Test Bank for Essentials of Medical Genetics for Nursing and Health Professionals 1st McClary

Essentials of Medical Genetics for Nursing and Health Professionals: An Interprofessional Approach

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Test Bank

Chapter 1 – Introduction

1. A new patient comes to establish care at the family medicine clinic where you work. During your history taking, you create a diagram representing the familial relationships among the patient's relatives to analyze Mendelian inheritance of certain traits. What is this diagram called?
    a. Karyotype
    b. Genotype
    c. Pedigree
    d. Genome

    Answer: C
    Rationale: A diagram that represents relationships between family members is called a pedigree. Answer A is incorrect because a karyotype is the number and appearance of chromosomes within a cell. Answer B is incorrect because a genotype is the genetic makeup of a cell. Answer D is incorrect because a genome is the genetic material of an organism. Therefore, C is the correct answer choice.

2. You are counselling your patient with albinism, which is a homozygous recessive allele. She is worried about the chances of her future children being affected by this condition. Which of the following is a correct statement regarding your education of this patient?
    a. More males than females are affected.
    b. Affected individuals would not have affected offspring.
    c. Affected individuals typically have affected parents.
    d. Approximately 50% of siblings with the same parents are affected.

    Answer: B
    Rationale: Answer B is correct because her mating partner is likely homozygous for the normal (non-albino) allele, so her offspring would be heterozygous and therefore not affected. Answer A is incorrect because this is the description of an X-linked recessive condition; males are more affected because they only have one X chromosome and therefore require normal functioning of that chromosome. Answer C is incorrect because it contradicts the correct answer (B). Answer D is incorrect because all this patient's offspring are likely to be unaffected.

3. A karyotype halts mitosis in what stage?
    a. Prophase
    b. Metaphase
    c. Anaphase
    d. Telophase

    Answer: B

Rationale: During a karyotype, mitosis is stopped in metaphase, just as the chromosomes are aligned and most condensed. Answer A is incorrect because the chromosomes are just beginning to condense. Answers C and D are incorrect because sister chromatids have separated and are being pulled apart. Thus, B is the only correct answer.

4. What genetic test often uses the metaphase arrangement of chromosomes to analyze the lengths and positions of their centromeres?
    a. Cytogenetics
    b. Karyotyping
    c. Probing
    d. Fluorescence in situ hybridization (FISH)

    Answer: B
    Rationale: During a karyotype, mitosis is stopped in metaphase, just as the chromosomes are aligned and most condensed. Answer A is incorrect because cytogenetics is when light microscopy is used to analyze chromosomes. Answer C is incorrect because a probe is a small, fluorescent-dyed piece of nucleic acid that is similar to the gene being analyzed in fluorescence in situ hybridization (FISH). Answer D is incorrect because FISH is a diagnostic technique that uses fluorescence, or light emission, to analyze chromosomes with the use of a probe.

5. While counselling a couple regarding the likelihood that their child will inherit a certain genetic disorder, you explain that some genotypes do not always express their corresponding phenotype while other genotypes are always expressed. What concept are you explaining?
    a. Cytogenetics
    b. Expressivity
    c. Dominance
    d. Penetrance

    Answer: D
    Rationale: Penetrance is the probability to which a genotype will be phenotypically expressed. Traits with full penetrance will be expressed in all patients of that genotype. With incomplete penetrance, the disease is not expressed in all patients of that genotype. Answer A is incorrect because cytogenetics is using light microscopy to study chromosomes. Answer B is incorrect because variable expressivity refers to differences in the clinical or phenotypical presentation of a disease. Answer C is incorrect because dominance is the ability of a trait to manifest itself in heterozygous carriers. Thus, answer D is the correct choice.

6. You are a graduate research student studying HIV and need to create large numbers of the viral DNA to use in experiments. What process will you use to replicate it exponentially?

a. Polymerase chain reaction
b. Chromosome painting
c. Fluorescence in situ hybridization
d. Karyotyping

Answer: A
Rationale: Polymerase chain reaction (PCR) is a technique used to amplify DNA exponentially. Answer B is incorrect because chromosome painting is when chromosomes are painted difference colors to help identify pairs of homologous chromosomes. Answer C is incorrect because fluorescent in situ hybridization (FISH) is a diagnostic technique that uses fluorescence, or light emission, to analyze chromosomes with the use of a probe. Answer D is incorrect because a karyotype is the number and appearance of chromosomes within a cell. Therefore, answer A is the most appropriate choice.

7. You suspect your patient has an enzymatic defect. What type of testing is most appropriate at this time?
   a. Polymerase chain reaction
   b. Biochemical analysis
   c. Fluorescence in situ hybridization
   d. Karyotyping

   Answer: B
   Rationale: Biochemical analysis is the most appropriate choice because it is used to detect the presence of proteins. Answer A is incorrect because Polymerase chain reaction (PCR) is a technique used to amplify DNA exponentially. Answer C is incorrect because fluorescent in situ hybridization (FISH) is a diagnostic technique that uses fluorescence, or light emission, to analyze chromosomes with the use of a probe. Answer D is incorrect because a karyotype is the number and appearance of chromosomes within a cell. Thus, answer choice B is the best answer.

8. A 6-year-old female presents to your pediatrics office with facial tenderness, cough, purulent rhinorrhea, and fever. A brief review of her history reveals frequent antibiotic use for recurrent lung infections. You recommend biochemical analysis to examine for an enzymatic defect in chloride and water transport, also known as:
   a. Phenylketonuria
   b. Primary ciliary dyskinesia
   c. Primary immunodeficiency
   d. Cystic fibrosis

   Answer: D
   Rationale: Biochemical analysis is used for the detection of cystic fibrosis because it is used to detect the presence of proteins, namely enzymes. Answer A is incorrect because phenylketonuria (PKU) is an absence or deficit in phenylalanine hydroxylase, which

catabolizes phenylalanine. Answer B is incorrect because primary ciliary dyskinesia is a congenital condition where mucus clearance is impaired due to defective or absent cilia. Answer C is incorrect because primary immunodeficiency is a susceptibility to diseases due to poor antibody production. Thus, answer D is the correct choice.

9. The parents of a newborn baby girl were told that their neonate was screened for inborn errors of metabolism this morning. You educate the parents regarding what an inborn error of metabolism is and give examples, including phenylketonuria and
    a. Down Syndrome
    b. Cystic fibrosis
    c. Hemophilia
    d. Familial Hypercholesterolemia

    Answer: B
    Rationale: Answer A, answer C, and answer D are not examples of inborn errors of metabolism. Only the only choice that is an inborn error of metabolism is cystic fibrosis. Therefore, answer B is the best answer.

10. A small piece of nucleic acid that is labeled with fluorescent dye used to identify specific mutated genes in fluorescence in situ hybridization (FISH) is called a:
    a. Probe
    b. Homolog
    c. Karyotype
    d. Genome

    Answer: A
    Rationale: A probe matches a short part of the DNA being analyzed and is labeled with fluorescent dye to help potentially identify mutated genes. Answer B is incorrect because homologs are homologous chromosomes that pair up before separating during meiosis. Answer C is incorrect because a karyotype is the number and appearance of chromosomes within a cell. Answer D is incorrect because a genome is the genetic material of an organism. Thus, answer A is the best choice.