**Mendelian genetics** - particulate (instead of blending) theory of inheritance

* inherited characters determined by genes
* genes occur in pairs (from maternal/paternal homologous chromosomes)
* **Law of Segregation**- only 1 chromosome from each pair found in gametes
* **Law of Independent Assortment** - genes on different chromosomes distributed randomly into gametes
* **gene** - unit of heredity on chromosomes
* allele - alternate states of genes, contributed from parents
* **dominant alleles** - masks expression of other alleles (designated by capital letter)
* **recessive alleles** - expression masked by dominant alleles (designated by lower-case letter)
* **genotype** - all alleles present in the cell
* **phenotype** - physical appearance of a trait
* homozygous - when paired alleles are identical
* heterozygous - when paired alleles are different

**types of inheritance**

* **simple dominance** - certain allele completely dominant over another
* **incomplete dominance** - heterozygous genotype results in mixed characteristic
  + red/white flowers create pink flowers when cross-bred
* **lethal inheritance** - inherits gene that kills offspring
  + albino seedlings cannot photosynthesize, eventually die
* **codominance** - evident in blood types
  + 4 blood types - A, B, AB, O; determined by **antigens** (proteins) on surface of cells
  + A blood - has A antigens, antibodies against B blood cells
  + B blood - has B antigens, antibodies against A blood cells
  + AB blood - has A/B antigens, no antibodies
  + O blood - has antibodies against A/B blood, no antigens

**human traits** - many determined by just single gene

* widow's peak - pointed hairline dominant over straight hairline
* bent little finger - little finger bends toward 4th finger (dominant)
* albinism - lack pigment in skin (recessive)
* pigmented iris - pigments (dominant) hides blue/gray color of iris back layer
* attached earlobes - free earlobes dominant over attached earlobes
* hitchhiker's thumb - last joint of thumb bends back over 60 degrees (recessive)
* interlacing fingers - crossing left thumb over right dominant over crossing right over left
* PTC tasting - ability to taste bitterness dominant over inability
* mid-digital hair - hair on middle segment of fingers (dominant)

**human diseases** -

* cystic fibrosis - chronic bronchial obstruction, growth reduction
* galactosemia - can't metabolize galactose in human milk (autosomal recessive)
* phenylketonuria (PKU) - inability to metabolize phenylalanine, leads to mental retardation
* Huntington's disease - uncontrollable, involuntary muscle movements
* occurs late in life, often gets passed on to offspring
* juvenile retinoblastoma - cancer of retina

**transposons** - fragments of DNA that can move in chromosome

* useful in genetic engineering
* can insert foreign DNA into chromosome

AP Biology

DNA History & Structure

(Associated Learning Objectives: 1.14, 1.15, 2.22, 3.1, 3.2, 3.29, 4.1, 4.23)

Important concepts from previous units:

