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Advanced Physiology and Pathophysiology: Essentials for Clinical Practice

Second Edition

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Chapter 2

Chemical and Biochemical Foundations

Multiple Choice Questions

1. Which of the following best describes why atoms have an overall neutral charge?

   a. Electrons orbit on the outside of the atom.

   @ Incorrect answer. Although it is true that electrons orbit outside the nucleus, this is not the reason why atoms have an overall neutral charge.

   *b. The number of protons is equal to the number of electrons.

   @ Correct answer. There is an equal number of protons, which have a positive charge, and electrons, which have a negative charge. The positive and negative charges balance one another out for an overall neutral charge.

   c. There are more neutrons than protons.

   @ Incorrect answer. There is an equal number of neutrons and protons. Additionally, neutrons have a neutral charge.

   d. There are four electrons in the outer shell.

   @ Incorrect answer. Some elements have four electrons in the outer shell, but this is not the reason for the overall neutral charge. The number of electrons will match the number of protons.

2. Why do hydrogen and potassium readily interact with other atoms?

   a. They only have electrons in the first shell.

   @ Incorrect answer. Hydrogen has one electron, but potassium has 19 electrons. Therefore, potassium has two electrons in the first shell, eight in the second and third shells, and one in the fourth shell. Hydrogen has one electron in the first shell.

b. They have electrons in all shells.

@ Incorrect answer. Hydrogen has one electron in the first shell. Potassium has two electrons in the first shell, eight in the second and third shells, and one in the fourth shell.

c. They both have four electrons in the outer shell.

@ Incorrect answer. Both hydrogen and potassium have one electron in their outer shells.

*d. They both have one electron in the outer shell.

@ Correct answer. Both hydrogen and potassium have one electron in their outer shells, meaning they can readily interact with other atoms.

3. How many electrons does carbon use to interact with other atoms?

*a. 4

@ Correct answer. Carbon has four electrons in its outer shell. Only the electrons in the outer shell, or valence electrons, are used to interact with other electrons.

b. 6

@ Incorrect answer. While carbon has a total of six electrons, only the electrons in the outer shell are used to interact with other electrons. Two of carbon's six electrons are in the first shell.

c. 1

@ Incorrect answer. Carbon has four valence electrons.

d. 2

@ Incorrect answer. While carbon has two electrons in the first shell, it also has four valence electrons that are used to interact with other elements.

4. Which of the following is true regarding the role of bicarbonate in maintaining homeostasis?

a. Bicarbonate dissociates from hydroxide to form a free hydroxide that neutralizes acid.

@ Incorrect answer. The dissociation of sodium from a hydroxide ion creates a free hydroxide that neutralizes acid.

b. Bicarbonate releases protons when diluted with water, which neutralizes acids.

@ Incorrect answer. Acids release protons when placed in water, but bicarbonate is a base.

c. Bicarbonate is partially ionized in water and is considered to be a weak base.

@ Incorrect answer. Acids are ionized in water, not bases like bicarbonate. Both carbonic acid and phosphoric acid are partly ionized in water and are considered weak acids.

*d. Bicarbonate functions as a buffer to neutralize acids.

@ Correct answer. Bicarbonate is produced in the pancreas for the purpose of neutralizing the acidity in chyme as it moves from the stomach to the small intestine. It also plays a major role in the carbonic acid buffer system.

5. Which of the following occurs when carbonic acid is placed in water?

a. Almost all of the protons are released into the water.

@ Incorrect answer. Carbonic acid is a weak acid. Only in strong acids do most of the protons dissociate into the water.

*b. Some of the protons are released into the water.

@ Correct answer. Carbonic acid is a weak acid, meaning it is only partly ionized in an aqueous solution. Some of the protons are released into the water.

c. Some of the electrons are released into the water.

@ Incorrect answer. When acids are placed in water, the protons are released. They are classified based on their tendency to lose protons. Carbonic acid is a weak acid because only a few of its protons are released into the water.

d. Almost all of the neutrons are released into the water.

@ Incorrect answer. Protons are released when acids are placed in water.

6. Which of the following most fully describes how proteins are used to buffer pH?

a. Proteins are used to neutralize excess acids because they have a carboxylic acid group.

@ Incorrect answer. The carboxylic acid group enables proteins to neutralize excess bases.

b. Proteins have an amino group, which is an acid, and therefore they can neutralize excess base.

@ Incorrect answer. The amino group is a base, which enables proteins to neutralize excess acids.

c. Proteins are able to neutralize either excess base or excess acid because they have a neutral amino group.

@ Incorrect answer. The amino group is a base, which enables proteins to neutralize excess acids. The carboxylic acid group is an acid, which enables proteins to neutralize excess bases.

*d. Proteins are made up of basic amino groups and acidic carboxylic acid groups, which can be used to neutralize either excess bases or acids.

@ Correct answer. Proteins can neutralize either excess bases or excess acids because they are made up of an amino group (base) and a carboxylic acid group (acid).

7. Which of the following correctly describes the bond between two oxygen atoms?

*a. Two oxygen atoms share a double bond, enabling them both to fulfill the octet rule.

@ Correct answer. One oxygen atom will share two of its electrons with the other atom, forming a double bond and fulfilling the octet rule for both atoms.

b. Two oxygen atoms share bonds with two hydrogen atoms, enabling them to fulfill the octet rule.

@ Incorrect answer. Two oxygen atoms only need to share two electrons with one another in order to form a double bond that fulfills the octet rule.

c. Two oxygen atoms share protons, forming a double bond and fulfilling the octet rule.

@ Incorrect answer. Two oxygen atoms share electrons, not protons, to form a double bond and fulfill the octet rule.

d. Two oxygen atoms share two electrons to form a double bond and eight neutrons to fulfill the octet rule.

@ Incorrect answer. Two oxygen atoms share only two electrons to form a double bond and fulfill the octet rule.

8. Which of the following correctly describes a molecule that is most likely to be polar?

*a. A molecule enriched in hydroxyl groups

@ Correct answer. A molecule that contains O-H or N-H bonds is most likely to be polar.

b. A molecule modified with additional methyl groups

@ Incorrect answer. Methyl groups only contain carbon and hydrogen, making them nonpolar.

c. A molecule with 14 carbons, all bonded to hydrogens

@ Incorrect answer. A molecule containing only carbon and hydrogen is a nonpolar molecule.

d. A molecule of carbon dioxide with linear symmetry

@ Incorrect answer. Carbon dioxide cannot have an electrical dipole because it is a straight, symmetrical molecule.

9. Electronegativity is the tendency of an element to attract a bonding pair of electrons. Which of the following elements is the most electronegative?

a. Hydrogen

@ Incorrect answer. Electronegativity increases across the periodic table. Hydrogen has one electron and is more likely to release a valence electron.

b. Carbon

@ Incorrect answer. Carbon has four valence electrons.

c. Potassium

@ Incorrect answer. Potassium has one valence electron and is more likely to release it.

*d. Oxygen

@ Correct answer. Oxygen has six valence electrons and is more likely to attract two more electrons to fulfill the octet rule.

10. Which of the following accurately describes a fact about hydrophobic molecules?

a. Water is nonpolar so hydrophobic molecules cannot readily dissolve in water.

@ Incorrect answer. Water is polar. Nonpolar molecules are unable to form hydrogen bonds or ionic interactions with polar molecules. Therefore, hydrophobic molecules cannot dissolve in water.

*b. Hydrophobic molecules help determine the three-dimensional structure of proteins.

@ Correct answer. Many proteins contain both hydrophilic and hydrophobic areas. Hydrophobic molecules are attracted to other hydrophobic molecules and this interaction contributes to the three-dimensional structure of the protein.

c. Hydrophobic molecules are unable to dissolve in water because they form hydrogen bonds.

@ Incorrect answer. Hydrophobic molecules are unable to dissolve in water because they cannot form hydrogen bonds or ionic interactions with water.

d. Hydrophobic molecules cannot be transported in aqueous solutions because they do not dissolve.

@ Incorrect answer. Hydrophobic molecules cannot dissolve in aqueous solutions, but they can be transported with special packaging.

11. Why do some individuals have lactose intolerance?

*a. Some adults are unable to produce lactase.

@ Correct answer. Some adults are unable to produce lactase, the enzyme needed to breakdown lactose.

b. In some adults, the enzyme changes shape.

@ Incorrect answer. Some adults are unable to produce lactase.

c. In some adults, the enzyme lactase is unable to break down the milk sugars.

@ Incorrect answer. Some adults lack the ability to produce lactase, which is the enzyme needed to break down the milk sugar lactose.

d. In some adults, an inappropriate immune response occurs as a result of exposure to lactose.

@ Incorrect answer. An allergic response is not why some individuals have lactose intolerance. It is a lack of production of lactase, the enzyme needed to break down the milk sugar that causes lactose intolerance.

12. How do carbohydrates impact the brain?

a. Carbohydrates decrease brain activity.

