FINALS REVIEW SHEET:

Biology is the study of life.

Scientists formulate **hypotheses** which are **tested by experiments**. A hypothesis is a supposition or **proposed explanation** drawn from limited evidence as a starting point for further investigation which needs to be validated with experiments. You observe that flashlight doesn't work, so you propose that it must be dead batteries? This must be tested. It may be due to dead batteries, but it might be a burned out or defective light bulb. Experiments need controls and test one variable. Clinical trials for an experimental drug must include a cohort of study patients comprising the control group who do not take the drug, but take a placebo or 'fake pill.' Furthermore, the data must be analyzed by a third party who is not invested in the results of the clinical trial or confounded by bias. The study participants in the experimental and control group are not revealed to the third party. This is called a 'double blind' study.

A scientific paper has **4 sections**: 1) Introduction section in which hypotheses might be stated and background provided, 2) Materials and Methods section delineates equipment, reagents, and experimental procedures 3) Results section of data including tables, graphs, etc. 4) Discussion section in which results are compared to existing published data and inferences are drawn.

The **nucleus** of an atom contains **proton(p) and neutron(n)** surrounded an **electron cloud**. What is **atomic number** (number of protons **only or p**), **atomic weight** (roughly the number of protons plus the neutrons or **p+n**). Usually the atomic weight is roughly **double** the atomic number unless there is an isotope (different number of neutrons to protons.)**.** The mass of an electron is infinitesimally small compared to protons and neutrons. - Electrons are similar to tiny moons orbiting around a large planet.

What is meant by energy levels or shells? First shell has 2 electrons, next shell has 8 electrons to fill the shell. The **outermost** electron shell is the most reactive. As two atoms approach, the outermost electrons of each atom may interact.

Differences amongst three types of bonds **1) covalent, 2) ionic, and 3) hydrogen bonding**?

Electrons are **shared in covalent bonding**. Example Carbon dioxide gas, CO_2. These bonds tend to be the strongest and are charge neutral.

On the other hand, **ionic bonds**, the electrons are lost from the donor atom (resulting in a positive charge) to the recipient atom (resulting in a negative charge). In other words, electrons are **transferred** between atoms. For an ionic bond, the atoms are in close proximity to each other since there is an attraction between the positive and negative charges, but the electrons are **NOT shared** between the atoms as in covalent bonds. An example of an ionic bond is table salt or NaCl.

Water is slightly polar, oxygen has a partial positive charge and a partial negative charge, hence **hydrogen bonding** can occur between the hydrogen atom of one water molecule and the oxygen atom of another water molecule. Hydrogen bonding exists amongst water molecules. The hydrogen bonding between the nucleotides (bases) of the DNA double helix keeps the strands together. Water Molecules which are polar are attracted to water's partial negative and positive charge or are **Hydrophilic** thus may be **solubilized** (i.e., salt and sugar dissolve in water). Molecules which are not attracted to water are **hydrophobic** including **nonpolar** substances such as oxygen gas and many lipids (most fats are not soluble in water).

Hydrogen bonding is a critical component for life on earth because of the **intrinsic properties of water** such as the following: water's high surface tension, adhesion of water to other materials, cohesion of water molecules, water's high heat capacity/high heat of vaporization, and water as a 'universal' solvent. Examples: High surface tension- water bugs 'walks on water'; Cohesion- water from roots 'travels' to the top of a tree or capillary action of water 'travels' up a straw. High heat of vaporization- sweating in Houston-our natural A/C; High heat capacity: prevents large fluctuations of temperature in oceans, so aquatic organisms can maintain body temperature; Universal solvent-water can dissolve many molecules such as salt, sugar, proteins, etc. (water is polar due to hydrogen bonding)

What is **pH?** The negative logarithm of the hydrogen proton concentration or -log[H+]. Therefore, acids have a **higher hydrogen proton** concentration than bases. The pH scale is 1-14. A substance with a pH of 7 is neutral, acids have pH < 7, bases have pH>7;

Examples of pH: Stomach secretions (pH2) and Coke(pH3) are **acidic**; whereas, seawater (pH 8) and oven cleaner (pH13) are **basic.** Blood pH is 7.4 or slightly basic or alkaline. Why is an acid of pH 2, 1000 times more acidic than an acid of pH 5? Logarithm scale. If you mix an equal volume (same molarity) of a pH 9 solution with a pH 5 solution, then you should get a neutral solution of pH 7.

Dehydration and Hydration reactions as they pertain to Synthesis and Decomposition Reactions? Dehydration often confers **synthesis or joining of monomers to build a polymers(Anabolic).** The opposite is a hydration reaction in which **polymers are degraded into individual monomer components (Catabolic)**. Typically, energy input is required for anabolic reactions and released during catabolic reactions**.**

What is **homeostasis**? A balance control mechanism such as keeping blood pH at 7.4 or body temperature to 37C or 98.6F degrees. These features are **critical to life**- adaptation, growth, development, and the ability to transfer genetic information from one generation to the next?

Understand the **chief monomers and polymers of carbohydrates**. -Examples of a carbohydrate monomers are the *Monosaccharides*, glucose, and fructose (which incidentally are isomers), as opposed to *Disaccharides* such as table sugar, sucrose or milk sugar, lactose.

++Glucose is the chief monomer for metabolic activity

Polysaccharides are polymers that includes chains of **GLUCOSE** molecules the formed branched structures of **glycogen** (glucose storage for animals), **starch & cellulose** (glucose storage for plants). Table sugar is a disaccharide comprised of two monomers which are isomers to each other- glucose and fructose. Glucose is added to Gatorade/Power Aid.

Difference between saturated and unsaturated fatty acids? **Saturated lipids don't** have double carbon bonds, and saturated fats are solid at room temperature, typically, of animal origin. Trans-saturated fats are very unhealthy. **Unsaturated fats** have double bonds which **kink** the fatty acid chains allowing for increased flexibility and fluidity. Many unsaturated fats are plant origin and are liquid at room temperature. as some vitamins (A,D,E,K). Lipids are often nonpolar and do not dissolve in water. The Phospholipid bilayer of cell membrane has polar fatty acid heads and nonpolar hydrophobic fatty acid tails.

Proteins are comprised of **amino acids** linked by **peptide bonding**.