1. The basic unit of DNA or RNA is a nucleotide; composed of a Nitrogen base, 5 Carbon sugar, and phosphate.
2. DNA is like a “million dollar blueprint” having the genetic information for *making* proteins and enzymes.
3. Base pairing is always a pyrimidine (C, T, U) with a purine (A, G).
4. Frederick Griffith (in 1928)
   1. He was a British Army doctor who was studying Pneumonia in the hopes of finding a cure.
   2. He is given credit for the **transformation** experiment, even though this was not his original intent.
      1. In the experiment, he took pathogenic (disease causing) bacteria and non-pathogenic bacteria and injected them into mice. The pathogenic bacteria killed the mice. The non-pathogenic did not kill the mice. He then took some pathogenic bacteria and killed them by exposing them to *heat*. He took the dead bacteria and injected them into more mice. The mice did not die. He then took some of the dead pathogenic bacteria and mixed them with the non-pathogenic bacteria. He then injected the mixture into some more mice. THEY DIED. His reasoning was some “instructional agent” was *exchanged* between the dead pathogenic bacteria and the living non-pathogenic bacteria allowing them to “learn” a new trick. How to make the toxin (poison). So we say they were **transformed** from non-pathogenic into pathogenic bacteria.
5. Oswald Avery and associates (in 1944)
   1. He retests Griffith’s experiment, but with the purpose to find out what the “instructional agent” was that led to the transformation of the non-pathogenic bacteria.
   2. After the testing, he states that the transformation agent was DNA.
   3. This statement sparks lots of controversy as DNA is too simple a molecule most scientists believe. It must be proteins, as they are very large complex molecules. So now the race is on to prove which was it, DNA or proteins.
6. Alfred Hershey and Martha Chase (in 1952)
   1. They worked with the T2 Bacteriophage (a virus that infects bacteria) and E. Coli bacteria.
   2. This becomes the **Hershey-Chase Experiment**.
      1. They used *radioactive Sulfur 35* to label the virus’s *protein outer capsid* in one container. (Remember, the amino acid Cysteine contains sulfur. The radioactivity allows them to follow where the *proteins* go by using a Geiger counter. A Geiger counter is used to measure radioactivity.)
      2. They then used *radioactive Phosphorus 32* to label the *DNA* inside the virus in a *different* container. (Remember, phosphorus is one piece of a **nucleotide**. They can also follow the DNA using the Geiger counter.)
      3. The radioactive viruses where then exposed to bacteria. The viruses infected the bacteria. In the radioactive Sulfur container, the radioactive sulfur did NOT enter the bacteria. It remained outside the bacteria. When the viruses reproduced inside the bacteria, the reproduced viruses that came out of the dead bacteria were NOT radioactive. In the radioactive Phosphorus container, the radioactive phosphorus did enter the bacteria. When they reproduced inside the bacteria, the reproduced viruses that came out of the dead bacteria *were* radioactive from the phosphorus the possessed.
      4. This proved with 100% accuracy, that DNA was the “transformation agent” and that this carries the information “blueprint” from one generation to the next.
7. Erwin Chargaff (in 1947)
   1. He develops what becomes known as **Chargaff’s Rule**.
   2. The rule states that, *FOR ALL ORGANISMS***,** [A] = [T] and [C] = [G].
      1. This helps support the theme of *Unity* and *Diversity.* Unifying complementariness, as it *always the same* pairing of nucleotides. Diversity is in the percentages of each *grouped* nucleotide pairs *between* species.
      2. For example: If you know a species has 32% Thymine; then there must ALSO be 32% Adenine. (32+32= 64%.) This means that there is 36% unaccounted for. (100- 64 = 36.) Since this 36% is BOTH Cytosine and Guanine, divide by 2 to find the percentage of each. (36÷ 2 = 18) There exists 18% Cytosine and 18% Guanine.
8. Rosalind Franklin (in the 1950’s)
   1. She performed X-ray Crystallography on DNA. This picture was *extremely* important in helping Watson and Crick develop their model of DNA.
      1. The picture indicates the Double Helix structure of DNA (The picture would be from the view of *looking down* a strand of DNA. It would be similar to looking down a paper towel cardboard tube.)
      2. The picture also indicates that the Nitrogen Bases (the X in the center) point inward and are *equal* lengths in binding, because it is *always* one Pyrimidine (C and T) and one Purine (A and G).
      3. The large areas *around* the “X” are the sugar phosphate backbone of DNA.
9. James Watson and Francis Crick (in 1953)
   1. They constructed the first *accurate* model of DNA.
   2. They used Chargaff’s work and Franklin’s work to fill in the gaps that they could not figure out.
   3. The Double Helix backbone is composed of Phosphorus and the 5 Carbon sugar Deoxyribose. (It would be like the side supports on a ladder.)
   4. The “rungs or steps of the ladder” would be the Purine base + Pyrimidine Bases. (A=T and C=G)
   5. **Hydrogen Bonds** hold the two sides together and it is twisted into the Double Helix shape (It looks like a twisted ladder.) Remember, **Hydrogen bonds** are *weak* bonds. We will want to “open up” the DNA during DNA replication AND Protein Synthesis.