@ Incorrect answer. The brain relies on the carbohydrate glucose for most functions.

b. Glycolipids enhance neural activity.

@ Incorrect answer. Glycolipids are lipids with an added carbohydrate. Glycolipids form a sugary membrane coat.

*c. The brain relies on glucose for most functions.

@ Correct answer. The carbohydrate glucose is what the brain relies on for most functions.

d. The brain relies on glycoproteins as its main energy source.

@ Incorrect answer. Glycoproteins are proteins with an added carbohydrate. They are abundant in the plasma membrane.

13. Phospholipids are two fatty acids bound to a glycerol molecule. Which of the following is one reason why phospholipids are able to form cell membranes?

a. Phospholipids are amphipathic, meaning they have two polar ends.

@ Incorrect answer. Phospholipids are amphipathic, but this means they have one polar end and one nonpolar end.

b. Phospholipids have two nonpolar ends, so they can form a barrier against water.

@ Incorrect answer. Phospholipids make good plasma membranes because they have one polar end and one nonpolar end.

c. Phospholipids are not able to spontaneously aggregate, making them more stable as plasma membranes.

@ Incorrect answer. Phospholipids do spontaneously aggregate, making them stable as plasma membranes.

*d. Phospholipids have one polar end and one nonpolar end, enabling them to form plasma membranes.

@ Correct answer. Phospholipids are amphipathic, the basis for their major biological role in forming cell membranes.

14. Which of the following compounds is soluble in the phospholipid bilayer and thus can easily cross cell membranes without the need of a channel or carrier protein?

a. Na^+ ions

@ Incorrect answer. Charged ions are not soluble in lipid solutions due to their charge.

b. A hemoglobin tetramer

@ Incorrect answer. A large protein is unable to cross the membrane due to its size.

*c. O_2

@ Correct answer. Molecular oxygen is made of two oxygen atoms, which equally share four electrons to create a double covalent bond. Therefore, these two oxygen atoms are equally electromagnetic and thus the molecule is nonpolar.

d. Glucose

@ Incorrect answer. Glucose is a polar molecule and thus it is not soluble in the phospholipid bilayer. In fact, glucose is a water-soluble molecule due to its hydroxyl groups that can form hydrogen bonds with water.

15. The production of prostaglandins and leukotrienes during inflammation depends on the presence of a specific:

a. Cell membrane carbohydrate

@ Incorrect answer. Prostaglandins and leukotrienes are made from plasma membrane arachidonic acid.

*b. Cell membrane lipid

@ Correct answer. Cell membrane phospholipids are the source of the fatty acid arachidonic acid.

c. Cell membrane protein

@ Incorrect answer. Prostaglandins and leukotrienes are made from plasma membrane arachidonic acid.

d. Intracellular nucleotide

@ Incorrect answer. Prostaglandins and leukotrienes are made from plasma membrane arachidonic acid.

16. Which of the following is true regarding energy storage in the body?

*a. Triglycerides are the major form of energy storage because hydrocarbon chains release large amounts of energy when they undergo catabolic reactions.

@ Correct answer. Triglycerides, mainly stored in adipose tissue, release large amounts of energy when the hydrocarbon chains undergo catabolic reactions. Therefore, they are the major source of energy in the body.

b. Glycoproteins are the major form of energy storage because they are readily broken down into glucose.

@ Incorrect answer. Triglycerides, not glycoproteins, are the major form of energy storage for the body.

c. Triglycerides are a minor form of energy storage because the hydrogen chains break apart easily.

@ Incorrect answer. Triglycerides are made up of hydrocarbon chains that release large amounts of energy when they undergo catabolic reactions.

d. The main storage area for energy is in the mitochondria.

@ Incorrect answer. The main storage area for triglycerides is adipose tissue.

17. The endogenous pathway transports energy packaged in the liver to the tissues for use. Which of the following delivers the triglycerides to the tissues?

a. High-density lipoproteins (HDL)

@ Incorrect answer. HDL transfers apolipoproteins to VLDL and into the circulation.

b. Low-density lipoproteins (LDL)

@ Incorrect answer. LDL is mostly made of cholesterol and functions in the delivery of cholesterol to the cells.

c. Intermediate-density lipoproteins (IDL)

@ Incorrect answer. IDLs are remnants of VLDL after tissues have removed triglycerides.

*d. Very low-density lipoproteins (VLDL)

@ Correct answer. VLDLs are rich with triglycerides, which are delivered to the tissues.

18. What is the source of familial forms of hypercholesterolemia?

*a. Mutations in the gene that codes for apolipoprotein B-100 (Apo B-100)

@ Correct answer. Familial forms of hypercholesterolemia are usually associated with mutations in the gene that codes for Apo B-100.

b. Mutations in the gene that codes for very low-density lipoproteins

@ Incorrect answer. Mutations in the gene that codes for Apo B-100 are associated with familial forms of hypercholesterolemia.

c. Increased intake of saturated fatty acids

@ Incorrect answer. Mutations in the gene that codes for Apo B-100 are inherited in a dominant fashion in familial forms of hypercholesterolemia.

d. Environmental factors that cause mutations of the genes that code for triglycerides

@ Incorrect answer. Familial forms of hypercholesterolemia are usually associated with normal triglyceride levels.

19. Intrachain bonds between protein chains are dependent on the presence of this amino acid:

a. Alanine

@ Incorrect answer. Alanine has a hydrophobic side chain and cannot make cross-links with other amino acid side chains.

b. Aspartate

@ Incorrect answer. Aspartate has an acidic side chain and cannot make cross-links with other amino acid side chains.

*c. Cysteine

@ Correct answer. Cysteine has a thiol side chain and can form a disulfide bond with another cysteine.

d. Cytosine

@ Incorrect answer. Cytosine is a pyrimidine base, not an amino acid.

20. Which nucleic acid is responsible for transporting information to the ribosomes for protein synthesis?

a. Deoxyribonucleic acid (DNA)

@ Incorrect answer. DNA stores genetic information necessary to produce proteins.

*b. Ribonucleic acid (RNA)

@ Correct answer. RNA transports genetic instructions for production of proteins from the DNA to the ribosomes.

c. Guanosine triphosphate (GTP)

@ Incorrect answer. GTP provides a timing switch for membrane receptors.

d. Cyclic adenosine monophosphate (cAMP)

@ Incorrect answer. cAMP is an intracellular second messenger turning on a signaling cascade.

21. What is the cause of hypopigmentation in children with PKU (phenylketonuria)?

a. There is too little phenylalanine.

@ Incorrect answer. There are high levels of phenylalanine because the enzyme to convert phenylalanine into tyrosine is missing.

b. There is an accumulation of phenylacetate.

@ Incorrect answer. Through a secondary metabolic pathway, phenylalanine is converted into phenylacetate, which is then excreted through the urine. Phenylacetate gives urine a mousy odor.

*c. Tyrosine is in short supply.

@ Correct answer. Melanin is produced from tyrosine. Tyrosine is reduced in PKU because they are missing the enzyme responsible for converting phenylalanine into tyrosine.

d. There is an accumulation of melanin.

@ Incorrect answer. There is too little melanin due to a short supply of tyrosine. Too little melanin leads to hypopigmentation.

22. The difference between chemically reactive and chemically inert elements is determined by:

a. The number of protons in the nucleus

@ Incorrect answer. Protons in an atom's nucleus do not interact with other atoms.

b. The number of neutrons in the nucleus

@ Incorrect answer. Neutrons in an atom's nucleus do not interact with other atoms.

*c. The presence or absence of electrons that completely fill the outer, valence shell

@ Correct answer. An incompletely filled valence shell allows an atom to react with other atoms.

d. The total number of electrons in the atom

@ Incorrect answer. The determining factor in an element's reactivity is the filling of the outermost valence electron shell.

23. Which of the following correctly describes the process of sodium chloride dissolving in water?

a. The atoms of sodium and chloride remain attached via an ionic bond.

@ Incorrect answer. The ions separate when a sodium chloride molecule dissolves in water.

b. The sodium and chloride atoms separate, retaining their initial atomic valence shell electron configuration.

@ Incorrect answer. The sodium and chloride atoms become ions, with sodium losing one electron to become a positive ion and chloride gaining one electron to become a negative ion.

*c. The sodium ion acquires a hydration shell of water molecules, with the oxygens oriented toward the sodium ion; the chloride ion acquires a hydration shell of water molecules, with the hydrogens oriented toward the chloride ion.

@ Correct answer. The electronegative oxygen within polar water molecules is attracted to the sodium's positive charge; the electropositive hydrogens within water molecules are attracted to the chloride's negative charge.

d. The sodium ion acquires a hydration shell of water molecules, with the hydrogens oriented toward the sodium ion; the chloride ion acquires a hydration shell of water molecules, with the oxygens oriented toward the chloride ion.

@ Incorrect answer. The electronegative oxygen within polar water molecules is attracted to the sodium's positive charge; the electropositive hydrogens within water molecules are attracted to the chloride's negative charge.