Carbs are comprised on **monosaccharides** linked by **glycosidic bonding**

The backbone of Nucleic acids (phosphate & ribose sugar) are linked by **phosphodiester bonding.**

ATP is a high-energy compound with unstable bonds.

The cell is the basic functional unit of all life forms (organisms). Note, some organisms are unicellular (just one cell).

Eukaryotic organisms, including fungi, plants, and animal cells have subcellular compartments called **ORGANELLES**. Listed are some organelles:

*The **Nucleus** is the control center (similar to 'city hall' of a town), that contains the genetic blueprint or DNA of the cell which is typically bound to proteins called **chromatin**. The DNA is not 'naked' in the nucleus. The **Nucleolus** is also located in the nucleus and manufactures the ribosomal ribonucleic acid subunits of the ribosome.*

*Adjacent to the nucleus in the cell's cytoplasm lies the **rough endoplasmic reticulum** (ER) that is a network of tubular structures 'studded' with ribosomes (proteins are translated by the ribosome in the rough ER and folded inside the Endoplasmic reticulum compartment). On the other hand, the **smooth ER** does not have attached ribosomes, but generates lipids and carbohydrates, **not** proteins like the rough ER, and helps detoxify poisons and store calcium ions. **Vesicles** transport proteins and other cellular material from one organelle to the next in membrane bound 'balls' that 'bud off' of one organelle and fuse to another organelle. The **Golgi Apparatus** is a cluster of flattened sacs that abuts the ER and is responsible for further chemical modification of proteins such as **glycosylating** proteins or adding carbohydrate side chains to proteins for subsequent exit of proteins out of the cell in a process called **exocytosis**. For example, a protein is formed in the rough ER, packaged in a vesicle, buds off the rough ER and fuses to the cis face (closest to Golgi) of the Golgi Apparatus where it is further modified in the Golgi itself, and eventually another vesicle, then buds off from the trans Golgi (next to the*

cell membrane) and eventually fuses to the cell membrane thereby expelling the contents of the vesicle to the outside of the cell (exocytosis). Please know this sequence of events as proteins are moving from the ribosome to the outside of the cell.

Lysosomes *and* ***peroxisomes*** *have digestive functions that destroy and recycle cellular debris and help digest invading pathogens.* ***Centrosomes*** *are structure that become evident during mitosis and are instrumental to initiate cell division in animal cells. The* ***phospholipid bilayer*** *is the cell membrane is selectively permeable and does not permit everything from entering or leaving the cell.* ***Mitochondria*** *generate ATP and are the powerhouse of the cell (e.g. like an electrical power plant).* ***Cytoskeleton*** *are 'rope-like' in structure and function in maintaining the structure of the cell and play a prominent role in cellular movement as well as vesicular transport (a pseudo-highway system in the cell). The smallest are the* ***microfilaments****, which are concentrated at the periphery of the cell; whereas, the largest diameter of these cellular 'cables' are the microtubules. The* ***microtubules*** *are essential in moving the attached chromosomes during cellular division or mitosis. The* ***intermediate*** *filaments are the most abundant and are critical for the rigidity and architecture of the cell.*

The extracellular matrix (ECM) *or the material outside of the cell is highly organized and its formation is highly regulated which containing proteins and ground substances that provide a scaffolding for cellular attachment. An example of ECM is collagen fibers.*

What is DNA made of? How about RNA? What are some differences?

DNA *is a stable double stranded deoxyribose nucleic acid containing* ***nucleotides*** *comprised of a phosphate head, ribose 5-carbon sugar, and nitrogenous base. The sequence and number of bases determines the genetic blueprint. DNA has 4 bases:* ***C****ytosine,* ***G****uanine,* ***A****denine, and* ***T****hymine. (A pairs with T, and G pairs with C in the double helix). RNA is less stable; single stranded; uses base* ***U****racil instead of thymine; and has 3 broad categories (mRNA or messenger ribonucleic acid, rRNA or ribosomal ribonucleic acid, and tRNA or transfer ribonucleic acid).*

What are the differences between plant cells and animals cell as far as structure?

Plants have a cell wall, chloroplasts for photosynthesis, and a large central vacuole that harbors water and other cell nutrients. Plant cells do ***not*** *have lysosomes or centrioles. Plant cells* ***do*** *have mitochondria, though.*

Bacteria have a cell membrane and cell wall, but ***do not have defined organelles****. They do not have a nuclear membrane. Bacteria have free ribosomes, but do not have any of the organelles mentioned on the previous page.*

Cells have intercellular junctions such as:

Tight junctions*- prevent water loss between adjacent cells,* ***Gap junctions*** *or tiny tunnels between neighboring cells that foster intercellular communication, and* ***Desmosomes*** *or belt-like structures that help bundle and keeps clusters of cells 'fastened together.' Plant cells have*

*perforations or holes in their cell walls analogous to gap junctions called **plasmodesmata** (not present in animal cells) that help nutrients pass from neighboring plant cells.*

Why are cells small? *Answer to **maximize surface area** for nutrient entry and waste exit.*

Concept of **potential** energy versus **kinetic** energy. Examples: Food you digest is potential energy, muscles moving is kinetic.

Define **ATP** as the important 'energy currency of the cell.'

What are the three components of ATP? 1) has **3** phosphates 2) ribose sugar 3) nitrogenous base (adenosine)

Some reactions are **exergonic**, gives off heat. A + B > C + D **+ heat**

Distinguish between **anabolic** (synthesis) versus **catabolic** reactions (degradation). Often catabolic reactions are accompanied by hydrolysis or addition of water; whereas, anabolic reactions are often accompanied by dehydration or loss of water. **Synthesis of complex polymers from simpler monomeric subunits often requires energy (e.g. ATP in living systems).**

Example: The breaking of a bond in ATP resulting in ADP (adenosine **di**phosphate) and a free Phosphate [ATP → ADP + P] during exercise in the muscles liberates heat. The energy is released from the bond between the second and third phosphates.

Some reactions are **endergonic,** absorb heat from surroundings. A + B + **heat** > C + D

Example: Photosynthesis is **endergonic** as well as **anabolic** (synthesis).

Equation of photosynthesis: **{solar energy** + CO_2 + H_2O > glucose +O_2**}**

Example: Cellular respiration is the **opposite** of photosynthesis, heat is liberated (**exergonic**) and is **catabolic** (decomposing).

