptoms.
D) The clinical phenotype in individuals with mutations in the same gene can be different. This is called variable expressivity. Reduced penetrance means that not everyone who inherits a mutation will have clinical symptoms.
Page Ref: 35
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX.7. Provide appropriate patient teaching that reflects developmental stage, age, culture, spirituality, patient preferences, and health literacy considerations to foster patient engagement in their care
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

11) Which statement indicates that an individual with an autosomal recessive disorder understands its pattern of inheritance?
A) A carrier will not express the trait.
B) The trait will be expressed with one copy of the gene.
C) The affected gene is on the sex chromosome.
D) More females are affected than males
Answer: A
Explanation: A) The word *autosomal* means that the gene of interest is on one of the non-sex chromosomes. Thus, males or females can be affected. Recessive inheritance indicates that both copies of a gene must not be working to express the trait. A person who inherits only one copy of a mutated gene will be a carrier but will not express the trait.
B) The word *autosomal* means that the gene of interest is on one of the non-sex chromosomes. Thus, males or females can be affected. Recessive inheritance indicates that both copies of a gene must not be working to express the trait. A person who inherits only one copy of a mutated gene will be a carrier but will not express the trait.
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Page Ref: 35
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Evaluation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
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MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

12) The nurse explains to parents that if both of them are carriers of an autosomal recessive genetic mutation, the chance of each child inheriting the disease is:
A) 0%.
B) 25%.
C) 50%.
D) 100%.
Answer: B
Explanation: A) When both parents are carriers of an autosomal recessive gene mutation, there is a 25% chance in each pregnancy that the child will inherit two mutated genes and thus have the disease.
B) When both parents are carriers of an autosomal recessive gene mutation, there is a 25% chance in each pregnancy that the child will inherit two mutated genes and thus have the disease.
C) When both parents are carriers of an autosomal recessive gene mutation, there is a 25% chance in each pregnancy that the child will inherit two mutated genes and thus have the disease.
D) When both parents are carriers of an autosomal recessive gene mutation, there is a 25% chance in each pregnancy that the child will inherit two mutated genes and thus have the disease.
Page Ref: 35
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

13) How should the nurse respond when the parents of a child with cystic fibrosis ask how their male child inherited this autosomal recessive disorder when neither parent has the disease?
A) "Two carriers may pass on the mutation to their child."
B) "The disease often skips a generation."
C) "The gene for this disorder is on the Y chromosome."
D) "Are you sure there is no history of cystic fibrosis in either family?"
Answer: A
Explanation: A) Often in an autosomal recessive condition, such as cystic fibrosis, there is not a family history of the condition. The disease occurs only when two carriers have a child together, and both carriers pass on the mutation to that child.
B) Often in an autosomal recessive condition, such as cystic fibrosis, there is not a family history of the condition. The disease occurs only when two carriers have a child together, and both carriers pass on the mutation to that child. The disease does not skip a generation.
C) The disorder is not sex-linked.
D) Often in an autosomal recessive condition, such as cystic fibrosis, there is not a family history of the condition. The disease occurs only when two carriers have a child together, and both carriers pass on the mutation to that child.
Page Ref: 36
Cognitive Level: Analyzing
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

14) When counseling parents about genetic transmission of an X-linked disorder, which concept does the nurse keep in mind?
A) A male can pass an X-linked mutation to a son or a daughter.
B) A female can only pass an X-liked disorder to a son.
C) A male with a mutation in an X-linked gene will manifest the disease.
D) An X-linked disorder can be traced through the paternal lineage.
Answer: C
Explanation: A) A female can pass an X-linked mutation to a son or daughter, but only the son will manifest the severe form of the disease. A father cannot pass on the X-linked condition to his sons because a male inherits his X chromosome from his mother and the Y chromosome from his father. Therefore, in an X-linked condition, it is likely that only males will be affected, and the condition can be traced through the maternal lineage.
B) A female can pass an X-linked mutation to a son or daughter, but only the son will manifest the severe form of the disease. A father cannot pass on the X-linked condition to his sons because a male inherits his X chromosome from his mother and the Y chromosome from his father. Therefore, in an X-linked condition, it is likely that only males will be affected, and the condition can be traced through the maternal lineage.
C) A female can pass an X-linked mutation to a son or daughter, but only the son will manifest the severe form of the disease. A father cannot pass on the X-linked condition to his sons because a male inherits his X chromosome from his mother and the Y chromosome from his father. Therefore, in an X-linked condition, it is likely that only males will be affected, and the condition can be traced through the maternal lineage.
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Page Ref: 36
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Planning | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