24. Which of the following is not a role of ions in the body?

*a. Formation of hydrophobic membrane barriers

@ Correct answer. Ions are hydrophilic.

b. Maintaining acid–base balance

@ Incorrect answer. Acids and bases that buffer body fluids are also ions.

c. Carrying electrical charges

@ Incorrect answer. Ions are also electrolytes; when in solution, they help carry electrical charges.

d. Generation of membrane potentials

@ Incorrect answer. Differential distribution of ions and varying membrane ion permeability help create membrane potentials.

25. The components of the bicarbonate buffer system include which of these molecules?

a. Amino acid lysine

@ Incorrect answer. Amino acids contribute to buffering by proteins.

b. Ammonia

@ Incorrect answer. Buffering by ammonia is independent of the bicarbonate buffer system.

*c. Carbonic acid

@ Correct answer. Carbonic acid is the central compound of the bicarbonate buffer system, formed when bicarbonate ion is exposed to excess acid.

d. Hydrochloric acid

@ Incorrect answer. Hydrochloric acid is a strong acid involved in gastric secretion and digestion.

26. Nonpolar biomolecules are distinguished by their abundance of which of the following?

*a. Carbon–hydrogen bonds

@ Correct answer. Carbon–hydrogen bonds are nonpolar covalent bonds.

b. Carbon–nitrogen–hydrogen bonds

@ Incorrect answer. The presence of nitrogen bonded to hydrogen in a biomolecule constitutes a polar covalent bond.

c. Carbon–oxygen–hydrogen bonds

@ Incorrect answer. The presence of oxygen bonded to hydrogen in a biomolecule constitutes a polar covalent bond.

d. Carbon–oxygen–oxygen–hydrogen bonds

@ Incorrect answer. Carbon bonded to two oxygens, one of which is bonded to a hydrogen, describes a carboxyl group with polar and charged characteristics.

27. Transfer of a phosphate group from adenosine triphosphate (ATP) to a biomolecule (also known as phosphorylation) involves this type of chemical reaction:

a. Degradation

@ Incorrect answer. It is an exchange reaction; reactants are not being degraded.

*b. Exchange

@ Correct answer. The phosphate group is exchanged for a hydrogen on the accepting biomolecule.

c. Hydroxylation

@ Incorrect answer. A phosphate is being transferred, rather than a hydroxyl group.

d. Synthesis

@ Incorrect answer. It is an exchange reaction; reactants are not being built into larger molecules.

28. Which category of biomolecules has the greatest hydrophobicity?

a. Amino acids

@ Incorrect answer. Carbohydrates have charged and polar carboxyl and amine groups and are very polar and hydrophilic.

b. Carbohydrates

@ Incorrect answer. Carbohydrates are rich in hydroxyl groups and are very polar and hydrophilic.

*c. Lipids

@ Correct answer. Fatty acids, triglycerides, and cholesterol are rich in C-H bonds and are very nonpolar and hydrophobic.

d. Nucleotides

@ Incorrect answer. Nucleotides have N-H bonds that increase their polar, hydrophilic qualities.

29. What is the major biomolecule group that modifies membrane lipids and proteins forming glycolipids and glycoproteins used as cell markers?

a. Amino acids

@ Incorrect answer. Amino acids are rarely found as modifying attachments to membrane lipids and proteins.

b. Fatty acids

@ Incorrect answer. Carbohydrates, particularly hexose sugars, are the most common biomolecules that modify membrane lipids and proteins.

*c. Hexose sugars

@ Correct answer. Hexose sugars, singly and in chains, are common biomolecules that modify membrane constituents, forming glycolipids and glycoproteins.

d. Nucleotides

@ Incorrect answer. Nucleotides are not commonly found on the outer membrane surface attached to lipids and proteins.

30. Which of the following food products contain the least amount of water?

a. Almonds

@ Incorrect answer. Although almonds have low water content (3%–6%) due to the fact that as seeds they undergo the process of desiccation, they still do contain water.

b. Dried mushrooms

@ Incorrect answer. Although dried mushrooms have been artificially dehydrated, their water content may still range between 5% and 8%.

*c. Oil

@ Correct answer. Water content in extra virgin olive oil is about 700 to 2,000 ppm, which is significantly lower than the water content in other foods simply because it is hydrophobic.

d. Salami

@ Incorrect answer. While rich in fat, salami still has high water content, about 60%.

31. What is a common membrane phospholipid that is amphipathic, having two fatty acid tails and a polar head group?

a. Blood group antigens

@ Incorrect answer. Blood group antigens are membrane sphingolipids with attached carbohydrate groups.

b. Cholesterol

@ Incorrect answer. Cholesterol is a steroid-type membrane lipid.

*c. Phosphatidylcholine

@ Correct answer. Phosphatidylcholine has two fatty acid tails and the polar head group choline.

d. Triglyceride

@ Incorrect answer. Triglycerides have three fatty acid chains and no polar head group; therefore, they are not amphipathic.

32. Which of the following is not a prominent component of a lipoprotein particle?

*a. Carbohydrate

@ Correct answer. Lipoproteins consist of a lipid core with a phospholipid and protein coat.

b. Cholesterol

@ Incorrect answer. Cholesterol is found in varying degrees in many lipoproteins.

c. Triglyceride

@ Incorrect answer. Triglyceride molecules are found in varying degrees in many lipoproteins.

d. Phospholipids

@ Incorrect answer. Phospholipids are found in the coat of many lipoproteins.

33. Which description best characterizes hydrogen bonding in protein secondary structures like α-helices and β-pleated sheets?

a. Oxygen and hydrogen atoms attached to polar R groups form intrachain hydrogen bonds.

@ Incorrect answer. Intrachain hydrogen bonds form between oxygens and hydrogens of the peptide backbone.

b. Oxygen and nitrogen atoms attached to polar R groups form intrachain hydrogen bonds.

@ Incorrect answer. Intrachain hydrogen bonds form between oxygens and hydrogens of the peptide backbone.

c. Oxygen and hydrogen atoms attached to charged R groups form intrachain hydrogen bonds.

@ Incorrect answer. Intrachain hydrogen bonds form between oxygens and hydrogens of the peptide backbone.

*d. Oxygen and hydrogen atoms attached to the peptide backbone form intrachain hydrogen bonds.

@ Correct answer. Intrachain hydrogen bonds form between oxygens and hydrogens of the peptide backbone.

34. Which portions of a folded globular protein are stabilized by hydrophobic attraction between R groups?

*a. The protein core

@ Correct answer. Hydrophobic R groups cluster in the core of globular proteins, minimizing interactions with the aqueous solution of intracellular or extracellular fluid.

b. The protein surface

@ Incorrect answer. Hydrophobic R groups are found in the core of folded proteins, while the outer surface is rich in hydrophilic charged and polar R groups.

c. Regions of both protein core and surface

@ Incorrect answer. Hydrophobic R groups are found in the core of folded proteins.

d. None of these; folding of globular proteins does not involve R groups

@ Incorrect answer. Hydrophobic amino acid R groups influence protein folding by promoting attraction into the core region.

35. Individual amino acids can function as precursor molecules for biological signals. An example of a signaling molecule derived from the amino acid tyrosine is:

a. Cholesterol

@ Incorrect answer. Cholesterol is a lipid and is not derived from an amino acid.

b. Glycogen

@ Incorrect answer. Glycogen is a polymer of glucose and is not derived from an amino acid

c. Insulin

@ Incorrect answer. Insulin is a protein hormone made up of many amino acids.

*d. Norepinephrine

@ Correct answer. Norepinephrine is a neurotransmitter derived from the amino acid tyrosine.

36. Which of the following is composed of a single nitrogenous base, phosphate group, and ribose sugar molecule?

a. Nucleoside

@ Incorrect answer. A nucleoside has a nitrogenous base and pentose sugar, but no phosphate group.

*b. Nucleotide

@ Correct answer. A nucleotide has a nitrogenous base, a pentose sugar, and one or more phosphate groups.

c. Ribonucleic acid (RNA)

@ Incorrect answer. RNA is a polymer of many nucleotides.

d. Deoxyribonucleic acid (DNA)

@ Incorrect answer. DNA is a polymer of many nucleotides.

37. Which of the following cell functions cannot be performed by a nucleotide?

a. Energy storage and use

@ Incorrect answer. Adenosine triphosphate (ATP) is the principal unit of cell energy storage and use.

*b. Protein synthesis

@ Correct answer. Proteins are synthesized from amino acids, not nucleotides.

c. Formation of nucleic acids

@ Incorrect answer. Nucleic acids are polymers of nucleotides.

d. Intracellular second messenger signaling

@ Incorrect answer. Cyclic adenosine monophosphate (AMP) and cyclic guanosine monophosphate (GMP) are intracellular second messengers.

38. Based on the principle of complementary base pairing, in double-stranded DNA the number of adenine molecules must equal the number of molecules of:

a. Cytosine

@ Incorrect answer. Cytosine forms base pairs with guanine, not adenine.

b. Guanine

@ Incorrect answer. Guanine forms base pairs with cytosine, not adenine.