AP Biology

DNA Structure and Replication

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.8, 2.9, 2.22, 2.31, 2.32. 2.34, 2.36, 3.1, 3.3, 3.7, 3.8, 4.1, 4.3)

Important concepts from previous units:

1. Monomers of Nucleic Acids are called Nucleotides; Polymers are DNA or RNA.
2. Nucleotides are *linked* together by a *covalent* Phosphodiester bond*.*
3. The *sequence* of nucleotides *determines* what protein or enzyme is made (expressed).
4. **DNA Replication**
   1. The process of making of a **complete copy of an entire length of DNA.** (Applies to all Chromosomes.)
      1. This occurs *during*the **S-Phase** of the Cell Cycle for Mitosis or Meiosis.
      2. In bacteria, it is referred to as *Circular* or Theta replication. (Symbol for Greek letter Theta is: Θ.)
      3. In other organisms that possess chromosomes, it is referred to as *Linear* Replication.
   2. It is easy to do for cells because the two sides are **complimentary** (A with T and C with G always.)
   3. The **Semi-conservative Model** best explains the process of DNA replication.
      1. It shows one *original* DNA side serving as a *template (guide)* for making the *other DNA side*.
      2. Easy as A = T and C = G.
      3. The replication work is being done *in opposite directions*, but on *both sides at the same time*.
   4. In humans, it takes just a *few hours* to copy over 6 Billion nucleotides in our cells thanks to ENZYMES!
5. **Origins** Of Replication (**Starting points**)
   1. These are *specific nucleotide sequences* encoded in the DNA strands that act as “starting points”.
   2. The enzyme **helicase** *unwinds the DNA double helix* to create a **Replication Bubble** (This provides *“space”* to do the actual *building* work of making the *new complimentary side* of the *new DNA molecule* by other enzymes.)
      1. The *ends* of the bubbles are called **Replication Forks**. There is one on each end of the bubble.
      2. Work *is happening* on both sides of the forks *and* both sides of the bubbles.
      3. Many bubbles can be on the same DNA strand. (This speeds up the process of replication.)
6. DNA Replication *Elongation* 
   1. Elongation of the *new* DNA complimentary side *will require* the enzyme **DNA Polymerase III.** (This enzyme performs the *addition of new nucleotides*to the new DNA complimentary side and also acts as a *proofreader* to help prevent *errors* in construction from occurring. (Look at the name and see the function. Remember, “polymers” means “many units” or “many monomers”. In this case, the monomers are called nucleotides. The ending “ase” tells you it is an enzyme.)
      1. The enzyme works at a rate of about 500 nucleotides being *added per second*.
   2. **DNA Nucleosides** are brought to the enzyme from the *cytoplasm* of a cell. (Nucleosides were “created” from broken down DNA strands found in the cells or particles of *food during the process of digestion*.
      1. A **nucleoside** has *three* phosphates to supply the bonding process with energy. (Remember, to create a bond requires “free” energy.)
      2. The **nucleoside** will *lose two phosphates* in the bonding (attachment) process to the new DNA.
         1. Lose of phosphates makes it a nucleotide.
      3. This *saves* ATP for other cellular processes.
   3. The two sides of the Double Helix are said to be **Anti-parallel.** (This means that the DNA information runs in *different directions*.)
      1. DNA is **ALWAYS READ AND MADE 5’🡪 3’. (REMEMBER THIS IMPORTANT FACT!)**
         1. The 5’ Carbon of the sugar (Deoxyribose or Ribose) has a phosphate attached to it.
         2. The 1’ Carbon of the sugar has the Nitrogen Base attached to it.
         3. The 3’ Carbon of the sugar has an *open* bond. (This is the *connector site* for the *next* nucleoside.)
   4. **Helicase** enzyme causes the Double Helix to *unwind*.
   5. **Single-strand binding protein** keeps the two sides *apart and stable*. (Look at the name and see the function.)
   6. **Lead strand** of the replication fork (Remember, there are TWO forks going in OPPOSITE directions.)