Equation of respiration: {glucose + O_2 > CO_2 + H_2O + **Heat**}

What is an **enzyme**? A protein. What is the active site (a cleft or pocket in the enzyme where the reaction takes place similar to a lock. The substrate (or molecule which is catalyzed by the enzyme or reactant) fits into the groove of the active site (the substrate is analogous to the key where the catalysis occurs giving rise to the products.

Enzymes are highly folded proteins that **lower** the activation energy of a reaction and are **not** consumed in the reaction. Understand the concepts of the heat of reactants and heat of products as they are displayed on a graph (the 'hill' between heat of reactants and heat of products is often the activation energy. The enzyme **reduces the activation energy**, making the 'hill smaller.'

Enzymes increase the rate of the reaction

What happens when you increase the enzyme concentration? ***Faster***

What happens when you increase the substrate concentration?' ***Faster***

Enzymes are optimal at certain temperatures and pH conditions. The enzymes of warm blooded mammals tend to have an optimal temperature at 37C or 98.6F degrees. What about the optimal

temperature for enzymes of bacteria that live close to volcanoes called thermophilic bacteria? *Answer: Much warmer, even 70C.* Interestingly, during digestion, certain enzymes in your stomach are optimal at acid pH whereas other enzymes are optimal at basic pH, thus depends on *local environment*?

What are the differences between competitive and noncompetitive inhibition of enzymes? How do they work?

If a drug is a competitive inhibitor, then increasing the substrate concentration can overcome drug action. However, a noncompetitive drug has an allosteric effect and thus increasing the substrate concentration cannot overcome the drug concentrations, since the drug is interacting allosterically or away from the active site binding area.

Plasma membrane is a phospholipid bilayer. Hydrophilic phosphate heads face the outside of the cell and face the interior of the cell; whereas, the nonpolar hydrophobic fatty acid chains are contained in the inside of the bilayer. Cholesterol molecules reside in the hydrophobic interior.

The bilayer has transmembrane (traverse the bilayer) **INTEGRAL proteins** that are embedded in the bilayer**.** The text lists 6 types. Define each

1.CHANNEL- a tunnel that permits the flow into or out of the cell

2.CARRIER- facilitate movement of molecules into or out of the cell

3.CELL RECOGNITION- Immune system recognition

4.RECEPTOR-- Binds to ligands on the cell surface and leads to downstream regulatory processes

5.ENZYMATIC- activates or inhibitory enzymes localized to membrane

6. JUNCTION - Binds neighboring cells

PERIPHERAL PROTEINS, on the other hand, are ONLY on the inside of the cell (on the cytoplasm side, not the extracellular side) and do not penetrate the lipid bilayer.

Often**, receptor integral proteins embedded within the cell surface** bind to extracellular **ligands or signaling molecules**, such as, hormones, growth factors, neurotransmitters, ions, etc. and as these ligands dock to the receptor on the outside of the cell, the receptor protein changes its conformation thereby allowing peripheral proteins on the I**NSIDE** of the cell to **transduce** additional signals inside of the cell. **Signal transduction** inside of the cell can be mediated by additional enzymatic modifications (a domino effect like a cascading waterfall) leading to a **cellular response**. Sometimes, signal transduction cascades can reach the nucleus where genes are activated or silenced.

Remember, the cell membrane is selectively permeable. In other words, most molecules cannot pass freely from one side of the membrane to the other. Polar molecules have difficulty going the membrane because of ionic bonding and hydrogen bonding. Nonpolar molecules can cross the membrane, more easily, including lipids and small uncharged molecules. The movement is highly regulated.

DIFFUSION or PASSIVE TRANSPORT (*high concentration to lower concentration* along a concentration gradient). Examples are oxygen and carbon dioxide gas. Many lipids which are nonpolar can go across the membrane via passive transport.

FACILITATED diffusion: **A carrier or transport protein** that facilitates movement of one compound that is 'dragged along with the help of another compound.'

BULK TRANSPORT: What is **exocytosis** (particles going out); **Endocytosis** (particles going in); **Pinocytosis** (cell imbibes or drinks); **Phagocytosis** (cell engulfs particles, e.g. macrophage of immune system 'swallows' a pathogen or bacteria). Often exocytosis and endocytosis are mediated by **Vesicles**.

