Chapter 2: Chemistry of life

2.3 Molecules of life

- Carbohydrates, lipids, proteins, and nucleic acids are organic molecules with specific functions in cells.
- Dehydration reactions form macromolecules from their building blocks. Hydrolysis reactions break down macromolecules.

2.4 Carbohydrates

- Carbohydrates are short-term energy storage molecules.
- Simple carbohydrates are monosaccharides or disaccharides. Glucose is a monosaccharide used by cells for quick energy.
- Complex carbohydrates are polysaccharides. Starch, glycogen, and cellulose (fiber) are polysaccharides containing many glucose units.
- Plants store glucose as starch, whereas animals store glucose as glycogen. Cellulose forms plant cell walls. Cellulose is dietary fiber. Fiber plays an important role in digestive system health.

2.5 Lipids

- Lipids are nonpolar molecules that do not dissolve in water. Fats, also called triglycerides, and oils are lipids that act as long-term energy storage molecules.
- Fatty acids can be saturated or unsaturated. Trans fats are unsaturated fatty acids that have adverse effects on your health.
- Plasma membranes contain phospholipids.
- Steroids are complex lipids composed of four interlocking rings. Testosterone and estrogen are steroids. Cholesterol is a steroid that is transported by proteins called lipoproteins (LDLs and HDLs)

2.6 Proteins

- Proteins may be structural proteins (keratin, collagen), hormones, or enzymes that speed chemical reactions. Proteins account for cell movement (actin, myosin), enable muscle contraction (actin, myosin), or transport molecules in blood (hemoglobin).

- Proteins are macromolecules with amino acid subunits. A peptide is composed of two amino acids linked by a peptide bond, and a polypeptide contains many amino acids.
- A protein has levels of structure: A primary structure is determined by the sequence of amino acids that forms a polypeptide. A secondary structure is an α (alpha) helix. A tertiary structure occurs when the secondary structure forms a three-dimensional, globular shape. A quaternary structure occurs when two or more polypeptides join to form a single protein. Denaturation represents an irreversible change in the shape of a protein.

2.7 Nucleic Acids

- Nucleic acids are macromolecules composed of nucleotides. Nucleotides are composed of a sugar, a base, and a phosphate. DNA and RNA are polymers of nucleotides.
- DNA (deoxyribonucleic acid) contains the sugar deoxyribose; contains the bases adenine (A), guanine (G), thymine (T), and cytosine (C); is double-stranded; and forms a helix. The helix exhibits complementary base pairing between the strands of DNA.
- RNA (ribonucleic acid) contains the sugar ribose; contains the bases adenine, guanine, uracil (U), and cytosine; and does not form a helix.
- ATP (adenosine triphosphate) is a high-energy molecule, because its bonds are unstable.
- ATP undergoes hydrolysis to ADP (adenosine diphosphate) + P , which releases energy used by cells to do metabolic work.

Chapter 3: Cell structure and function

3.1 What Is a Cell?

- The cell theory states that cells are the basic units of life and that all life comes from preexisting cells. Microscopes are used to view cells, which must remain small to have a favorable surface-area-to-volume ratio.

3.2 How Cells Are Organized?

- The human cell is a eukaryotic cell with a nucleus that contains the genetic material. Prokaryotic cells, such as the bacteria, are smaller than eukaryotic cells and lack a nucleus. The cell is surrounded by a plasma membrane, a selectively permeable barrier that limits the movement of materials into and out of the cell. Between the plasma

membrane and the nucleus is the cytoplasm. In eukaryotic cells, the cytoplasm contains various organelles, each with specific functions.

3.3 The Plasma Membrane and How Substances Cross It

- The fluid-mosaic model describes the structure of the plasma membrane. The plasma membrane contains:
    1. A phospholipid bilayer that selectively regulates the passage of molecules and ions into and out of the cell.
    2. Embedded proteins, which allow certain substances to cross the plasma membrane.
- Passage of molecules into or out of cells can be passive or active:
    1. Passive mechanisms do not require energy. Examples are diffusion, osmosis, and facilitated transport. Tonicity and osmotic pressure control the process of osmosis.
    2. Active mechanisms require an input of energy. Examples are active transport (sodium-potassium pump), endocytosis (phagocytosis and pinocytosis), receptor-mediated endocytosis, and exocytosis.