      1. This strand runs in a *continuous 5’🡪3’ direction* as it opens. (It is leading the way in the process.)
      2. To start *adding* nucleosides, we first need to attach an **RNA Primer**. (Remember, RNA is a *disposable* form of DNA.) using **Primase** enzyme and go! (A “primer” is a *starting segment* of nucleotides. It will be *removed* later in the process and replaced with DNA or cut off if it is attached to a **telomere**, which are located at the chromosome ends.)
      3. Lead strands on *both* sides of the replication bubble are LOCATED DIAGONALLY from each other. (If it is on top on one end of the bubble, it will be on the bottom on the other side of the bubble.)
         1. This is because the two DNA strands are anti-parallel.
   7. **Lagging Strand**
      1. This side of the replication fork has DNA *not running in a 5’🡪3’ direction*. (Therefore it will always be *lagging* behind.)
      2. This side of the fork has to wait for *a long segment* of DNA to become *exposed* first before we can start by adding a primer.
      3. When a long segment has been “opened” by **Helicase**, a **RNA Primer** (disposable) will attach and then **DNA Polymerase III** will *work backwards* making an **Okazaki fragment**.
      4. When the **DNA Polymerase III**, on the *newly created* **Okazaki fragment**, reaches the *previous* RNA primer of the *previous* Okazaki fragment, the DNA Polymerase III will *remove* the old RNA primer and replace it with new DNA nucleotides. This keeps the *DNA intact*.
      5. The Okazaki fragments are “stitched” together using the enzyme **Ligase**.
      6. The lagging strands of *each fork* on BOTH sides of the replication bubble are LOCATED DIAGONALLY ALSO.
7. Correction of Errors (Proofreading)
   1. This function is performed by **DNA Polymerase III** as the *new* DNA strand is being made.
      1. **Mismatch Repair** is when the *wrong nucleotide* is added to the new sequence. DNA Polymerase will *reverse* a spot, *remove* the wrong nucleotide, and then *replace* with the *correct* nucleotide. (This would be equivalent to you hitting the following computer keyboard buttons Backspace/Delete and then continue when you make a typo while you are trying to write an English paper.)
   2. For errors that are “created” (what are called Mutations) *after* the DNA has been made – **Nucleotide** **Excision Repair** is used to correct these, if possible.
      1. Step 1: **Nuclease** –cuts *around* the faulty pairing so they can be removed.
      2. Step 2: **DNA Polymerase III** – *replaces* the missing nucleotides.
      3. Step 3: **Ligase** - *stitches back together* the fragments.
8. **Telomeres** (TTAGGG is the nucleotide sequence) (“Telo” means “last”; “mere” means “unit”)
   1. These are *repeated* nucleotide sequences found at the *ends of chromosomes* that are used for RNA primers to attach to start replication, without having a bubble.
   2. The *number* of telomeres depends on the cell type. (It can range from 1 –10,000 telomeres. Heart cells and brain cells have VERY few. Skin cells have thousands.)
   3. Having these *protects* the important DNA information from replication erosion. Telomeres are *disposable.*
   4. **Apoptosis** (This is **programmed** cell death.) This is important in creating the spaces between your toes and fingers. Otherwise you would have fins for feet and hands. It is because the cells run out of Telomeres, so they *do not reproduce*. Thus when they die, they “create” the gaps.
9. **Telomerase**
   1. This is the enzyme that *replaces telomeres during fetal development*. After the fetus is *fully* developed, this enzyme *shuts off* and degrades over time. The DNA segment (called a gene) that is responsible for providing the “blueprint” on how to make this enzyme will become heavily methylated.
   2. Normally the active gene is found in gamete producing **germ** cells – Cancer? When this enzyme is turned back on *in children or adults* it leads to *abnormally fast growth of cells*. This abnormal growing group of cells is called a **tumor.** Some can be malignant and some can be benign.