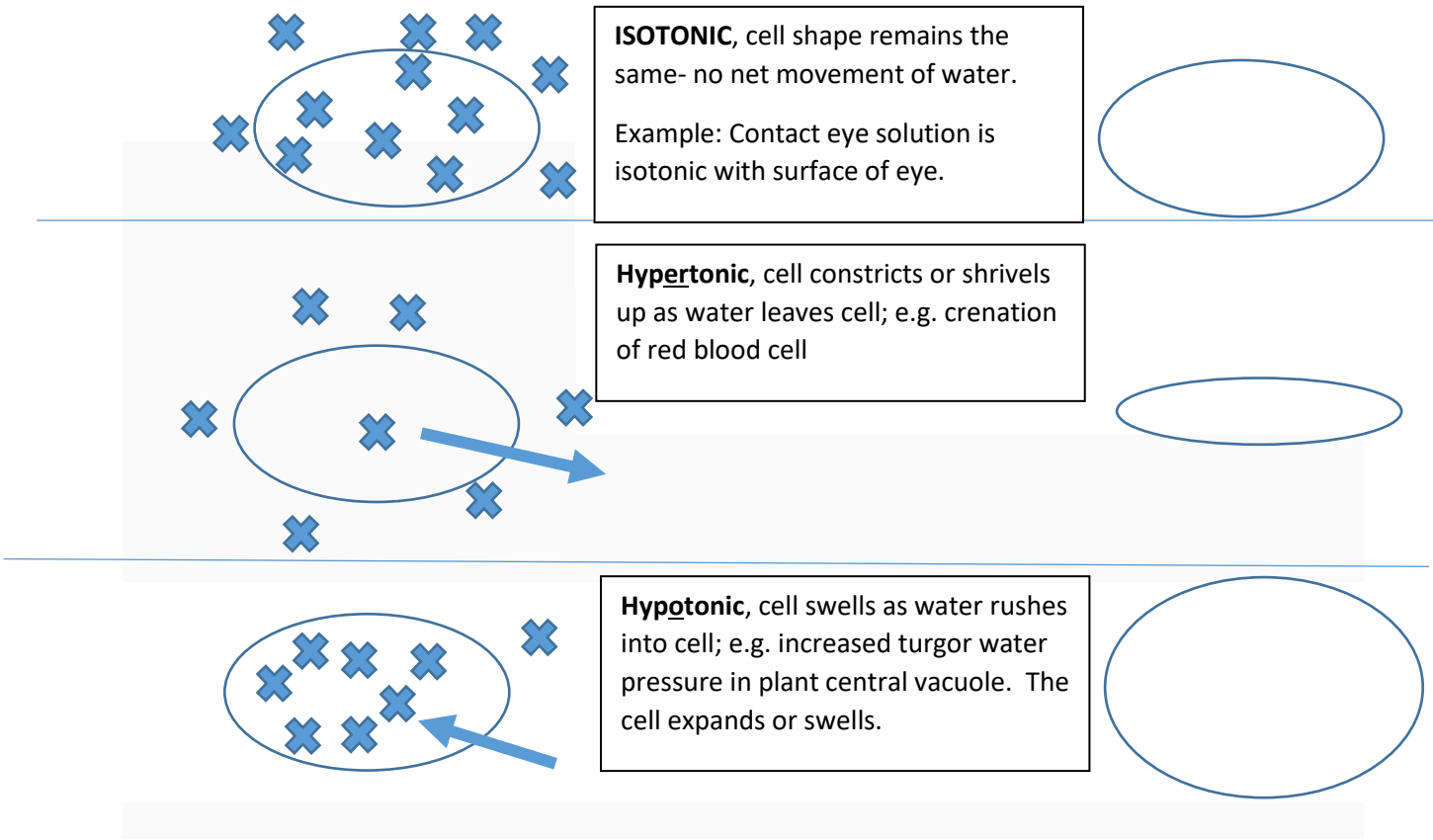
Know the concept of **ACTIVE TRANSPORT** or pumps usually requires energy or ATP and typically occurs as particles AGAINST the concentration gradient or going from ***low to higher concentration***. A classic example is the sodium/potassium ion pump (Na/K)

Note that molecules can bind to receptors and be brought into the cell by a process called **RECEPTOR-Mediated ENDOCYTOSIS.** Often, lysosomes (or endosomes) fuse to vesicles and unload their cargo (molecules from the outside of the cell and then bring the receptors back to surface, in effect, recycling the receptors.

What are SOLUTES or SOLVENTS. Define?

Understand the concepts of **TONICITY** as it applies to **OSMOSIS** or movement of water.

Define the terms ISOTONIC, HYPOTONIC, and HYPERTONIC Solution. In the drawings depicted below, the oval represents a cell, the 'X' is a solute particle, and the arrow points in the direction of water movement or osmotic flow. The cell shape on the left is the result!



CELL CYCLE

Dividing cells must enter the cell cycle; whereas, nondividing cells such as most neural cells and some muscle cells that do **not** divide in humans remain in **G_0** (Gzero or Gnaught). Cells divide during the growth and development of the organism as well as during repair/regeneration. For the most part, cells do not grow *ad infinitum* unless they are cancer cells in which they are 'immortalized.' In other words, cells undergo 'programmed cell death' or **apoptosis** after several rounds of cell divisions or cell doublings. This 'cellular suicide 'or apoptosis mechanism is a normal process for most organisms, since 'worn out parts' of the cell, and the accumulation of unwanted mutations may accumulate over time, hence the cell must eventually 'die.'

The stages of the cell cycle proceed in a highly-regulated sequence that is marked by two major divisions- **Interphase** and **Mitosis**. Interphase consists of the G_1 phase followed by S phase followed by G_2 phase (G_1>S>G_2). During **S** phase, the **chromosomes replicate (or copy, duplicate)** prior to **Mitosis or cell division in the M phase**. The entire cycle proceeds in this sequence (G_1>S>G_2>M), but before a cell can enter the next stage, a checkpoint (e.g., stop sign) or cellular quality check (similar to QC) is required. NOTE- after 1 turn of the cell cycle, 2 copies of the original cell (called the mother cell) result in 2 identical daughter cells. For example, a 46-human chromosome liver mother cell generates 2 **identical** 46 chromosome liver cells after one turn of the cell cycle. Understand what happens at each stage.

*****Dear class, you must know what happens in each **stage** of the cell cycle G_1>S>G_2>M. Furthermore, the M phase or Mitosis phase is divided into several sub stages, in this order, **prophase, prometaphase, metaphase, anaphase,** and **telophase**

In which phase do the chromosome condense and become visible? Prophase

Which phase do the chromosomes align in the center or equator of the cell? Metaphase

Which phase is marked by the initial 'pulling' of the chromosomes to the opposite pole? Anaphase

Which phase is prior to cytokinesis (animal cells) in which the individual cells are separated? Telophase

The spindle apparatus that moves the chromosomes during mitosis are components of the cytoskeleton. The chromosomes are attached to **microtubules.** The **sister chromatids** are the duplicated chromatids resulting from the S phase of interphase held together a centromere.

If a picture taken from a tissue section of cells undergoing mitosis (microscope of stained chromosomes), then could you identify and differentiate between the stages? **prophase, prometaphase, metaphase, anaphase,** and **telophase.**

Understand the **differences** between cellular division in plant and animal cells?

Animal cells undergo cytokinesis with a cleavage furrow which pinches off the daughter cells by a contractile ring. Plant cells, on the other hand, use **temporary cell plate** formed by fused vesicles. Plants do not have centrioles, either.

Prokaryotes duplicate or replicate their circular DNA and undergo **binary fission**.

If you have 66 chromosomes in a cell undergoing mitosis, then after mitosis the two daughter cells will have 66 chromosomes each as well because they are copies of the original mother cell.

CANCER:

What are the differences between a normal cell and a cancer cell with respect to cell division?

Cancer has uncontrolled growth

What is a **proto-oncogene**? A normal gene responsible for cell growth

Mutations in proto-oncogenes become oncogenes resulting in uncontrolled growth-no checkpoint control in the cell cycle

What is a **tumor suppressor gene**? Often these genes are important in DNA repair.