3.4 The nucleus and the endomembrane system

- The nucleus:
    1. The nucleus houses DNA, which contains genes that specify the order of amino acids in proteins. Chromatin is a combination of DNA molecules and proteins that make up chromosomes.
    2. The nucleus is surrounded by a nuclear envelope that contains nuclear pores for communication and the movement of materials.
    3. The nucleolus produces ribosomal RNA (rRNA).
    4. Protein synthesis occurs in ribosomes, small organelles composed of proteins and rRNA.
- The endomembrane System:

  The endomembrane system consists of the nuclear envelope, endoplasmic reticulum (ER), Golgi apparatus, lysosomes, and vesicles:
    1. The rough ER has ribosomes, where protein synthesis occurs.
    2. Smooth ER has no ribosomes and has various functions, including lipid synthesis.

3. The Golgi apparatus processes and packages proteins and lipids into vesicles for secretion or movement into other parts of the cell.
4. Lysosomes are specialized vesicles produced by the Golgi apparatus. They fuse with incoming vesicles to digest enclosed material, and they auto digest old cell parts.

3.5 The Cytoskeleton, Cell Movement, and Cell Junctions

- The cytoskeleton consists of microtubules, actin filaments, and intermediate filaments that give cells their shape; and it allows organelles to move about the cell. Microtubules are organized by centrosomes. Cilia and flagella, which contain microtubules, allow a cell to move.
- Cell junctions connect cells to form tissues and to facilitate communication between cells.
- The extracellular matrix (ECM) is located outside the plasma membrane. It may provide structure and regulate the movement of materials into the cell.

3.6 Metabolism and the Energy Reactions

- Metabolic Pathways:
  - Metabolism represents all the chemical reactions that occur in a cell. A metabolic pathway is a series of reactions, each of which has its own enzyme. The materials entering these reactions are called reactants, and the materials leaving the pathway are called products.
- Enzymes:
  - Enzymes bind their substrates in the active site.
  - Enzymes accelerate chemical reactions by lowering the energy of activation (Ea) needed to start the reaction.
  - Coenzymes, such as NAD+ (nicotinamide adenine dinucleotide), are nonprotein molecules that assist enzymes.
- Mitochondria and Cellular Respiration:
  - Mitochondria are involved in cellular respiration, which uses oxygen and releases carbon dioxide.
  - During cellular respiration, mitochondria convert the energy of glucose into the energy of ATP molecules.
- Cellular Respiration and Metabolism
  - Cellular respiration includes three pathways: glycolysis, the citric acid cycle, and the electron transport chain.
  - Glycolysis occurs in the cytoplasm and is anaerobic. It produces two pyruvate molecules and small amount of ATP and NADH.

- The pyruvate molecules are modified by the preparatory reactions in the mitochondria before entering the citric acid cycle.
- The citric acid cycle occurs in the matrix of the mitochondria. Its role is to break C—C bonds and generate ATP, NADH, and FADH2.
- The electron transport chain is located along the cristae of the mitochondria. It is an aerobic pathway that uses the electrons in the NADH and FADH2 molecules to generate the majority of the ATP in the cell.
- If oxygen is not available in cells, the electron transport chain is inoperative, and fermentation (which does not require oxygen) occurs. Fermentation recycles NAD+ molecules so that the cell can produce a small amount of ATP by glycolysis.

Chapter 19: Patterns of Chromosome Inheritance

19.1 Chromosomes

- The genetic material of the cell is organized as chromosomes. Chromosomes contain a combination of proteins and DNA called chromatin.
- Most human cells are diploid—therefore, chromosomes occur in pairs.
- Prior to mitosis, or duplication division, the chromosomes are replicated, forming sister chromatids. The sister chromatids are joined at the centromere.
- A karyotype is a visual display of an individual's chromosomes.

19.2 The Cell Cycle

- The cell cycle occurs continuously and has several stages: G1, S, G2 (the interphase stages), and M (the mitotic stage), which includes cytokinesis and the stages of mitosis.
    1. In G1, a cell doubles organelles and accumulates materials for DNA synthesis.
    2. In S, DNA replication occurs.
    3. In G2, a cell synthesizes proteins needed for cell division.
- Checkpoints and external signals control the progression of the cell cycle. Cells that fail to pass checkpoints may enter G0 phase and undergo apoptosis.

19.3 Mitosis

- Mitosis is duplication division that ensures that the daughter cells have the diploid (2n) number and the same types of chromosomes as the parent cell. The mitotic spindle plays an important role in the separation of the sister chromatids during mitosis. The mitotic spindle is organized by the centrosomes of the cell. Centrosomes contain clusters of microtubules called centrioles.