*c. Thymine

@ Correct answer. Thymine forms base pairs with adenine; therefore, there must be an equal number of thymine and adenine molecules.

d. Uracil

@ Incorrect answer. Uracil is a base in RNA, not DNA.

39. A characteristic that distinguishes DNA from RNA molecules is:

a. The presence of a pentose sugar attached to each base

@ Incorrect answer. Both DNA and RNA have pentose sugar molecules; however, DNA has deoxyribose sugars, while RNA has ribose sugars.

b. The presence of a phosphate group attached to each nucleotide

@ Incorrect answer. Both DNA and RNA have phosphate groups in their backbones.

c. The specific purines in the molecule

@ Incorrect answer. Both DNA and RNA use the same purine bases: adenine and guanine.

*d. The specific pyrimidines in the molecule

@ Correct answer. DNA uses cytosine and thymine, while RNA uses cytosine and uracil.

40. Which treatment has been the main approach to reduce the pathophysiological consequences of phenylalanine hydroxylase deficiency leading to phenylketonuria (PKU)?

a. Gene therapy administered at birth to replace the phenylalanine hydroxylase enzyme

@ Incorrect answer. Gene therapy is not currently available for this condition.

b. In vitro fertilization with preimplantation screening to avoid implanting an embryo with phenylalanine hydroxylase deficiency

@ Incorrect answer. Preimplantation screening for this condition has not been used.

c. Management with medications that reduce phenylalanine concentrations

@ Incorrect answer. There are no readily available medications to clear phenylalanine from the body.

*d. Nutrition therapy with a low phenylalanine diet to reduce hyperphenylalaninemia

@ Correct answer. The mainstay of therapy is to feed a low phenylalanine diet as soon as the enzyme deficiency is identified in a newborn, thus preventing the damaging effects of excess phenylalanine.

41. Which of the following molecules is most likely to be hydrophobic and poorly soluble in an aqueous environment?

a. Alanine

@ Incorrect answer. Alanine is an amino acid with polar or charged carboxyl and amine groups that improve its solubility in water.

*b. Estrogen

@ Correct answer. Estrogen is a steroid hormone that is based on the lipid cholesterol; therefore, it is hydrophobic and poorly soluble in an aqueous environment.

c. Glucose

@ Incorrect answer. Glucose molecules are enriched in polar hydroxyl groups, making glucose very hydrophilic.

d. Guanosine

@ Incorrect answer. Guanosine has a nitrogenous base bonded to a pentose sugar, with both molecular components contributing to its polar and hydrophilic nature.

42. What components of lipoprotein molecules contribute to their ability to travel freely in the circulation despite their overwhelmingly hydrophobic components?

a. They are rich in cholesterol.

@ Incorrect answer. Cholesterol is not water-soluble and cannot easily travel in the circulation.

b. They are rich in triglyceride.

@ Incorrect answer. Triglycerides are not water-soluble and cannot easily travel in the circulation.

*c. They are coated with phospholipids and proteins.

@ Correct answer. Hydrophobic cholesterol and triglyceride are coated with amphipathic phospholipids and proteins, allowing lipoproteins to travel in the aqueous plasma within the circulation.

d. They have varying densities.

@ Incorrect answer. Although lipoproteins have different densities, they all have a hydrophobic core that would prevent free travel in the circulation without a coat of phospholipid and protein.

43. When cholesterol molecules are assembled in lipoproteins, what is their orientation?

*a. The hydroxyl group is facing outside of the lipoprotein, toward the blood.

@ Correct answer. This is the hydrophilic component and thus it is exposed to the blood, which is an aqueous solution.

b. The whole molecule is exposed toward the outside of the lipoprotein, toward the blood.

@ Incorrect answer. Cholesterol contains hydrophobic components that cannot be exposed to the blood, which is an aqueous solution.

c. The hydrocarbon chain is facing outside of the lipoprotein, toward the blood.

@ Incorrect answer. This is a hydrophobic component and thus cannot be exposed to the blood, which is an aqueous solution.

d. The whole molecule is immersed in the inside of the lipoprotein; none of it is exposed to the blood.

@ Incorrect answer. The hydroxyl group is hydrophilic and thus it is not a fat-soluble molecule.

44. Which of the following genetic mutations could theoretically cause the least disruption to protein folding?

a. Replacing a leucine with an arginine

@ Incorrect answer. Leucine is nonpolar while arginine is positively charged and thus this change will likely affect protein folding.

*b. Replacing a leucine with an isoleucine

@ Correct answer. Both leucine and isoleucine are nonpolar and thus this change is less likely to affect protein folding.

c. Replacing a leucine with a tyrosine

@ Incorrect answer. Leucine is nonpolar while tyrosine contains a hydroxyl group and thus this change will likely affect protein folding.

d. Replacing a leucine with a glutamate

@ Incorrect answer. Leucine is nonpolar while glutamate is negatively charged and thus this change will likely affect protein folding.

Chapter 3

Molecular Biology, Genetics, and Genetic Disorders

Multiple Choice Questions

1. Why is it important to screen newborns for phenylketonuria (PKU)?

   a. It is necessary to supplement the newborn's diet with digestive enzymes.

   @ Incorrect answer. PKU does not involve missing digestive enzymes.

   b. Surgical correction can prevent progression.

   @ Incorrect answer. Surgical correction does not prevent progression of PKU.

   c. Signs and symptoms often do not appear until school age.

   @ Incorrect answer. Signs and symptoms of PKU may appear early or later, depending on the severity of the phenotype.

   *d. Immediate dietary changes can mitigate some disease manifestations from developing.

   @ Correct answer. Immediate adoption of a low-phenylalanine diet mitigates many of the phenotype manifestations from developing.

2. Which of the following is responsible for repairing errors that occur during replication?

   *a. DNA polymerase

   @ Correct answer. DNA polymerases repair DNA during the replication process.

   b. Micro RNA

   @ Incorrect answer. Micro RNAs (miRNAs) are capable of binding to messenger RNA (mRNA) to accelerate its degradation and reduce the expression of associated proteins.

   c. RNA polymerase

   @ Incorrect answer. RNA polymerase transcribes the DNA into pre-mRNA.

   d. Introns

@ Incorrect answer. Introns are the noncoding regions of genes.

3. During processing after transcription, which of the following forms the anchor to attach the messenger RNA (mRNA) to the ribosome?

   a. Promoter region

   @ Incorrect answer. The promoter region of the DNA tells the RNA polymerase where to begin transcription.

   b. Terminating region

   @ Incorrect answer. The terminating region of the DNA tells the RNA polymerase where to stop transcription.

   c. Poly-A tail

   @ Incorrect answer. The poly-A tail is attached to one end of the exons to add stability and extend the time it takes for the molecule to be degraded.

   *d. 5′ cap

   @ Correct answer. The 5′ cap forms an anchor for the ribosome to attach the mRNA and begin translation.

4. Which of the following regulates the degradation of messenger RNA (mRNA)?

   *a. siRNA

   @ Correct answer. siRNAs, or small interfering RNAs, are short segments of RNA that bind to mRNA and accelerate its degradation.

   b. tRNA

   @ Incorrect answer. tRNA, or transfer RNA, is a carrier molecule that carries amino acids to contribute to a growing protein chain.

   c. rRNA

   @ Incorrect answer. rRNA, or ribosomal RNA, is the site of protein synthesis.

d. snRNA

@ Incorrect answer. snRNA, or small nuclear RNA, splices pre-mRNA as one component of a spliceosome complex.

5. Which of the following is the most significant red flag that may indicate a genetic component to the disease?

*a. A 38-year-old female with heart disease

@ Correct answer. Early onset of cardiovascular disease is a red flag for a genetic link to the disease.

b. Breast cancer in a 64-year-old female

@ Incorrect answer. Breast cancer occurring in women before the age of 60 is a red flag for a genetic link to the disease.

c. Diabetes (DM) type II in a parent and child

@ Incorrect answer. Multiple family members across multiple generations may be a red flag, but with DM type II in a parent and child it may also be environmental or lifestyle-related.

d. Prostate cancer in a 72-year-old male

@ Incorrect answer. Prostate cancer in an older male patient is not a red flag for a genetic link to the disease.

6. Which of the following epigenetic mechanisms is found in breast, cervical, and thyroid cancers?

a. Global hypermethylation

@ Incorrect answer. Global hypomethylation is found in many cancers.

b. Histone modification

@ Incorrect answer. Histone modification is an epigenetic mechanism, but not specific to breast, cervical, and thyroid cancers.

c. Transgenerational epigenetic inheritance

@ Incorrect answer. Transgenerational epigenetic inheritance is an epigenetic mechanism, but not specific to breast, cervical, and thyroid cancers.

*d. Global hypomethylation

@ Correct answer. Global hypomethylation is found in many cancers, including breast, cervical, thyroid, lung, prostate, bladder, stomach, esophagus, colon, and liver cancer.