15) Which statement by a patient indicates an understanding of why phenotypic variations in disease occur?
A) "Disease presentation is the same in all people with the same genetic mutations."
B) "Disease presentation only relies on lifestyle choices."
C) "Disease presentation is only due to environmental influences."
D) "Disease presentation usually depends on multiple genes, environment, and lifestyle."
Answer: D
Explanation: A) Manifesting multifactorial disease depends on multiple genes, environmental influences, and lifestyle choices. Therefore, significant variation in disease presentation exists. However, single-gene disorders can also have quite a bit of variation in clinical presentation, owing to factors such as reduced penetrance, as in familial cancer syndromes, and variable expressivity.
B) Manifesting multifactorial disease depends on multiple genes, environmental influences, and lifestyle choices. Therefore, significant variation in disease presentation exists. However, single-gene disorders can also have quite a bit of variation in clinical presentation, owing to factors such as reduced penetrance, as in familial cancer syndromes, and variable expressivity.
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Page Ref: 38
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Evaluation | Learning Outcome: 2.7 Differentiate the mechanisms responsible for phenotypic variations in human disease. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX.9. Monitor client outcomes to evaluate the effectiveness of psychobiological interventions
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.

16) The plan of care for a woman newly diagnosed with a BRCA1 mutation should include which strategy?
A) Preoperative planning for immediate prophylactic double mastectomies
B) A recommendation for enhanced screening for breast and ovarian cancer
C) Reassurance that male offspring with BRCA1 do not develop breast cancer
D) Reassurance that the BRCA1 gene protects against ovarian cancer
Answer: B
Explanation: A) Up to 5% of all cases of breast cancer are due to autosomal dominant inheritance of a mutation in either the BRCA1 or BRCA2 gene. Having a BRCA1 or BRCA2 mutation confers a very high lifetime risk of both breast cancer and ovarian cancer. Even males with a BRCA1 or BRCA2 gene mutation have an increased risk of breast cancer. Early identification of a gene mutation may allow for enhanced screening and some prophylactic options for early detection and/or decreasing the risk of cancer. The choice of prophylactic mastectomy is very personal and should be made by the woman in conjunction with her family and healthcare team.
B) Up to 5% of all cases of breast cancer are due to autosomal dominant inheritance of a mutation in either the BRCA1 or BRCA2 gene. Having a BRCA1 or BRCA2 mutation confers a very high lifetime risk of both breast cancer and ovarian cancer. Even males with a BRCA1 or BRCA2 gene mutation have an increased risk of breast cancer. Early identification of a gene mutation may allow for enhanced screening and some prophylactic options for early detection and/or decreasing the risk of cancer.
C) Up to 5% of all cases of breast cancer are due to autosomal dominant inheritance of a mutation in either the BRCA1 or BRCA2 gene. Having a BRCA1 or BRCA2 mutation confers a very high lifetime risk of both breast cancer and ovarian cancer. Even males with a BRCA1 or BRCA2 gene mutation have an increased risk of breast cancer. Early identification of a gene mutation may allow for enhanced screening and some prophylactic options for early detection and/or decreasing the risk of cancer.
D) Up to 5% of all cases of breast cancer are due to autosomal dominant inheritance of a mutation in either the BRCA1 or BRCA2 gene. Having a BRCA1 or BRCA2 mutation confers a very high lifetime risk of both breast cancer and ovarian cancer. Even males with a BRCA1 or BRCA2 gene mutation have an increased risk of breast cancer. Early identification of a gene mutation may allow for enhanced screening and some prophylactic options for early detection and/or decreasing the risk of cancer.
Page Ref: 39
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Planning | Learning Outcome: 2.8 Understand the role of genetic and epigenomic factors in cancer. | QSEN Competencies: I.C.10 Value active partnership with patients or designated surrogates in planning, implementation, and evaluation of care | AACN Essential Competencies: IX.3. Implement holistic, patient-centered care that reflects an understanding of human growth and development, pathophysiology, pharmacology, medical management and nursing management across the health-illness continuum, across lifespan, and in all healthcare settings
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 4: Recognize diagnostic tests, genetic- and genome-based therapies, and future advances in the provision of personalized health care.