AP Biology

Protein Synthesis – Part 1

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.22, 2.31, 2.33, 2.36, 2.37, 3.4, 3.6, 3.18, 3.24, 3.25, 4.1, 4.2, 4.3, 4.4)

Important concepts from previous units:

1. Proteins are constructed from the macromolecules called amino acids.
2. Amino Acids are linked by a covalent peptide bond by a hydrolysis reaction.
3. DNA is more stable than RNA; but RNA evolved before DNA did.
4. George Beadle and Edward Tatum (1934)
   1. They develop the one gene-one enzyme hypothesis. This proposes that a single gene has the genetic information for making one enzyme. This is later changed to become the **one gene - one polypeptide** (protein) hypothesis; as enzymes are a *type* of polypeptide (protein).
5. **Transcription** (means “ the process of making a *working copy* of an original”)
   1. This process is the making of a *disposable copy* of DNA but in the form of RNA**.** The *disposable* copy will become known as mRNA – messenger RNA. It is a *disposable copy* of the “Million Dollar DNA Blueprint”.
      1. The *message* (mRNA) will be sent to the *construction site* (ribosomes) *for building* the protein.
      2. RNA nucleotides use Ribose *instead* of Deoxyribose as the five carbon sugar. This makes the RNA *less stable* than DNA; that is good since the cell only needs it to send a *temporary* message on how to construct the protein.
      3. In RNA, Uracil replaces Thymine. Thymine can’t exit nuclear pores. Remember, ribosomes are out in the cytoplasm, so Thymine needs to be substituted by Uracil.
   2. DNA serves as a **template** (guide) for making the mRNA. A = U and C = G (Still can use **Chargaff’s Rule**.)
   3. In Eukaryotic cells, the mRNA *must*first be “modified” before translation can occur. The modification occurs in the nucleus. (Prokaryotes **DO NOT** modify the mRNA. They sent it to the ribosome “as is”.)
      1. **Primary Transcript** (before modification) is modified to produce the **secondary transcript** (*after*modification has occurred). The **secondary transcript** is what will be sent out of the nucleus to the ribosome.
   4. This is considered the *first part* of **Protein Synthesi**s**.**
6. **Translation** (means “The process of taking from one language and *changing* to another language”)
   1. In this process, the cell is *turning* nucleotide language (DNA/RNA) into amino acid language to make proteins. Remember, amino acids are the building blocks of proteins.
   2. This process occurs at the Ribosome. The ribosome has a nickname… “the Translator”. It is also considered a “construction site” since the cell is *building* a protein.
   3. This is considered the *second part* of **Protein Synthesis.**
7. **Codon** “A.K.A Triplet Code” (This is the amino acid language.)
   1. Codons are *determined* by the template strand of DNA (Important Blueprint Information) but are READ ON THE RNA! (The mRNA is what is being translated; not the DNA.)
      1. **The codons MUST be read 5’ 🡪3’on the mRNA!** (Because this is how the mRNA was made. You do not write a sentence and then read it backwards do you. It would make no sense.)
   2. **RNA Codon Chart** for Amino Acids (Contains the 20 known amino acids for living organisms.)
      1. The chart was started by Marshall Nirenberg (early 1960’s) (He won a Noble Prize for this.)
         1. UUU- Phenylalanine was the first one recorded.
      2. 61of the 64 possible codons ( 4³) codes for an Amino Acid.
         1. 4 refers to the four nucleotides possible (A, C, U, G); 3 refers to the number of pieces in a UNIT (codon).
      3. **AUG** is the **start codon** or Methionine. It depends on the *position* in the mRNA. If it is the first codon on the 5’ end, it will be the start codon. If it is not the first, it will be regular methionine.
      4. **UAA, UAG**, and **UGA** are the **stop codons.** These codons stop the process of transcription.
      5. Redundancy is *wasteful*, so enter Inosine. This nucleotide can act as a “Wild Card”. When put in the *third* position of an **anticodon**, it can represent *any* nucleotide. (This is useful for such amino acid sequences such as Serine or Arginine.)
      6. This chart is *universal*  for *all* living organisms and vituses. **(**Viruses are *not* living.) This hits on the theme of *Unity* and *Diversity*. Unity in that it indicates Common Ancestry among all organisms and viruses. Diversity is in the differences of the *sequences* of amino acids strung together to make a protein.)
         1. Remember, RNA evolved first (but it is unstable) then mutated to DNA (which is more stable). This is why ALL living organisms and most viruses are DNA based.
   3. **Reading Frame** - This term refers to a set of *3 consecutive nucleotides* read in **5’ 🡪 3’** direction.
8. **RNA** **Synthesis** and **Modification** -The *making* of mRNA. This process occurs at the **nucleolus**. (Remember,