7. Which of the following best describes the epigenetic mechanisms at work in aging?

*a. Environmental exposures add up over time and influence epigenetic mechanisms.

@ Correct answer. Epigenetic mechanisms are influenced by environmental chemical exposures, drugs, aging, and diet.

b. Generalized hypermethylation pattern occurs across the genome.

@ Incorrect answer. Generalized hypomethylation occurs in aging.

c. Gene-specific CpG island hypomethylation sites increase.

@ Incorrect answer. There are some gene-specific CpG island hypermethylation sites.

d. Decreased clearance of drugs inhibits epigenetic mechanisms.

@ Incorrect answer. Decreased clearance of drugs influences epigenetic mechanisms.

8. Which of the following statements is true regarding DNA?

a. DNA in germ cells contains 46 chromosomes.

@ Incorrect answer. DNA in germ cells contains 23 chromosomes. Upon fertilization, there are 46 chromosomes within the resulting zygote.

b. The complementary base of adenine is guanine.

@ Incorrect answer. The complementary base pair of adenine is thymine. The complementary base pair of guanine is cytosine.

*c. Replication of DNA proceeds from the 5′ to the 3′ direction.

@ Correct answer. DNA replication involves several complex enzyme processes. Building the new strand proceeds from the 5′ to 3′ direction, reading off a template strand oriented in the 3′ to 5′ direction.

d. Proofreading prevents nucleotides from being inserted incorrectly.

@ Incorrect answer. Proofreading removes the nucleotides that have been inserted incorrectly so that they cannot cause a permanent change in DNA.

9. Which of the following is true in the process of DNA coding?

*a. The transcription factor's primary purpose is in the production of premessenger RNA.

@ Correct answer. Transcription factor influences the separation of DNA strands and binding of RNA polymerase to form premessenger RNA.

b. Noncoding DNA is not involved in the process.

@ Incorrect answer. Noncoding DNA relates transcription factor binding and influences gene expression.

c. The transfer RNA's primary purpose is protein folding.

@ Incorrect answer. Transfer RNA brings amino acids to the ribosome for protein building.

d. 95% of human DNA is identical.

@ Incorrect answer. 99.6% of human DNA is identical.

10. Which of the following is caused by a single base change and results in an amino acid change?

a. Nonsense mutation

@ Incorrect answer. A nonsense mutation occurs due to a premature stop codon and produces a nonfunctional protein.

b. Deletion mutation

@ Incorrect answer. A deletion mutation occurs when one or more bases are deleted.

*c. Missense mutation

@ Correct answer. A missense mutation is a change in a single base that results in an amino acid change.

d. Frameshift mutation

@ Incorrect answer. A frameshift mutation occurs due to a deletion or insertion mutation altering the messenger RNA's reading frame.

11. One parent carries the gene for the autosomal dominant disorder familial hypercholesterolemia and the other parent does not. One child has the disorder. What is the chance using Mendelian inheritance principles that the next child will have the disease?

a. 0%

@ Incorrect answer. Familial hypercholesterolemia is an autosomal dominant disorder. If one parent has the disorder, there is a 50% chance they will pass on the abnormal copy of the gene to each offspring.

*b. 50%

@ Correct answer. Familial hypercholesterolemia is an autosomal dominant disorder. If one parent has the disorder, there is a 50% chance they will pass on the abnormal copy of the gene to each offspring.

c. 25%

@ Incorrect answer. Familial hypercholesterolemia is an autosomal dominant disorder. If one parent has the disorder, there is a 50% chance they will pass on the abnormal copy of the gene to each offspring.

d. 75%

@ Incorrect answer. Familial hypercholesterolemia is an autosomal dominant disorder. If one parent has the disorder, there is a 50% chance they will pass on the abnormal copy of the gene to each offspring.

12. Which of the following is not true regarding autosomal recessive disorders?

a. Affected offspring usually have unaffected parents.

@ Incorrect answer. With autosomal recessive disorders, affected individuals are usually born to unaffected carriers.

b. There are fewer affected individuals in a family pedigree.

@ Incorrect answer. With autosomal recessive disorders, there are overall fewer individuals affected in the family pedigree.

c. The disorder may appear to "skip" a generation.

@ Incorrect answer. With autosomal recessive disorders, the pattern may not present in every generation, appearing to "skip" a generation.

*d. Females are affected more often than males.

@ Correct answer. With autosomal recessive disorders, either sex can be affected equally.

13. Which of the following is an X-linked dominant disorder?

a. Huntington disease

@ Incorrect answer. Huntington disease is an autosomal dominant disorder.

b. Tay–Sachs disease

@ Incorrect answer. Tay–Sachs disease is an autosomal recessive disorder.

*c. Rett syndrome

@ Correct answer. Rett syndrome is an X-linked dominant disorder.

d. Neurofibromatosis

@ Incorrect answer. Neurofibromatosis is an autosomal dominant disorder.

14. A father has been diagnosed with Leber hereditary optic neuropathy (LHON) at age 28. What is the chance that one of his three children will develop the disease?

a. Each has a 25% chance because LHON is an autosomal recessive disorder.

@ Incorrect answer. LHON is a mitochondrial DNA disorder. A male with LHON will not pass it on to any of his children as the sperm contributes little to no mitochondrial DNA.

*b. There is no chance the children will develop this disorder because it is linked to mitochondrial DNA.

@ Correct answer. LHON is a mitochondrial DNA disorder. A male with LHON will not pass it on to any of his children as the sperm contributes little to no mitochondrial DNA.

c. Each female child has a 50% chance of developing this X-linked dominant disorder.

@ Incorrect answer. LHON is a mitochondrial DNA disorder. A male with LHON will not pass it on to any of his children as the sperm contributes little to no mitochondrial DNA.

d. There is little chance the female children will develop this X-linked recessive disorder.

@ Incorrect answer. LHON is a mitochondrial DNA disorder. A male with LHON will not pass it on to any of his children as the sperm contributes little to no mitochondrial DNA.

15. Which protein family is required for tight packaging of DNA?

a. DNA polymerase

@ Incorrect answer. DNA polymerase synthesizes the complementary DNA strand during replication.

b. Helicase

@ Incorrect answer. Helicase unwinds the DNA strand to prepare for replication.

*c. Histone

@ Correct answer. Histone proteins are the sights of winding to package DNA.

d. Primase

@ Incorrect answer. Primase forms complementary RNA primers required to start DNA replication.

16. In DNA proofreading, how is an incorrect nucleotide removed?

a. Incorrect nucleotides are removed from the 3′ end by DNA ligase.

@ Incorrect answer. Incorrect nucleotides are removed from the 3′ end by DNA polymerase.

b. Incorrect nucleotides are removed from the 5′ end by helicase.

@ Incorrect answer. Incorrect nucleotides are removed from the 3′ end by DNA polymerase.

*c. Incorrect nucleotides are removed from the 3′ end by DNA polymerase.

@ Correct answer. DNA polymerase removes incorrect nucleotides at the growing (3′) end of the complementary DNA strand.

d. Incorrect nucleotides are removed from the 5′ end by topoisomerase.

@ Incorrect answer. Incorrect nucleotides are removed from the 3′ end by DNA polymerase.

17. Which of the following is different when comparing the processes of replication and transcription?

a. A polymerase enzyme is required.

@ Incorrect answer. Both replication and transcription use polymerase enzymes (DNA and RNA polymerases, respectively).

*b. Adenine forms base pairs with thymine.

@ Correct answer. A-T base pairs are used in DNA replication, while transcription uses A-U base pairs.

c. Synthesis proceeds in the 5′ to 3′ direction.

@ Incorrect answer. Nucleic acid strands are synthesized from 5′ to 3′ in both replication and transcription.

d. Nucleotides are joined together by a sugar–phosphate backbone.

@ Incorrect answer. Nucleotides are joined together by a sugar–phosphate backbone in both replication and transcription.

18. Which molecule is formed by gene transcription and codes for cytoplasmic protein synthesis?

*a. Messenger RNA

@ Correct answer. Transcription of a gene produces a complementary messenger RNA (mRNA) that codes for a protein synthesized by ribosomes in the cytoplasm.

b. Ribosomal RNA

@ Incorrect answer. Ribosomal RNA is transcribed from DNA and is used to build ribosomes.

c. Small nuclear RNA

@ Incorrect answer. Small nuclear RNA remains in the nucleus and directs pre-mRNA splicing.

d. Transfer RNA

@ Incorrect answer. Transfer RNA molecules each brings a single amino acid to a ribosome during translation.

19. Which of the following describes the effect of a nonsense mutation?

a. As a result of the mutation, a single amino acid is altered in the resulting protein.

@ Incorrect answer. This is the effect of a missense mutation.

b. As a result of the mutation, several additional amino acids are found in the resulting protein.

@ Incorrect answer. This is the effect of an insertion mutation, such as a trinucleotide repeat expansion.

