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

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***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.

# Chapter 2 – Diagnostic Techniques in Medical Genetics

1. You create a pedigree for a client who reports several family members experiencing the same genetic disease. Which aspect of the pedigree indicates that the disorder has complete penetrance?
    a. Two cousins have the same disorder.
    b. Males and females are affected equally.
    c. The disorder does not appear in one family.
    d. The disorder appears in the female parents and male children.

    Answer: B
    Rationale: Penetrance is the proportion of organisms having a particular genotype that actually expresses the corresponding phenotype. If the phenotype is always expressed, penetrance is complete; otherwise, it is incomplete. One characteristic of complete penetrance is both males and females being equally affected by the disorder. Cousins having the same disorder or the disorder missing an entire family does not demonstrate complete penetrance. A disorder appearing in females and male children a characteristic of an x-linked genetic disorder.

2. A young adult client reports having several genetic disorders in the immediate family and wants to know the chances of having a child with a disorder. Which specimen should you consider collecting in anticipation of having a chromosomal analysis completed for this client?
    a. Stool
    b. Urine
    c. Blood
    d. Saliva

    Answer: C
    Rationale: Chromosomal analysis is done by growing human cells in tissue culture, chemically inhibiting mitosis, and staining, observing, photographing, sorting, and counting the chromosomes. Samples can be obtained from peripheral blood, amniotic fluid, trophoblastic cells from the chorionic villus, bone marrow, and cultured fibroblasts, usually obtained from a skin biopsy. Stool, urine, or saliva samples are not used to complete a chromosomal analysis.

3. A chromosomal analysis is being completed for a client with a questionable genetic disorder. Which technique is used to identify chromosomal abnormalities?
    a. Treat with Giemsa stain
    b. Grow the cells in a tissue culture
    c. Use light microscopy to study the sample
    d. Identify the order of the chromosome pairs

Answer: A
Rationale: Treating chromosomes with Giemsa stain causes the chromosomes to exhibit transverse bands or G-bands that are specific for each pair of homologs. These G-bands allow smaller segments of each chromosome arm to be identified in order to identify chromosomal abnormalities. Growing the cells in a tissue culture and using light microscopy to study the sample is the process used in chromosomal analysis, in which the chromosomes are routinely rearranged systematically in pairs from longest to shortest and numbered from 1 through 22 to represent the autosomes.

4. A blood sample from a client having genetic testing is being tested through fluorescence in situ hybridization (FISH). What will this type of testing reveal about the client?
    a. Detect risk for fertility issues
    b. Validate the chromosomal pairs
    c. Identify individual gene mutations
    d. Predict if offspring will develop an abnormality

Answer: C
Rationale: Fluorescence in situ hybridization (FISH) makes it easier to visualize and map chromosomal and gene abnormalities. This technique does not identify the risk for fertility issues. It will not validate chromosomal pairs and does not predict if offspring will develop an abnormality.

5. A client is having genetic testing through fluorescence in situ hybridization (FISH). What is the purpose of the probe with this genetic test?
    a. Search for repeating DNA cycles
    b. Identify the longest chromosomes
    c. Validate the shortest chromosomes
    d. Search for genes with the same sequence

Answer: D
Rationale: The probe is a short sequence of nucleic acid that matches a portion of the gene in question. The probe is then allowed to hybridize to suitably prepared cells or histological sections; hybrids are formed with complementary sequences of nucleic acids in a chromosome. Through rough nucleic acid hybridization, the degree of sequence identity can be determined and specific sequences located on specific chromosomes detected. A probe does not search for repeating DNA cycles. This laboratory test does not identify the longest or validate the shortest chromosomes.

6. You review data collected from a client during a health history. For which reason should cytogenetic testing be considered for this client?
    a. Experiences seasonal eczema
    b. Older sibling spouse has twins

c. Parents alive and both in their 70s
d. Infertility with no physiologic cause

Answer: D
Rationale: There are specific situations in which cytogenetic testing is appropriate. One of these situations is infertility occurring without an obstetric or urogenital cause. Cytogenetic testing would not be indicated for a seasonal allergic health problem. Having both parents alive in their 70s is not a reason for cytogenetic testing. An older sibling's spouse having twins does not indicate a genetic anomaly.

7. A client having genetic testing is asked to provide a strand of hair. How will this sample be used for DNA analysis?
   a. Treat with Giemsa stain
   b. Create nucleic acid chains
   c. Analyze under a microscope
   d. Create a polymerase chain reaction

   Answer: D
   Rationale: In a DNA analysis, probes are used to identify specific genes that may be mutated in a certain hereditary disease. The probe may be a piece of the actual gene, a sequence close to the gene, or just a few nucleotides at the actual mutation. The closer the probe is to the actual mutation, the more accurate and the more useful the information. When even a minute amount of DNA from a patient such as a hair bulb is combined with the primers in a reaction mixture that replicates DNA—called polymerase chain reaction (PCR) —the region of DNA between the primers will be amplified exponentially after about a dozen cycles. Giemsa stain is used to identify chromosomal abnormalities. Fluorescence in situ hybridization (FISH) uses nucleic acid chains. DNA are not visible through microscopic analysis.

8. A client is scheduled for biochemical analysis to identify a genetic disorder. Which is the focus of this analysis?
   a. Genus of intestinal bacteria
   b. Amount of subcutaneous tissue
   c. Presence or absence of proteins
   d. Levels of hormones required for development

   Answer: C
   Rationale: The primary goal of biochemical testing is to determine whether certain proteins are present or absent. Biochemical testing also identifies the proteins' characteristics and effectiveness in vitro. Biochemical analysis does not focus on intestinal bacteria, amount of subcutaneous tissue, or hormone levels.

9. A 3-month old baby is diagnosed with cystic fibrosis. On which body systems should you prepare teaching for the parents about this disorder?
    a. Respiratory and digestion
    b. Musculoskeletal and renal
    c. Cardiovascular and lymphatics
    d. Neurologic and electrolyte balance

    Answer: A
    Rationale: Cystic fibrosis is caused by a gene that disrupts chloride and water transport, causing thick sticky mucus that blocks the airways in the lungs and pancreatic ducts. This leads to problems with breathing and digestion. Cystic fibrosis does not affect the musculoskeletal, renal, cardiovascular, lymphatic, neurologic, or electrolyte balance systems.

10. A client agrees to have genetic testing for a potential disorder. Which statement should you make so that the client understands the potential issues of having this kind of testing?
    a. "All of the test findings are confidential."
    b. "Genetic testing costs the same as other diagnostic testing."
    c. "Most genetic anomalies can be altered by lifestyle changes."
    d. "You will not be denied health insurance, but your premiums may increase."

    Answer: D
    Rationale: There are no laws preventing insurance rate spikes that may follow genetic Testing. Insurance companies cannot deny people coverage, but they can raise premiums. A family member may inadvertently learn of a genetic disorder through an inheritance pattern, which is a violation of the Health Insurance Portability and Accountability Act (HIPAA). Genetic testing can be very expensive and is not always conclusive. Once a client has a positive diagnosis, it cannot be changed. Diet, exercise, and lifestyle modifications can do nothing to change one's genome.