the nucleolus is “like” a copy machine because we are making a *cheap*

*disposable copy*of the DNA sequence.)

* 1. Three Phases of production to **a transcription unit** - piece of mRNA.
     1. **Initiation** - This is building our *factory* to make mRNA basically.
        1. A protein called a **Transcription Factor** attaches to the **TATA box** to *determine* the *direction* the “factory” will proceed. The **TATA box** is part of the **promoter** sequence. (Look at the TATA sequence, can you see it running in *different* directions. This orients the “factory” to the direction it will transcribe.)
        2. Then additional transcription factors (proteins and enzymes) are added to the “factory”.
        3. Finally**, RNA Polymerase II** joins to *complete* the factory. The whole “factory” is called a **Transcription Initiation Complex**. (Can you see the definition in the term? Transcription is the process being done. Initiation refers to the beginning process. Complex indicates we have many parts involved in making the structure.)
        4. This is a *step by step**controlled process*. (The cell “controls” each step to help make sure nothing goes wrong.)
     2. **Elongation** - This refers to the *actual making* of the mRNA molecule.
        1. This *must* bemade in the **5’ 🡪 3’** direction!
        2. **RNA Polymerase II** *separates* the DNA Double Helix to *make room to work*.
        3. **RNA Polymerase II** also *adds nucleosides* to the growing molecule.

i. Nucleosides come from the cytoplasm. **Lysosomes** recycle and provide them.

* + - 1. After **RNA Polymerase II** has past the DNA transcription point, the DNA reforms the helix.
      2. Cells can make *multiple copies* of RNA because the DNA is *left intact and protected* in the nucleus.
    1. **Termination** (Just like it sounds… stop the transcription.)
       1. A **stop codon** is made (for the ribosome) and the “factory” molecule slows down.
       2. **RNA Polymerase II** slows down *until*  it stops transcription by forming an AAUAAA sequence and is then released from the DNA.
  1. **Modification** of the Primary Transcript for EUKARYOTIC Cells (This also occurs in the nucleus.)
     1. **Front end (5’) modification** of the mRNA molecule.
        1. A 5’ *protective cap* is added. (This would be like you putting on a hard hat to protect your head when you go outside into a “construction site”.)
        2. This cap acts as a *signal* to the ribosome particles, telling it where to attach.
     2. **Back end (3’) modification** of the mRNA molecule.
        1. A Poly A Tail is added. (“poly” means “many”; 50-250 Adenines will be added onto the tail.)
        2. This acts as protection against *digestive enzymes* in the cytoplasm. (Remember, it is a construction site and things are being broken down as well as being built.)
     3. **Middle modification** of the mRNA molecule. (This modification is referred to as **RNA** **Alternative Splicing**.)
        1. During this step*, remove* the non-coding **introns** (These act as spacers) using **Spliceosomes**. (A **spliceosome** is a *collection* of snRNPs.)

i. **snRNP’s** (**s**mall **n**uclear **r**ibo**n**ucleic **p**roteins act as scissors.) Remember, Ribozymes are RNA molecules that *act as enzymes*.