*c. As a result of the mutation, a premature stop codon is inserted and the protein is shortened or is not produced at all.

@ Correct answer. A nonsense mutation results in insertion of a premature stop codon.

d. As a result of the mutation, there is no change in the final protein amino acid sequence.

@ Incorrect answer. This is the effect of a silent mutation.

20. Which familial pattern is seen in a trinucleotide repeat disorder such as Huntington disease?

a. Inheritance is always traced from an affected mother.

@ Incorrect answer. Matrilineal descent characterizes genetic mitochondrial disorders.

b. Inheritance shows an autosomal recessive pattern.

@ Incorrect answer. Trinucleotide repeat expansion disorders are neither strictly dominant or recessive; instead, the degree of pathology depends on the number of copies of the abnormal DNA triplet in the responsible gene.

c. Inheritance shows carrier mothers who have affected sons and carrier daughters.

@ Incorrect answer. X-linked descent is usually associated with carrier females and affected males.

*d. Inheritance shows greater severity and earlier onset in the later generations.

@ Correct answer. Expanded numbers of trinucleotide sequences in vulnerable genes can be further expanded during meiosis, so later generations have higher numbers and greater chance of developing the disease phenotype.

21. Phenotype severity in individuals with mutations of the cystic fibrosis transmembrane conductance regulator (CFTR) gene varies from mild gastrointestinal symptoms due to faulty pancreatic enzyme secretion to severe recurrent lung infections due to abnormal epithelial mucus production. This is correctly understood as an example of:

a. Decreased penetrance

@ Incorrect answer. In diseases that display decreased penetrance, some affected individuals are completely free of the disease phenotype.

*b. Variable expressivity

@ Correct answer. In diseases with variable expressivity, a variety of disease phenotypes are possible.

c. Codominance

@ Incorrect answer. Codominance describes genetic inheritance in which a child inherits one allele from each parent, and both alleles are expressed. An example is a child born to a mother with type A blood and a father with type B blood—that child has a chance of having type AB blood.

d. X-linked inheritance.

@ Incorrect answer. In X-linked inheritance, there is a difference in probability of inheritance based on chromosomal sex of the child.

22. What principle underlies the use of hybridization arrays and gene chips to detect subject genotypes?

*a. The principle of complementary base pairing

@ Correct answer. Gene chips used synthetic DNA strands designed to bind subject DNA sequences based on complementary base pairing.

b. The principle of gene silencing

@ Incorrect answer. Gene silencing occurs during development when one allele is destined to be repressed and the other allele is allowed to remain active.

c. The principle of dominant inheritance

@ Incorrect answer. Dominant inheritance occurs when one allele of a gene is expressed and determines the phenotype, while the other allele does not contribute to the phenotype.

d. The principle of familial inheritance

@ Incorrect answer. The principle of familial inheritance is used by documenting a subject's family history as thoroughly as possible.

23. Which test examines the length, banding, and number of chromosomes to assess for abnormalities?

a. Gene expression mapping

@ Incorrect answer. In gene expression mapping, messenger RNAs (mRNAs) are isolated from a biopsy and are tested with array hybridization to detect which genes are actively being expressed.

*b. Karyotyping

@ Correct answer. In karyotyping, a complete set of chromosomes is isolated from a subject and is examined for gross translocations, duplications, and omissions.

c. Single nucleotide polymorphism (SNP) mapping

@ Incorrect answer. In SNP mapping, a subject's DNA is broken down to small fragments that are tested on a hybridization array.

d. Whole genome sequencing

@ Incorrect answer. In whole genome sequencing, a subject's DNA is completely sequenced and compared with reference DNA.

24. Which of the following disorders usually detected during childhood is a disorder of abnormal chromosome number?

a. Cystic fibrosis

@ Incorrect answer. Cystic fibrosis is a monogenic, autosomal recessive disorder.

*b. Klinefelter syndrome

@ Correct answer. Males born with Klinefelter syndrome have an extra X chromosome (47,XXY).

c. Marfan syndrome

@ Incorrect answer. Marfan syndrome is a monogenic, autosomal dominant disorder.

d. Phenylketonuria (PKU)

@ Incorrect answer. PKU is a monogenic, autosomal recessive disorder.

25. Which of the following is correct about inheritance of an X-linked recessive disorder like Duchenne muscular dystrophy or glucose-6-phosphate dehydrogenase deficiency?

a. Having a father with the disorder means the probability is 25% of sons having the disorder.

@ Incorrect answer. The father does not donate the X chromosome to his sons so there is no chance he passed the affected gene or disorder to his son.

b. Having a father with the disorder means the probability is 25% of daughters having the disorder.

@ Incorrect answer. Having a father with the disorder means the probability is that 100% of daughters will be carriers and none will have the disorder.

c. Having a father with the disorder means the probability is 100% of sons being carriers.

@ Incorrect answer. The father does not donate the X chromosome to his sons so there is no chance he passed the affected gene to his sons.

*d. Having a father with the disorder means the probability is 100% of daughters being carriers.

@ Correct answer. Having a father with the disorder means that all daughters will inherit at least one copy of the affected gene and be carriers.

26. If an individual is homozygous for a given trait, this means they have:

a. Two different alleles on two homologous chromosomes

@ Incorrect answer. This describes heterozygosity.

*b. Two identical alleles on two homologous chromosomes

@ Correct answer. An individual is homozygous if they have identical alleles on both homologous chromosomes.

c. Two different alleles on two different chromosomes

@ Incorrect answer. Alleles are versions of genes that can vary but are generally found at a single locus on homologous chromosomes.

d. Two identical alleles on two different chromosomes

@ Incorrect answer. Alleles are versions of genes that can vary but are generally found at a single locus on homologous chromosomes.

27. Which of the following is not a mechanism of epigenetic programming of DNA?

*a. Alterations in the sequence of bases within chromosomes

@ Correct answer. Base sequences are altered by mutations, which are genetic changes, rather than epigenetic changes.

b. Alterations in methylation (addition/removal of methyl groups) of DNA

@ Incorrect answer. Methylation is one mechanism of epigenetic modification of DNA.

c. Alterations in acetylation (addition/removal of acetyl groups) of histone proteins

@ Incorrect answer. Acetylation of histones is an epigenetic modification of DNA.

d. Removal of methyl groups (hypomethylation) of gene promoter regions

@ Incorrect answer. Hypomethylation is an epigenetic modification of DNA.

28. Which of the following is a "genetic red flag" suggesting there is a genetic component to a disease or disorder?

a. Unknown family history

@ Incorrect answer. Having multiple family members with a given condition suggests a genetic component, but lack of or unknown family history does not rule out genetic components.

*b. Early age of disease onset with each generation

@ Correct answer. Early or extreme presentation is common in genetic diseases.

c. A married couple, both age 85, with the same neurodegenerative disease

@ Incorrect answer. The married couple do not share DNA.

d. An infant born with polydactyly but no other anomalies

@ Incorrect answer. While polydactyly may be one component of a genetic syndrome, it is often an independent diagnosis.

29. Which of the following is a key difference between meiosis and mitosis?

a. Mitosis produces daughter cells that are genetically different, whereas meiosis produces daughter cells that are genetically identical.

@ Incorrect answer. Mitosis produces two identical daughter cells; meiosis produces four genetically different daughter cells.

*b. Meiosis produces daughter cells that are genetically different, whereas mitosis produces daughter cells that are genetically identical.

@ Correct answer. Mitosis produces two identical daughter cells; meiosis produces four genetically different daughter cells.

c. Mitosis results in four daughter cells, whereas meiosis yields two.

@ Incorrect answer. Mitosis results in two identical cells, whereas meiosis results in four cells.

d. Mitosis occurs in gametes, whereas meiosis occurs in somatic cells.

@ Incorrect answer. Mitosis occurs in somatic cells, whereas meiosis occurs in gametes.

30. Which of the following describes the most commonly used routine screening test for inborn errors of metabolism and other genetic disorders?

a. A screening evaluation occurs during pregnancy using amniocentesis.

@ Incorrect answer. Amniocentesis is an elective invasive test that is not done in all pregnant women.

b. A screening evaluation occurs during pregnancy using cell-free DNA from a maternal blood sample.

@ Incorrect answer. A cell-free DNA test is an elective test that is not generally done without a specific reason.

c. A screening evaluation occurs during pregnancy using chorionic villus sampling.

@ Incorrect answer. Chorionic villus sampling is an invasive test that is not usually done without a specific reason.

*d. A screening blood sample is taken from the newborn shortly after birth.

@ Correct answer. Newborn testing is mandatory in many countries, testing for inborn errors of metabolism, as well as endocrine disorders, blood disorders, and other common single-gene disorders.

31. A 25-year-old patient comes to you with results from their “23 and me” testing showing a high likelihood of developing coronary artery disease (CAD). What healthcare education do you provide?

a. The genes you have inherited put you at increased risk for CAD; there is no treatment to decrease that risk.