The phases of mitosis are prophase, prometaphase, metaphase, anaphase, and telophase:

1. Prophase. The nucleus dissolves and the chromosomes condense.
2. Prometaphase. Chromosomes attach to spindle fibers.
3. Metaphase. Chromosomes align at the equator.
4. Anaphase. Chromatids separate, becoming chromosomes that move toward the poles.
5. Telophase. Nuclear envelopes form around chromosomes; cytokinesis begins.
6. Cytokinesis is the division of cytoplasm and organelles following mitosis. Cytokinesis in animal cells involves the formation of a cleavage furrow to separate the cytoplasm.

19.4 Meiosis

- Meiosis is reduction division that reduces the diploid (2n) chromosome number to a haploid (n) number. Meiosis involves two cell divisions— meiosis I and meiosis II.
- Meiosis I
    - Homologous chromosomes pair (synapsis) and then separate. Interkinesis follows meiosis I.
- Meiosis II
    - Sister chromatids separate, resulting in four cells with the haploid number of chromosomes that move into daughter nuclei.
    - Meiosis results in genetic recombination due to crossing-over; gametes have all possible combinations of chromosomes. Upon fertilization, the zygote is restored to a diploid number of chromosomes.
- Spermatogenesis and Oogenesis
- Spermatogenesis.
    - In males, spermatogenesis produces four viable sperm.
    - Oogenesis. In females, oogenesis produces one egg and several polar bodies. Oogenesis goes to completion if the sperm fertilizes the developing egg.

19.5 Comparison of Meiosis and Mitosis

- In prophase I, homologous chromosomes pair; there is no pairing in mitosis.
- In metaphase I, homologous duplicated chromosomes align at equator.
- In anaphase I, homologous chromosomes separate.

19.6 Chromosome Inheritance

Meiosis is a part of gametogenesis (spermatogenesis in males and oogenesis in females) and contributes to genetic diversity.

- Changes in Chromosome Number
    - Nondisjunction changes the chromosome number in gametes, resulting in trisomy (2n + 1) or monosomy (2n – 1).
    - Autosomal syndromes include Down syndrome.
- Changes in Sex Chromosome Number
    - Nondisjunction during oogenesis or spermatogenesis can result in gametes that have too few or too many X or Y chromosomes.
    - If more than one X chromosome is present in a cell, a Barr body may be formed.
    - Syndromes include Turner, Klinefelter, poly-X, and Jacobs.
- Changes in Chromosome Structure
    - Chromosomal mutations can produce deletions, duplications, inversions, and translocations.
    - These result in various syndromes, such as Williams, cri du chat (deletion), and Alagille, and certain cancers (translocation).

Chapter 21: Patterns of Genetic Inheritance

21.1 Genotype and Phenotype

An allele is a variation of a gene. Each allele exists at a specific locus on a chromosome. Genotype refers to the alleles of the individual, and phenotype refers to the physical characteristics associated with these alleles. Dominant alleles mask the expression of recessive alleles.

- Homozygous dominant individuals have the dominant phenotype (e.g., AA = normal pigmentation).
- Homozygous recessive individuals have the recessive phenotype (e.g., aa = albino).
- Heterozygous individuals have the dominant phenotype (e.g., Aa = normal pigmentation).

21.2 One- and Two-Trait Inheritance

- One-Trait Crosses
    1. The first step in doing a problem with a one-trait cross, or monohybrid cross, is to determine the genotype and then the gametes.
        - An individual has two alleles for every trait, but a gamete has one allele for every trait.
    2. The next step is to combine all possible sperm with all possible eggs. If there are more than one possible sperm and/or egg, a Punnett square is helpful in determining the genotypic and phenotypic ratio among the offspring.
        - For a monohybrid cross between two heterozygous individuals, a 3:1 ratio is expected among the offspring.
        - For a monohybrid cross between a heterozygous and homozygous recessive individual, a 1:1 ratio is expected among the offspring.
        - The expected ratio can be converted to the chance of a particular genotype or phenotype. For example, a 3:1 ratio = a 75% chance of the dominant phenotype and a 25% chance of the recessive phenotype.
- Two-Trait Crosses
    - A problem consisting of two traits is often referred to as a dihybrid cross.
    - If an individual is heterozygous for two traits, four gamete types are possible, as can be substantiated by knowledge of meiosis.
    - For a cross between two heterozygous individuals (AaBb × AaBb), a 9:3:3:1 ratio is expected among the offspring.
    - For a cross between a heterozygous and homozygous recessive individual (AaBb × aabb), a 1:1:1:1 ratio is expected among the offspring.