17) The nurse explains to a patient undergoing karyotyping that this test will:
A) examine nucleotide changes in a gene.
B) detect small deletions or structural abnormalities of the chromosomes and DNA.
C) examine the visual appearance of chromosome structure and number.
D) detect single gene mutations.
Answer: C
Explanation: A) Single-gene sequencing is designed to detect nucleotide changes anywhere in the gene.
B) Fluorescence in situ hybridization (FISH) has been utilized both for rapid detection of chromosome number and for targeting specific DNA sequences. This test can detect small deletions or structural abnormalities that are not seen in standard karyotyping.
C) Karyotyping is a test used to examine the visual appearance of chromosome structure and number. This type of genetic testing can identify aneuploidy and triploidy as well as translocations and other gross chromosomal structural abnormalities. However, it cannot detect single-gene mutations.
D) Single-gene sequencing is designed to detect nucleotide changes anywhere in the gene.
Page Ref: 39
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.9 Apply the following tests to the appropriate clinical situation: karyotyping, fluorescence in situ hybridization, single-gene testing, and genome-wide association studies. | QSEN Competencies: I.B.10 Engage patients or designated surrogates in active partnerships that promote health, safety and well-being, and self-care management | AACN Essential Competencies: IX.7. Provide appropriate patient teaching that reflects developmental stage, age, culture, spirituality, patient preferences, and health literacy considerations to foster patient engagement in their care
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 4: Recognize diagnostic tests, genetic- and genome-based therapies, and future advances in the provision of personalized health care.

18) The nurse explains to a patient with chronic myelogenous leukemia that the therapeutic action of Gleevec (Imatinib) is to:
A) bind directly DNA or RNA to block the aberrant cancer-causing gene product.
B) silence genes that are implicated in causing the growth of cancer causing cells.
C) selectively increase transcription levels of certain genes.
D) promote the replication of healthy genetic material.
Answer: A
Explanation: A) Cancer treatments have been developed in which an antisense oligonucleotide (sequence of complementary nucleotides) is used to directly bind the DNA or RNA to block the aberrant gene product. Gleevec (Imatinib) is the most publicized example of this approach in the treatment of chronic myelogenous leukemia, which is caused by the BCR-ABL fusion gene.
B) Gene replacement therapy works by expressing or silencing genes that are implicated in disease processes. DNA or RNA is engineered to be delivered (often via virus) into an individual so that target cells take up the gene and then express its protein product.
C) Transcription factor modulators selectively increase or decrease transcription levels of certain genes. Not being cell specific, transcription modulation must be used carefully, considering the potential side effects, but these may be limited in the future by cell-specific targeting.
D) Gene replacement therapy works by expressing or silencing genes that are implicated in disease processes. DNA or RNA is engineered to be delivered (often via virus) into an individual so that target cells take up the gene and then express its protein product. It does not promote the replication of healthy genetic material.
Page Ref: 40-41
Cognitive Level: Applying
Client Need & Sub: Physiological Integrity: Pharmacological and Parenteral Therapies
Standards: Nursing Process: Implementation | Learning Outcome: 2.10 Apply the following genetic-based therapies to appropriate clinical situations: gene replacement therapy, pharmacogenomics, antisense nucleotides, and transcription factor | QSEN Competencies: III.A.1. Demonstrate knowledge of basic scientific methods and processes | AACN Essential Competencies: IX.7. Provide appropriate patient teaching that reflects developmental stage, age, culture, spirituality, patient preferences, and health literacy considerations to foster patient engagement in their care
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 4: Recognize diagnostic tests, genetic- and genome-based therapies, and future advances in the provision of personalized health care.