@ Incorrect answer. Overall, CAD risk can be decreased by lifestyle changes to decrease modifiable risk despite an elevated inherited risk.

*b. The genes you have inherited put you at increased risk for CAD; it is important to make lifestyle changes to minimize risk.

@ Correct answer. Overall, CAD risk can be decreased by lifestyle changes to decrease modifiable risk despite an elevated inherited risk.

c. The genes you have inherited put you at increased risk for CAD, so you should have gene therapy to decrease overall CAD risk.

@ Incorrect answer. There are currently no gene therapies for CAD gene risk.

d. The genes you have inherited put you at increased risk for CAD, but since you are below the age of 40 you can wait to begin statin therapy to decrease overall CAD risk.

@ Incorrect answer. Someone with greater risk of developing CAD should commence lifestyle changes and have more frequent assessment of blood lipids.

Essay Questions

Type: E

1. Differentiate between autosomal, sex-linked, and mitochondrial disorders.

   SUGGESTED ANSWER: Autosomal disorders are passed from both parents as a mutation on one of the 22 chromosomes. Because offspring inherit one chromosome from each parent to form a new pair, dominant disorders are more readily passed than recessive disorders. Sex-linked disorders are carried on the X or Y chromosome. Mitochondrial disorders are only inherited from the mother because sperm contributes little to no mitochondrial DNA.

Type: E

2. Explain the difference between penetrance and expressivity.

   SUGGESTED ANSWER: Penetrance involves the presence of a mutated gene without the phenotype of the disease. Reduced penetrance can occur with both autosomal dominant and autosomal recessive

disorders. Reduced penetrance may be due to epigenetic factors, gene–environment interaction, age, sex, and other modifiers. Expressivity is a variability in the expression of the phenotype. For example, cystic fibrosis is 100% penetrant, meaning that every individual with the gene will have the disorder; however, it also has variable expressivity. The signs and symptoms of the disorder will vary depending on the exact mutation present. Phenylketonuria (PKU) is another example of a disease that is 100% penetrant, but a diet modification can prevent the expression of the gene from becoming severe.

Type: E

3. Explain the mechanism by which a single gene can code for more than one protein.

   SUGGESTED ANSWER: Many genes include sequences of DNA that are either transcribed and translated (exons), alternating with intervening sequences (introns) that are transcribed, but are then removed from the final mature messenger RNA (mRNA). If a gene has multiple exons and introns, alternative splicing of the pre-mRNA can generate more than one final mRNA. Translation of these different mRNA versions produces different proteins.

Type: E

4. Describe the difference between a messenger RNA (mRNA) vaccine and a traditional protein-based vaccine.

   SUGGESTED ANSWER: mRNA vaccines inject mRNA encoding a viral protein, prompting human cells to produce that protein and provoking their immune system to make antibodies to the viral protein. Traditional protein-based vaccines inject a portion of the virus, prompting an immune response.

Chapter 4

Cell Physiology and Pathophysiology

Multiple Choice Questions

1. Which of the following best describes most human cells?

   a. The cell membrane consists of a phospholipid bilayer with nonpolar components facing the outside.

   @ Incorrect answer. The cell membrane consists of a phospholipid bilayer with polar components facing the outside.

   *b. Small nonpolar substances can move across the membrane's lipid barrier.

   @ Correct answer. The cell membrane's lipid barrier allows small nonpolar substances to move between extracellular and intracellular fluid, such as oxygen and carbon dioxide.

   c. The membrane forms a hydrophilic barrier between intracellular fluid and extracellular fluid.

   @ Incorrect answer. The membrane forms a hydrophobic barrier between intracellular and extracellular fluid.

   d. Most molecules diffuse easily through the membrane's lipid barrier.

   @ Incorrect answer. Most molecules require transporters to cross the cell membrane.

2. Which of the following organelles is made in the nucleus and attach to messenger RNA (mRNA) for protein synthesis?

   a. Rough endoplasmic reticulum (ER)

   @ Incorrect answer. Rough ER is contiguous with the nucleus and is where proteins are folded.

   b. Smooth ER

   @ Incorrect answer. Smooth ER is connected to rough ER and holds enzymes for metabolic processes.

c. Golgi apparatus

@ Incorrect answer. The function of the Golgi apparatus is to sort, modify, and package proteins and lipids.

*d. Ribosome

@ Correct answer. The ribosome is manufactured in the nucleus. It has large and small subunits that attach to mRNA for protein synthesis.

3. Which of the following organelles functions in energy production with H_2O_2 as a common by-product?

a. Lysosomes

@ Incorrect answer. Lysosomes are involved in bacterial killing and cell death pathways. They contain degradative enzymes.

b. Mitochondria

@ Incorrect answer. The mitochondria produce adenosine triphosphate (ATP), the energy source for cellular chemical reactions and ion transport. They consume oxygen, with carbon dioxide as a common by-product.

*c. Peroxisomes

@ Correct answer. Peroxisomes use oxygen for enzymatic reactions. β oxidation of fatty acids occurs in peroxisomes, generating energy and producing the by-product of hydrogen peroxide.

d. Smooth endoplasmic reticulum

@ Incorrect answer. The smooth endoplasmic reticulum (ER) contains enzymes for metabolic processes.

4. Which process does oxygen use to move into cells with low oxygen levels?

*a. Simple diffusion

@ Correct answer. Simple diffusion is the movement of a substance across a barrier without a specific transport protein. It occurs down a concentration gradient. Oxygen moves from a relatively high concentration in the bloodstream to a relatively low concentration within cells.

b. Endocytosis

@ Incorrect answer. Endocytosis is when part of the extracellular fluid is engulfed by the cell membrane, pinched off, and moved into the cell. Phagocytic cells use this process to destroy bacteria.

c. Facilitated diffusion

@ Incorrect answer. Facilitated diffusion is used by most polar and hydrophilic substances to move across a cell membrane. Movement of the substance down their gradient requires the assistance of a protein carrier. Oxygen does not need the assistance of a protein carrier to diffuse across the cell membrane.

d. Primary active transport

@ Incorrect answer. Primary active transport moves a substance against its concentration gradient, requiring the use of energy.

5. Drugs that inhibit gastric acid secretion target which of the following types of transport?

a. Osmosis

@ Incorrect answer. Drugs that inhibit gastric acid secretion target proton pumps, or potassium–hydrogen ATPase, a pump that moves hydrochloric acid against its concentration gradient. This is a primary active transport pump.

b. Facilitated diffusion

@ Incorrect answer. Drugs that inhibit gastric acid secretion target proton pumps, or potassium–hydrogen ATPase, a pump that moves hydrochloric acid against its concentration gradient. This is a primary active transport pump.

*c. Primary active transport

@ Correct answer. Drugs that inhibit gastric acid secretion target proton pumps, or potassium–hydrogen ATPase, a pump that moves hydrochloric acid against its concentration gradient. This is a primary active transport pump.

d. Ion channels

@ Incorrect answer. Drugs that inhibit gastric acid secretion target proton pumps, or potassium–hydrogen ATPase, a pump that moves hydrochloric acid against its concentration gradient. This is a primary active transport pump.

6. Which of the following is the major transport mechanism for water in the kidney cells?

a. Diffusion

@ Incorrect answer. Water moves through aquaporins in kidney cells under the influence of osmotic pressure, not by diffusion.

b. Facilitated diffusion

@ Incorrect answer. Water moves through aquaporins in kidney cells under the influence of osmotic pressure, not by facilitated diffusion.

c. Secondary active transport

@ Incorrect answer. Water moves through aquaporins in kidney cells under the influence of osmotic pressure, not by secondary active transport.

*d. Aquaporins

@ Correct answer. Water moves through aquaporins in kidney cell membranes under the influence of osmotic pressure.

7. Which of the following is a characteristic of G protein–coupled receptors (GPCRs)?

a. Primary messengers turn on protein kinase enzymes to change the function of intracellular proteins.

@ Incorrect answer. Second messengers turn on protein kinase enzymes to change the function of intracellular proteins to meet the changing needs of the body.

b. Phosphorylation only increases the activity of proteins.

@ Incorrect answer. Phosphorylation of some proteins will increase their activity. Phosphorylation of other proteins decreases their activity.

*c. Inhibitory receptors limit the strength and duration of the effects of GPCRs.

@ Correct answer. Inhibitory receptors and off-switches limit the strength and duration of GPCR effects.

d. The action of second messengers is terminated by off-switches.

@ Incorrect answer. Second messengers are metabolized, which terminates their action.

8. Which of the following correctly explains the mechanism of action of nitric oxide?

a. Nitric oxide diffuses into the smooth muscle to decrease cyclic guanosine monophosphate (cGMP).

@ Incorrect answer. Nitric oxide diffuses into the smooth muscle cells to increase cGMP and cause vasodilation and lower blood pressure.

b. Nitric oxide is taken into smooth muscle cells via active transport and increases cGMP.