Mutations in tumor suppressor lead to a malfunction of the cell's DNA repair machinery leading to an accumulation of mutations which in turn can be "oncogenic"

Understand the concept of Cyclins and DNA repair checkpoint proteins.

Cyclins are responsible for checkpoints during transitions between phases of the cell cycle-critical as QC or Quality Control measure.

Malignant tumors metastasize. In these cases, cancer cells travel to distant site or secondary site via circulatory or lymph systems usually.

SEXUAL REPRODUCTION uses MEIOSIS.

*****NOTICE MEIOSIS and MITOSIS are spelled differently and are different**.**

To understand sexual reproduction, we must understand the difference between **diploid** (2n) and **haploid.** For instance, all the **somatic** (body) cells of our bodies have 46 chromosomes **or 2n**; however, the gametes of the sexual organs have **HALF** as many or 23 chromosomes. Eggs have 23, and sperm have 23 chromosomes. These gametes are (haploid or **1n)**. Upon fertilization of the egg with the sperm (23 + 23 chromosomes = 46 chromosomes.) A fertilized egg is called a **zygote** containing 46 chromosomes. During early embryonic development, the zygote (2 cell stage) grows and **differentiates** into the specialized tissues such as the muscle, nervous system, epithelia, and connective tissues.

How are the gametes formed in the sex organs?

The process is called **Meiosis** occurs in 2 stages (MEIOSIS I and MEIOSIS II).

Meiosis provides **genetic DIVERSITY. On the other hand,** Mitosis provides **EXACT** copies of cells.

Where does genetic diversity arise in Meiosis?

In Prophase I of Meiosis, Homologous Chromosomes pair together and genetic information can be exchanged by **CROSSING OVER** of **non-sister chromatids. This generates variation.**

Furthermore, more variation occurs as **the homologous chromosome pairs** align at the metaphase plate in **Metaphase I**, as the chromosomes are poised to separate in a **random** manner (law of independent assortment of homologous chromosomes). In humans, $(2^{23})^2$ or roughly 70 trillion combinations of zygotes are possible (huge variation).

The Homologous Chromosomes separate in Anaphase I of Meiosis I

***Be aware that **4 gametes (or 4 different sperm)** are formed during one round of Meiosis. Each of those gametes has 23 chromosomes. These gametes are **NOT** identical to the mother cell at the start of Meiosis.

Sister chromatids separate in Anaphase II of Meiosis 2.

What is **NONDISJUNCTION?** Chromosomes do **NOT** separate during Meiosis I or Meiosis II. Compare **Turner female** (XO) with **Klinefelter's male** (XXY). How do they arise?

Nondisjunction is aneuploid with irregular chromosome number, some cancer cells are aneuploid.

Another classic example of aneuploid is Down's Syndrome of **Trisomy 21** (an extra chromosome 21 in the karyotype of chromosome spread.

More CHROMOSOMAL rearrangements:

Deletions or **Duplications**.

Translocation- a segment (or fragment) of one chromosome moves to a non-homologous chromosome reassembles (or fuses).

Inversion: a segment breaks off and reseals in the opposite orientations. Gene order may be affected or disrupted.

Mutations in somatic cancer cells can experience chromosomal rearrangements. Environmental factors such as chemicals, diet, and radiation can induce such rearrangements.

********VIRUS*******

Two main classes, viruses use the cellular machinery of the host cell copy their nucleic acids.

Some **DNA VIRUSES** include small pox, chicken pox, Human Papilloma virus (HPV), and Herpes. There are both single and double stranded DNA viruses. An Adenovirus is a DNA virus.

II. The **RNA VIRUSES** may also be single stranded or double stranded. An example of a double stranded is the rotovirus.

Other single stranded RNA viruses include the **Retroviruses** which includes the common cold (rhinovirus), influenza, polio, measles, Zika, West Nile, and Hepatitis C.

A subclass of retroviruses is the lentivirus such as the human Immune Deficiency virus (HIV).

Retroviruses can infect non-dividing cells.

RNA viruses also contain the genes that encode for the **Reverse Transcriptase to convert their RNA genome to DNA.** ***Note this is the reverse order of the Central Dogma. RNA> DNA. The viral RNA is converted to DNA.***

Mendel's Law of segregation- **Allele** is a variant of a trait. One allele comes from each parent, and they segregate independently.

Understand concept of **Dominant** (Capital letter) and **Recessive** (lower case letter) alleles. For example, seed color has two alleles- Capital '**G'** and lower case '**g**.' A '**GG**' seed with be green. This is **homozygous** (the same for both alleles) **dominant**. The seed will look green. A "**Gg**" seed is **heterozygous dominant** (different alleles) and will also appear green. A **homozygous recessive** allele of **gg**' will appear yellow. A **test cross** will determine whether the original green plant is GG or Gg.

Know the difference between **phenotype** and **genotype.** The phenotype of GG and Gg is the same, both look green. The phenotype is the outward appearance of the alleles. The genotype of GG and Gg is **not** the same, however. GG is homozygous dominant, and Gg is heterozygous dominant.

You will have to execute some **Punnett Square** scenarios for the exam. The simplest Punnett square is shown in the PowerPoint slides for chapter 11 in slides 11-19. This is mono hybrid cross. Additional practice Punnett squares are available online. To perform Punnett Square problems, you will need to create breeding experiments between **Parental** strains, and resulting **F1 generation** (offspring, or 'children'), and subsequently, the **F2 generation** analogous to 'grandkids; etc.

The testcross can reveal whether a dominant phenotype such as tall plant height is either a TT(homozygous) or a Tt (heterozygous) genotype.

The concept of **Incomplete dominance**. Red rose (RR) crossed with white rose (rr) yields pink roses (RR= red, Rr = pink, rr= white, R is dominant allele, r is recessive allele, but Heterozygote Rr has an **Intermediate phenotype.**

Blood type is another exception. Type O is **recessive**. If one parent has A or B and the other parent has AB, then O blood phenotype type will **not** show in any children. If one parent is Type A, and the other is type B then a O blood type phenotype could show in the children, since the parents could both be carriers AO or BO giving OO kids or O blood phenotype. The Blood type AB is **codominant.**

Most alleles are located on **autosomal chromosomes** and are not carried on the sex chromosome. Very few genes are located on the Y chromosomes; however, some alleles are carried on the X-chromosome and are thus **sex-linked**. For the most part, genes that ***are sex-linked reside only on the X chromosome,*** not on the Y chromosome. Examples of sex-linked inheritance are red/green color blindness and hemophilia.