19) When teaching a genetics class to nursing students in a baccalaureate nursing program, the nurse educator explains that the International HapMap Project:
A) studies genomic elements to determine commonalities in the human genome.
B) explores the genetic basis for diseases around the world.
C) studies the human genome within a population.
D) analyzes genetic elements to determine what makes people different from one another.
Answer: D
Explanation: A) With data and technologies generated from the Human Genome Project, a worldwide collaborative study of different ancestries to develop a haplotype map of the human genome, called the International HapMap Project, was undertaken. The Human Genome Project revealed that humans are genetically 99.9% identical, and the International Hap-Map Project focuses on analyzing the 0.1% of the genomic elements that make us different from one another. Data from the International HapMap Project revealed 3.1 million common single nucleotide polymorphisms in the human genome across geographically diverse populations.
B) With data and technologies generated from the Human Genome Project, a worldwide collaborative study of different ancestries to develop a haplotype map of the human genome, called the International HapMap Project, was undertaken. The Human Genome Project revealed that humans are genetically 99.9% identical, and the International Hap-Map Project focuses on analyzing the 0.1% of the genomic elements that make us different from one another. Data from the International HapMap Project revealed 3.1 million common single nucleotide polymorphisms in the human genome across geographically diverse populations.
C) With data and technologies generated from the Human Genome Project, a worldwide collaborative study of different ancestries to develop a haplotype map of the human genome, called the International HapMap Project, was undertaken. The Human Genome Project revealed that humans are genetically 99.9% identical, and the International Hap-Map Project focuses on analyzing the 0.1% of the genomic elements that make us different from one another. Data from the International HapMap Project revealed 3.1 million common single nucleotide polymorphisms in the human genome across geographically diverse populations.
D) With data and technologies generated from the Human Genome Project, a worldwide collaborative study of different ancestries to develop a haplotype map of the human genome, called the International HapMap Project, was undertaken. The Human Genome Project revealed that humans are genetically 99.9% identical, and the International Hap-Map Project focuses on analyzing the 0.1% of the genomic elements that make us different from one another. Data from the International HapMap Project revealed 3.1 million common single nucleotide polymorphisms in the human genome across geographically diverse populations.
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Cognitive Level: Understanding
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.11 Analyze the impact of advances in genomics and epigenomics on personalized healthcare. | QSEN Competencies: III.A.1. Demonstrate knowledge of basic scientific methods and processes | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care

MNL Learning Outcome: LO 4: Recognize diagnostic tests, genetic- and genome-based therapies, and future advances in the provision of personalized health care.

20) The nurse is explaining the inheritance of Huntington disease to a newly diagnosed patient whose partner does not have the gene mutation. Which statement should the nurse make regarding family planning?
A) There is a 0% chance with each pregnancy that the child will inherit the gene for Huntington disease.
B) There is a 25% chance with each pregnancy that the child will inherit the gene for Huntington disease.
C) There is a 50% chance with each pregnancy that the child will inherit the gene for Huntington disease.
D) There is a 100% chance with each pregnancy that the child will inherit the gene for Huntington disease.
Answer: C
Explanation: A) Dominant inheritance indicates that only one copy of a mutated gene is required to cause disease. When a parent has an autosomal dominant gene mutation, there is a 50% chance in each pregnancy that the child will inherit the mutation and manifest disease.
B) Dominant inheritance indicates that only one copy of a mutated gene is required to cause disease. When a parent has an autosomal dominant gene mutation, there is a 50% chance in each pregnancy that the child will inherit the mutation and manifest disease.
C) Dominant inheritance indicates that only one copy of a mutated gene is required to cause disease. When a parent has an autosomal dominant gene mutation, there is a 50% chance in each pregnancy that the child will inherit the mutation and manifest disease.
D) Dominant inheritance indicates that only one copy of a mutated gene is required to cause disease. When a parent has an autosomal dominant gene mutation, there is a 50% chance in each pregnancy that the child will inherit the mutation and manifest disease.
Page Ref: 36
Cognitive Level: Applying
Client Need & Sub: Health Promotion and Maintenance
Standards: Nursing Process: Implementation | Learning Outcome: 2.6 Compare the characteristics and patterns of inheritance of genetic disorders caused by abnormalities of chromosome number or structure to those caused by autosomal, X-linked, and mitochondrial disorders. | QSEN Competencies: III.A.1. Demonstrate knowledge of basic scientific methods and processes | AACN Essential Competencies: IX. 2. Recognize the relationship of genetics and genomics to health, prevention, screening, diagnostics, prognostics, selection of treatment, and monitoring of treatment effectiveness, using a constructed pedigree from collected family history information as well as standardized symbols and terminology
NLN Competencies: Knowledge and Science: Relationships between knowledge/science and quality and safe patient care
MNL Learning Outcome: LO 3: Examine genetic factors and disorders and how they affect individual health.