@ Incorrect answer. Nitric oxide enters smooth muscle cells via diffusion, not active transport.

c. Nitric oxide enters smooth muscle cells through aquaporins and decreases cGMP.

@ Incorrect answer. Nitric oxide enters smooth muscles via diffusion and increases cGMP.

*d. Nitric oxide diffuses into the smooth muscle to cause vasodilation.

@ Correct answer. Nitric oxide diffuses into the smooth muscle cells to increase cGMP and cause vasodilation and lower blood pressure.

9. Epidermal growth factor receptor (EGFR) is an enzyme-linked receptor. Which of the following is true regarding EGFR?

*a. EGFR is mutated in many epithelial tissue cancers.

@ Correct answer. EGFR is mutated in many cancers, especially those involving epithelial tissues, such as the colon and breast.

b. EGFR inhibits cell signaling pathways that alter gene expression.

@ Incorrect answer. EGFR activates cell signaling pathways that alter gene expression, transcription, and translation to increase rates of cell division.

c. EGFR is commonly inhibited in breast cancer.

@ Incorrect answer. EGFR is commonly mutated in breast cancer.

d. EGFR is active only in adulthood.

@ Incorrect answer. EGFR is needed during embryonic and fetal development.

10. Activation of an epidermal growth factor receptor (EGFR) is followed by cell activity changes due to:

a. Phosphorylation of cell proteins on serine residues

@ Incorrect answer. G protein–coupled receptors (GPCR)-associated protein kinases phosphorylate on serine and threonine residues.

b. Phosphorylation of cell proteins on threonine residues

@ Incorrect answer. GPCR-associated protein kinases phosphorylate on serine and threonine residues.

c. Phosphorylation of cell proteins on tryptophan residues

@ Incorrect answer. Tryptophan is not a target of protein kinase phosphorylation.

*d. Phosphorylation of cell proteins on tyrosine residues

@ Correct answer. EGFR and other enzyme-linked receptors are tyrosine kinases.

11. What is the major outcome when a nuclear receptor for thyroid hormone is activated by thyroid hormone binding?

a. A three-part G protein dissociates into α and βγ subunits.

@ Incorrect answer. Membrane-associated G proteins are involved in actions of membrane-bound G protein–coupled receptors.

*b. Synthesis of new proteins is initiated.

@ Correct answer. Cytosolic and nuclear receptors bind to DNA when activated, initiating transcription and translation of new cell proteins.

c. Calcium is released from the endoplasmic reticulum.

@ Incorrect answer. Calcium is released from the endoplasmic reticulum by inositol triphosphate (IP_3) as part of phospholipase C-linked signaling.

d. Levels of cyclic guanosine monophosphate (cGMP) increase.

@ Incorrect answer. Levels of cGMP increase in vascular smooth muscle cells when guanylyl cyclase is activated by the vasodilators nitric oxide or natriuretic peptides.

12. How is the strength of contraction in striated muscle cells modulated?

a. It is modulated by the amount of calcium in the cytoplasm.

@ Incorrect answer. The strength of the contraction is modulated by muscle fiber length at the start of contraction and weight of the load.

b. Prior shortening to rest the sarcomeres strengthens the contraction.

@ Incorrect answer. Prior lengthening brings more sarcomeres to an optimal length for crossbridge cycling and strengthens the contraction.

c. A smaller load decreases the strength and speed of the contraction.

@ Incorrect answer. The greater the load, the slower the speed of contraction.

*d. Initial stretching generates a greater force of contraction.

@ Correct answer. Initial stretching generates a greater force of contraction.

13. Which of the following is most likely a cause of frailty in older adults?

a. Disuse atrophy

@ Incorrect answer. Disuse atrophy occurs due to a decrease in workload, usually due to an acute injury.

b. Fracture

@ Incorrect answer. A fracture is an acute limb injury that can result in disuse atrophy.

*c. Sarcopenia

@ Correct answer. Sarcopenia is a component of frailty in older adults that results from the increase of muscle protein breakdown as compared with synthesis.

d. Change in the length–tension relationship

@ Incorrect answer. The length–tension relationship describes the relationship between the length of a muscle prior to contraction and the tension it can develop during contraction.

14. How does relaxation of smooth muscle occur?

a. Receptor stimulation

@ Incorrect answer. When reception stimulation or depolarization stops, relaxation of smooth muscles will occur.

*b. Reuptake of calcium by the sarcoplasmic reticulum

@ Correct answer. When the sarcoplasmic reticulum calcium pump reduces the intracellular calcium, smooth muscle contraction will stop.

c. Increase in myosin light chain kinase activity

@ Incorrect answer. A decrease in myosin light chain kinase activity leads to smooth muscle relaxation.

d. Decreased production of cyclic adenosine monophosphate (cAMP) and cyclic guanosine monophosphate (cGMP)

@ Incorrect answer. cAMP and cGMP mediate smooth muscle relaxation.

15. In which type of cell death do cells undergo a hypoxic insult and become acidic, lose adenosine triphosphate (ATP) production, and accumulate reactive oxygen species that damage membrane lipids, cell proteins, and nucleic acids?

a. Apoptosis

@ Incorrect answer. Apoptosis is a programmed cell death that is part of a normal cell turnover.

b. Autophagy

@ Incorrect answer. Autophagy is a cellular adaptation to malnutrition. Organelles are digested into building blocks or, if the lack of nutrients is severe enough, the cell itself may be broken down.

*c. Necrosis

@ Correct answer. Necrosis is a result of hypoxic or ischemic insult. There is rapid loss of ATP production, depressed Na+/K+ pump activity, intracellular calcium overload, cell swelling, membrane rupture, loss of antioxidant function, accumulation of reactive oxygen species, acidosis, and activation of degradative enzymes.

d. Death program

@ Incorrect answer. The death program is the step followed in apoptosis.

16. The process of apoptosis is associated with:

*a. Cell shrinking and nuclear fragmentation

@ Correct answer. In apoptosis, cells get smaller and the nucleic acids are reduced to fragments and packaged.

b. Cell membrane rupture and leakage of cell contents

@ Incorrect answer. Cell membrane rupture occurs in necrosis.

c. Pathological damage and injury to tissue

@ Incorrect answer. Severe pathological damage and tissue injury causes necrosis.

d. Acute tissue inflammation

@ Incorrect answer. Necrotic cell death is associated with inflammation.

17. One hallmark of aging is theorized to involve increased sirtuin levels and reduced insulin signaling. Which of the following terms fits this definition?

a. Loss of proteostasis

@ Incorrect answer. Loss of proteostasis is the inability to correctly fold proteins or to degrade those that have been misfolded.

*b. Deregulated nutrient sensing

@ Correct answer. Deregulated nutrient sensing involves increased sirtuin levels and decreased insulin signaling.

c. Stem cell exhaustion

@ Incorrect answer. Stem cell exhaustion occurs when stem cells are no longer able to reproduce.

d. Cellular senescence

@ Incorrect answer. Cellular senescence occurs when cells that once had the ability to replicate are no longer able to do so.

18. Which of the following is a protein component of the cell membrane?

a. Cholesterol

@ Incorrect answer. Cholesterol is a lipid component of the cell membrane.

b. Ribosomal RNA

@ Incorrect answer. Ribosomal RNA is part of an intracellular ribosome.

c. Phosphatidylcholine

@ Incorrect answer. Phosphatidylcholine is a membrane phospholipid.

*d. Sodium–potassium pump

@ Correct answer. The sodium–potassium pump is an integral membrane transport protein.

19. Which compound has the greatest probability of crossing the cell membrane by simple diffusion?

*a. Carbon dioxide

@ Correct answer. Carbon dioxide is a small nonpolar molecule that can easily cross the hydrophobic membrane core.

b. Glucose

@ Incorrect answer. Glucose is a small, very polar molecule that attracts surrounding water molecules and cannot cross the hydrophobic membrane core.

c. Glutamate (an acidic amino acid)

@ Incorrect answer. Glutamate is a small, charged and polar molecule that attracts surrounding water molecules and cannot cross the hydrophobic membrane core.

d. Water

@ Incorrect answer. Water is a small, polar molecule that is attracted to neighboring water molecules by hydrogen bonds; it is unable to cross the hydrophobic membrane core.

20. What is the most common way for proteins to cross the cell membrane?

a. Active transport

@ Incorrect answer. Active transport proteins are responsible for ion transport.

*b. Endocytosis or exocytosis

@ Correct answer. Proteins are large molecules that generally move into or out of cells by endocytosis or exocytosis, respectively.

c. Facilitated diffusion

@ Incorrect answer. Facilitated diffusion transporters are responsible for moving small metabolites like glucose, fructose, and amino acids across the cell membrane.

d. Simple diffusion

@ Incorrect answer. Simple diffusion across the cell membrane is only possible for small, nonpolar, hydrophobic molecules.

21. One action of the hormone vasopressin is to increase water reabsorption in kidney tubules by increasing insertion of these transporters into cell membranes:

*a. Aquaporins