****REMEMBER some diseases are **DOMINANT** and others are **recessive**. Obviously, dominant diseases usually affect more family members in a **Pedigree**. If a disease is dominant, it may not be common as a congenital disease (lethal or very severe), but could be more common as an adult onset disease

Physical characteristics of DNA include a double helix that resembles a ladder with the sides of the ladder being the **phosphate/sugar backbone**, and the rungs of the ladder being the 4 **nucleotides (A, G, C, T**) which are paired together by hydrogen bonding. The nucleotides exhibit **complementary** base pairing or A always pairs with T; and G always pairs with C (i.e., A never pairs with G or C).

Therefore, as an example, if DNA analysis is performed on a sample, and it is revealed that 30% of the sample's chemical composition is Guanine (or 'G') then 30% must also be Cytosine (or 'C') because G always pairs with C. Since G and C are 60% of the sample DNA, then the remaining 40% of the DNA must be A and T (20%A, 20%T).

The two strands of DNA are **antiparallel** and have a polarity in which the 'head' is a negatively charged phosphate group at the **5'** or fifth carbon group of the pentose sugar group; whereas, the 'tail' of DNA is **3'**or the third carbon group of the pentose sugar group of one strand of the DNA helix.

During the **S phase** of the cell cycle, the DNA is replicated (duplicated, copied) in a process called **DNA replication**. The process is **semi-conservative** or one of the older strands is retained during DNA replication, and the new strand must be synthesized only in a 5' to 3' direction by DNA polymerase for both the leading and lagging strand.

CENTRAL DOGMA is critical for this exam. ***DNA>RNA>PROTEIN***

The tenant of the central dogma states that the **DNA** blueprint transcribes a **RNA** molecule in the nucleus (**TRANSCRIPTION**), and then the single stranded RNA molecule leaves the nucleus and is translated at the **ribosome** into a **protein** (**TRANSLATION**).

Be prepared to generate a complementary strand of RNA from DNA. Hint, RNA uses Uracil or U instead of Thymine in DNA but the other three nucleotides (A, G, and C are the same between DNA and RNA).

Example: **5'-GCCACTTA-3'** How would you generate an **mRNA** molecule for this? Answer is ---------- 3'-CGGUGAAU-5'. Notice RNA uses U instead of T. Also notice the 5' and 3' which shows that strands are **antiparalle**l. The top strand has 5' to start and the bottom has 3' to start below.

Understand the concept of immature mRNA in which introns are spliced out of the mRNA leaving only the exons and resulting in a mature mRNA.

Define these terms-**mRNA, rRNA, tRNA, codon, anticodon, triplet code, and code redundancy.** mRNA is messenger RNA that is complementary to the template strand of DNA (example show above), rRNA are the subunit components that comprise the ribosome structure, tRNA is the anticodon that pairs with the codon (or triplet) of the mRNA and brings an amino acid. Codon is a triplet of mRNA that is destined to be translated to one amino acid. An anticodon is a triplet of nucleotides on a tRNA hairpin molecule that is complimentary to the codon sequence of the mRNA chain

The **CENTRAL DOGMA** of Genetics starts with **DNA** in the nucleus (eukaryotic cell) which is transcribed to **mRNA** and further processed in the nucleus by post-transcription regulation (immature pre-mRNA is converted to mature mRNA). This mRNA leaves the nucleus for the cytoplasm and attaches to the ribosome where translation occurs resulting in a growing polypeptide chain (like beads of necklace) forming a protein. Later, this protein is folded and further modified by post-translational regulation (i.e., phosphates are added, covalent attachment of sugar or lipid groups, etc. in the Endoplasmic reticulum and Golgi Apparatus).
To reiterate, the sequence of events in Central Dogma as follow*: first transcription > post-transcription> translation > post-translation.* Transcriptional control is the most important step in this process because it is the first step and determines whether the gene will be transcribed in the first place.

Note: in prokaryotes or bacteria, both transcription and translation can occur simultaneously because bacterial DNA is not confined in a nuclear membrane.

Let's dissect these mechanism one-by-one:

I **Transcription** starts as the double helix but at the start of transcription it unwinds, and one of the strands acts as a template strand for transcription. **RNA polymerase** is the enzyme that pairs the complementary nucleotides (A, G, C, and U) into the RNA strand in the 5' to 3 direction during transcription. As far as base pairing, C pairs with G; A pairs with U (not T which is for DNA/DNA double strands). During transcription, a DNA/RNA hybrid double helix is temporarily formed. The DNA is the template strand, and the RNA is the mRNA strand. Eventually, the RNA is released from the DNA/RNA hybrid helix as a single strand called the immature or pre-mRNA.

II.**Post transcriptional** processing trims and splices out certain **introns** in the pre-mRNA or immature mRNA. The introns are the intervening sequences between **exons**. Most genes have more than one exon. The exons contain the triplet nucleotides or codons which eventually encode for the mature mRNA sequence that will eventually be translated into the protein. *Introns are not translated and thus are often called noncoding. The exons are coding*. The splicing or removing of introns is a regulatory mechanism in eukaryotic cells because sometimes exons are 'skipped' or **alternatively spliced** resulting in functionally smaller proteins or different isoforms of the protein. The final mature mRNA also undergoes additional modification such as a 5' guanine cap at the head of the RNA and a polyadenylation tail (many Adenosines added) on the 3' for stability.

III. **Translational control** depends on the lifespan of mRNA molecules. RNA is degraded by specific RNAse enzymes and other protein complexes, so their population can be tightly controlled. In other words, RNA stability is important outside of the nucleus. Of course, if less RNA is available outside the nucleus, than less mRNA molecules can be translated into proteins. Furthermore, there is some control over the initiation, elongation, and termination of the polypeptide chain on the ribosome itself.

IV**. Post translational control** are the final modifications to proteins that determine their folding. As proteins are folded, additional functional groups can be added to the protein such as phosphates, carbohydrate chains, and lipid modifications.