21.3 Inheritance of Genetic Disorders

- A pedigree shows the pattern of inheritance for a trait from generation to generation of a family. This first pattern appears in a family pedigree for a recessive disorder—both parents are carriers. The second pattern appears in a family pedigree for a dominant disorder. Both parents are again heterozygous.

- Genetic Disorders of Interest
    - Tay-Sachs disease, cystic fibrosis (CF), and sickle-cell disease are autosomal recessive disorders.
    - Marfan syndrome, osteogenesis imperfecta, and Huntington disease are autosomal dominant disorders.

21.4 Beyond Simple Inheritance Patterns

In some patterns of inheritance, the alleles are not just dominant or recessive.

- Polygenic Inheritance
    - Polygenic traits, such as skin color and height, are controlled by more than one set of alleles. The alleles have an additive effect on the phenotype. Multifactorial traits are usually polygenic with an environmental influence.
- Incomplete Dominance and Codominance
    - In incomplete dominance (e.g., familial hypercholesterolemia), the heterozygote is intermediate between the two homozygotes. In codominance (e.g., type AB blood), both dominant alleles are expressed equally.
- Multiple-Allele Inheritance

  In humans, an example of a trait involving multiple alleles is the ABO blood types. Every individual has two out of three possible alleles: IA, IB, or i. Both IA and IB are expressed. Therefore, this is also a case of codominance.

21.5 Sex-Linked Inheritance

- X-Linked Alleles
- Humans contain 22 pairs of autosomes and 1 pair of sex chromosomes. Traits on the sex chromosomes are said to be sex-linked. X-linked traits, such as those that determine normal vision as opposed to color blindness, are unrelated to the gender of the individual. Common X-linked genetic crosses are:
    - XBXb × XBY: All daughters will be normal, even though they have a 50% chance of being carriers, but sons will have a 50% chance of being color-blind.
    - XBXB × XbY: All children will be normal (daughters will be carriers).

- Pedigree for X-Linked Disorders
    - A pedigree for an X-linked recessive disorder shows that the trait often passes from grandfather to grandson by way of a carrier daughter. Also, more males than females have the characteristic.
    - Like most X-linked disorders, color blindness, Duchenne muscular dystrophy, fragile X syndrome, and hemophilia are recessive.

Chapter 22: DNA Biology and Technology

22.1 DNA and RNA Structure and Function

- DNA (deoxyribonucleic acid) is the genetic material. It is organized as genes, located on chromosomes. DNA replicates, stores information, and mutates for genetic variability.
- Structure of DNA
    - DNA is a double helix composed of two polynucleotide strands. Each nucleotide is composed of a deoxyribose sugar, a phosphate, and a nitrogen-containing base (A, T, C, G).
    - Complementary base pairing occurs between the strands of DNA. The base A is bonded to T, and G is bonded to C.
- Replication of DNA

  During DNA replication, the DNA strands unzip, and a new complementary strand forms opposite each old strand (the template), resulting in two identical DNA molecules. Mutations produce variation in the genetic material.
- The Structure and Function of RNA
    - RNA (ribonucleic acid) is a single-stranded nucleic acid in which the base U (uracil) occurs instead of T (thymine).
    - The four primary forms of RNA are ribosomal RNA (rRNA), messenger RNA (mRNA), transfer RNA (tRNA), and small RNAs.

22.2 Gene Expression

Gene expression leads to the formation of a product, either an RNA or a protein. Proteins differ by the sequence of their amino acids. Gene expression for proteins requires transcription and translation.

- Transcription:

  Transcription occurs in the nucleus using an enzyme called RNA polymerase. The three-base DNA codon is passed to an mRNA that contains codons. Introns are removed from mRNA during mRNA processing.
- Translation:

  Translation occurs in the cytoplasm at the ribosomes. The tRNA molecules bind to their amino acids, and then their anticodons pair with mRNA codons.
- The Regulation of Gene Expression

  Regulation of gene expression occurs at five levels in a human cell.
    - Pretranscriptional control: In the nucleus; the DNA is made available to transcription factors and enzymes
    - Transcriptional control: In the nucleus; the degree to which a gene is transcribed into mRNA determines the amount of gene product. Transcription factors are involved at this stage.
    - Posttranscriptional control: In the nucleus; involves mRNA processing and how fast mRNA leaves the nucleus.
    - Translational control: In the cytoplasm; affects when translation begins and how long it continues; includes inactivation and degradation of mRNA.
    - Posttranslational control: In the cytoplasm; occurs after protein synthesis.