Transcription initiates at the **promoter region** which is just upstream of the transcription start site. The minimal promoter region contains the essential DNA binding elements which attract a protein complex of the basic transcriptional machinery that 'pile ups' close to the start transcription start site. In Eukaryotes, RNA polymerase II is enzyme that polymerizes the mRNA strand. However, to initiate and start transcription itself, gene specific **TRANSCRIPTION factor proteins** assemble at the promoter to activate transcription. Often, the DNA bends as **enhancers and silencing DNA elements** located outside of the promoter region to tissue specific transcription factors.

For example, a brain cell and a liver cell both contain the same 46 chromosomes in humans. The differential expression of genes in the liver and the brain are due to the **different Tissue specific transcription factor proteins** which activate or silence gene in these tissues. For example, nerve specific transcription factors are not present in liver cells. On the other hand, brain specific transcription factors are not present in liver cells. Note the DNA promoter elements, enhancers, and silencers are part of the DNA which is the same in each cell. <u>The transcription factors are responsible for specificity.</u>

Translation occurs at the ribosome. The mature mRNA that leaves the nucleus and docks to the ribosome is comprised of a code in which every three nucleotides is called a **triplet or codon**. These codons can bind to **anticodons** which are three nucleotides that are complementary to the codon sequence**.** Each anticodon of three nucleotides is part of a **tRNA** molecule which is tethered to 1 amino acid. Note, there are 64 anticodon tRNA molecules but only 20 different amino acids, hence some individual amino acids are linked to more than one anticodon for a tRNA molecule.

During translation, the start is called **Initiation** as mRNA strand binds to the ribosome usually at the initiation codon or AUG.

Key words for the **3** parts of a prokaryotic (bacterial) **OPERON**

Part 1) **Promoter**- A DNA segment (or region) near the start of a gene in which RNA polymerase attaches.

RNA Polymerase: The enzyme responsible for transcription (**mRNA** in eukaryotes).

Part 2) **Operator:** A DNA segment where an <u>active</u> repressor binds.

Part 3) **Regulatory gene**: This gene that encodes for the repressor protein which in turn controls the operon. The regulatory gene is normally located outside of the operon. The repressor protein controls whether the operon is <u>active or non-active (silenced).</u>

Three types of operon are 1) trp operon, 2) lac operon, and 3) CAP. Know the differences amongst the three types.

Trp OPERON- The transcription of the gene which eventually is responsible for producing the amino acid trp (or tryptophan) is normally **always ON** (i.e. the light switch is normally on). In other words, the default is 'on' or constitutively active. By default, the repressor normally **does not bind** to the operator. As the levels of the amino acid tryptophan in the cell reach acceptably high levels, then the tryptophan protein binds to the repressor which results in an 'active repressor' thus binding to the operator and shutting off tryptophan gene expression. In this case, tryptophan acts as a **corepressor**.

Lac OPERON- The transcription of the gene which eventually is responsible for the enzymes that metabolize the sugar lac (or lactose) are normally **always OFF** (i.e. the light switch is normally off). In other words, the default is 'off' or constitutively inactive. By default, the repressor normally **binds** to the operator and inhibits the transcription of lactose-metabolizing genes. As the levels of sugar lactose in the cell reach acceptably high levels (lactose is a nutrient for the cell), then the **lactose binds to repressor** which frees from repressor from the operator, and then allows transcription of the genes that metabolize the sugar lactose. In this case, lactose acts as a **de-repressor.**

CAP- cap is normally inactive. E Coli bacteria prefer breaking down glucose, rather than lactose. If lactose is present in the cell and glucose is absent, then cyclic (cAMP) is high and then **binds** to the CAP (**C**atabolite **A**ctivator **P**rotein). The cAMP/CAP dimer complex binds to the LAC operator more efficiently thus activating the structural genes of the lac operon.

*** The trp and lac operons are negative feedback loops; whereas, the CAP operon is a positive feedback loop.

Eukaryotic Gene Expression:

DNA is 'coated' with proteins, and DNA is not naked in the cell. The DNA/histone complexed together is called **chromatin.** Histone proteins are positively charged as they cling to the negatively charged DNA molecules. The DNA is wound around the protein histones like beads on a string (the string is the DNA double helix). The beads of histone are called **nucleosomes.**

Loosely packed DNA is transcriptionally **active or Euchromatin**. Tightly packed DNA is transcriptionally **inactive or Heterochromatin**. The loosely packed DNA allows for the transcriptional machinery such as RNA Polymerase II, Transcription factors, and other proteins to sit on the DNA and gain access thus initiating gene transcription.

An example of how proteins can mask DNA and prevent genes from being expressed is random X-cell inactivation in mammals. A female Calico cat has three colored fur because one of its X chromosomes is 'inactivated' randomly.

EPIGENETICS -is inheritance patterns that are **not dependent on genes themselves**. The calico cat example above is epigenetic. Another example of epistasis provided by our fabulous textbook is the coat color of black Labrador retrievers, Chocolate Labs, and Yellow Labs. In this breed, black and brown are the two pigments that can be deposited into the fur- but in the yellow lab, both pigments are produced but neither is deposited in the fur. In other words, the 'yellow allele is 'epistatic' to brown and black. The Identical twins don't look the same, because the random assembly of proteins on the genes (the chromatin) during fetal development.

Another example of epigenetics is DNA methylation which **inactivates** genes.

How are truncated or smaller proteins with different subunits generated in eukaryotic cells? The **alternate processing of the mRNA**. Remember, introns are spliced out and exons are retained from the pre-mRNA (immature mRNA) to generate the mature mRNA. Sometimes exons can be skipped leading to different protein isoforms and shorter proteins.

The levels of regulation for gene activation and protein synthesis in plant and animal cells (e.g., eukaryotes) is more complex than bacteria (e.g., prokaryotes). Since eukaryotes have a defined nucleus, RNA processing mechanisms affect the final mRNA transcripts product. These mechanisms are classified as **POST-transcriptional** modifications. Furthermore, mRNA molecules are stabilized with a 5' guanine cap and a 3' polyadenylation tail which prevents mRNA degradation. The level of enzymes that cleave RNA molecules called **RNAses** are also tightly controlled.

Likewise, proteins are processed shortly after translation to adopt a tertiary folded structure. Additional modifications such as the addition of phosphates, sulfates, carbohydrate and/or lipid sidechains can continue in the Endoplasmic reticulum (ER) and the Golgi apparatus by a **POST-translational mechanism.** Misfolded proteins are also degraded by the **cellular proteasome complex.** The cell must have a QC or quality checkpoint to detect abnormalities.

MUTATIONS:

A **Somatic** mutation occur during the life of the organism in a **body** cell EXCEPT the gamete producing cells (e.g., testes and ovary). On the other, **Germline** mutations occur only in the **gamete** producing cells (e.g., testes and ovary) and are inherited to the offspring and subsequent generations.

Mutations can either by **Spontaneous or Induced**. Spontaneous mutations occur at a low frequency from normal cell activities such as DNA replication or DNA repair (usually random).

However, Induced mutations arise from the environment such as radiation or chemical. A classic example, cigarette smoke induces mutations in the lung alveolar cells.

Type of mutations- Point mutation of a nucleotide. Sickle-Cell anemia is a point mutation that changes one nucleotide base resulting in a change from a GLUTAMIC ACID amino acid changed to a VALINE amino acid. This occurs in one polypeptide change of the hemoglobin protein causing red blood cells to adopt a sickle shape.

Mutations can be **SILENT** or no change to the amino acids. Example codon redundancy of 61 possible combinations for 20 amino acids. Mutation can be **MISSENSE** as with sickle cell disease, changing one amino acid to a different one resulting in a change in function. Mutation can be **NONSENSE** resulting in a premature stop codon leading to a truncated or misfolded protein.

Micro insertions or deletions of nucleotide(s) can cause **FRAMESHIFT** mutations leading to premature stop codons and the encoding of different amino acids-often such proteins are misfolded or nonfunctional which could have disastrous consequences for the organism.

Understand the difference between **proto-oncogenes** and **tumor suppressor genes**. Note, both are **NORMAL** genes **not Cancer genes**. However, if mutations arise in either one, then cancer may occur. Proto-oncogenes control cellular division and tumor suppressor genes are DNA repair genes or genes that control the checkpoints of the cell cycle (cyclins). A mutation in a proto-oncogene results in *uncontrolled* growth leading to cancer. A mutated proto-oncogene is called an **oncogene.** Mutations in tumor suppressor gene impair the cell's normal DNA repair activity leading to the *accumulation of mutations* in cells which is a prelude to cancer.

NATURAL SELECTION

Darwin observed that individuals in a population vary in their traits, many of which are **heritable**. These variations occurred by chance and were random. More offspring are produced than survive, and competition is inevitable. Individuals that are best suited to their environment are more likely to survive and reproduce**.** *Over long periods of time*, more individuals in a population will have the advantageous traits. Evolution occurs as the unequal reproductive success of individuals (**reproductive fitness**)

Darwin proposed that natural selection could cause an **ancestral species** to give rise to two or more descendent species. He used **fossil evidence** to justify ancestral species. He also noticed similarities by looking at **comparative anatomy**. A bird arm and a human arm do **not** share a common ancestor, but the wing of a bat, a whale flipper, a dog arm, and human arm are similar (mammals), and thus share a common ancestor. He observed the beak length of finches (a species of birds) and tortoises on the Galapagos islands. He also noticed differences with **biogeography** (populations might be isolated depending on region, but the isolation may have occurred leading to the divergence from a common ancestor. Darwin did **not** have any **biochemical** evidence that DNA existed, but he proposed that ***inheritable variation*** occurred by chance usually over a long period of time.

Darwin's Theory of Evolution is in sharp contrast to Lamarck's Inheritance of Acquired Traits which proposed that modifications happen during an organism's lifetime and are passed onto the next generation. For example, according to Lamarck, a female giraffe stretches to get food in higher trees and thus the giraffe's progeny will also have taller necks. On the other hand, Darwin proposed that inheritable variation occurred randomly and took many generations for an acquired trait to provide reproductive fitness.

Photosynthesis:

Plants are producers or **Autotrophs**; whereas, animals are consumers or **Heterotrophs.**

Two Stages: 1) Light reactions and 2) Calvin Cycle.

KNOW this equation summarizing photosynthesis:

Solar energy + $6CO_2$ + $6H_2O$ --→ $C_6H_{12}O_6$ (glucose) + 6 O_2

Light reaction in presence of sunlight and absorbs solar energy by pigments such as Chlorophyll by energizing electrons as these electrons move down Electron transport chains by pumping Hydrogen protons or H^+ into thylakoids and generating ATP from ADP and NADPH out of $NADP^+$

In the LIGHT reactions of photosynthesis. H_2O is oxidized and O_2 is generated as the product of the light reactions and released as a gas.

Later in the Calvin Cycle, CO_2 is reduced in the Calvin Cycle to produce 3 carbon compounds. In effect, Glucose **(Carbs) are the product of Carbon dioxide fixation of the Calvin Cycle.**

Note: ATP and NADPH produced in the light reactions are necessary for the Calvin Cycle.

Glyceraldehyde-3-phosphate is the building blocks for glucose synthesis as well as protein or lipids in photosynthesis.

CELLULAR RESPIRATION

1. What are the 3 stages of the cellular respiration process?
   Glycolysis, Krebs Cycle, Electron Transport, in this order
2. Where in the cell does the glycolysis part of cellular respiration occur?
   in the cytoplasm
3. Where in the cell does the Krebs (Citric Acid) cycle part of cellular respiration occur?
   in the mitochondria
4. Where in the cell does the electron transport part of cellular respiration occur?
   in the mitochondria
5. How many **ATP** are made in the electron transport part of cellular respiration?
   30 THE **MOST**
6. In which phase of cellular respiration is carbon dioxide made?
   Krebs Cycle
7. In which phase of cellular respiration is water made?
   Electron Transport

8. In which phase of cellular respiration is oxygen a substrate that accepts electrons?
   Electron Transport

9. In which phase of cellular respiration is glucose a substrate?
   Glycolysis

++++GLYCOLYSIS DOES NOT REQUIRE OXYGEN but is still part of cellular respiration and happens outside of the mitochondria in the cytoplasm

ANAEROBIC fermentation requires NO oxygen.

1)Alcoholic Fermentation = Happens in Fungus (Yeast) and some bacteria, and it produces Carbon Dioxide and ATP

2)Lactic **Acid** = Happens in animal cells, what cause muscles to be sore, and produces Lactic Acid and ATP

Both photosynthesis and cellular respiration require Hydrogen Proton pumps for ATP synthesis