* + - 1. Then *rearrange* the *separated* coding **exons** (*important blueprint pieces*.) to the *needed* configuration. This is why it is called *alternative*; pieces (exons) can be *rearranged* in *different orders*. (This is REALLY important in the making of antibodies by our immune system. Remember the *variable* portion of the antibody structure.)
      2. “Stitch” the pieces together to make the *finalized* **secondary mRNA transcript** that is know *ready* for transport to the ribosomes for translation into proteins.

AP Biology

Protein Synthesis – Part 2

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.22, 2.31, 2.33, 2.36, 2.37, 3.4, 3.6, 3.18, 3.24, 3.25, 4.1, 4.2, 4.3, 4.4)

Important concepts from previous units:

1. Amino Acids are the building block macromolecules of proteins.
2. Amino acids are linked together by covalent peptide bonds in a dehydration reaction.
3. Proteins have to be *folded* in order to work; this involves *hydrogen bonds* (2’) and *disulfide bridges* (3’).
4. **Translation** - This is the part of *actually making* the protein.
   1. This process occurs at the **Ribosome** “the Translator”.
   2. The process *turns* the mRNA into a primary (1’) sequence of amino acids for *making* of the protein.
   3. This process needs the assistance of **tRNA (transfer RNA)** to transfer *free amino acids* from the cytoplasm to the construction site of the Ribosome.
      1. Free amino acids are provided by the digestive system, by **catabolism** (breakdown) of proteins in food, and then delivered to the cells by the blood vessels. Inside the cells, they are used for **anabolism** (building) of proteins or undergo **deamination** (removal of the amine functional group) for ATP production in cellular respiration.
   4. There are 45 different tRNA molecules for 61 possible codon combinations.
      1. **Inosine** (acts as a “wild card”) makes it possible for a cell to *conserve materials and energy*.
      2. The use of **Inosine** creates the “**Wobble effect**” - It does not fit *perfectly*, but gets the job done.
      3. **Inosine** is found in the *third slot* of the **anticodon** *only.*
         1. Remember, that the ANTICODON is found on the tRNA molecule, NOT the mRNA.
      4. The **Anticodon** “matches” the **codon** on the mRNA molecule ensuring the *correct* amino acid is brought to the construction site of the Ribosome. If they DO NOT match … it is the wrong Amino Acid!
      5. The amino acid is connected to *the 3’ end* of the tRNA molecule.
         1. Remember, the tRNA molecule is a nucleotide sequence; so there is a phosphate on the 5’ end and an *open bond* on the 3’ end… so this is where the amino acid gets attached so that it can be transported to the ribosome (construction site).
      6. This *connection* between the tRNA molecule and the amino acid is constructed using the **Aminoacyl – tRNA synthetase** enzyme. (Can you see the definition in the name?)
   5. **Ribosome** Structure (This cellular *particle* has 2 parts.)
      1. The **Small sub-unit** - This part acts as a *platform for work*; much like your desk.
      2. The **Large sub-unit** - This part is the *factory for making* the protein.
         1. The **A** **site** - This is where the *next* tRNA molecule is ADDED in the “factory”.
         2. The **P** **site** - This is the part of the “factory” where the PROTEIN is attached.
         3. The **E** **site** - This is where the “used tRNA molecule” EXITS the “factory” to be *reused*.
      3. The ribosome “*walks*” down the mRNA *one codon at a time* until it gets to the stop codon at the end of the mRNA molecule. Thus having completed the “message” on how to make that particular protein. This “walking” is called **Translocation.** (Can you see function in the name?)
      4. Remember, these are NOTorganelles. *All cells possess these structures*.
   6. The process of **translation** has three phases: (They are the *same 3* as **Transcription**.)
      1. **Initiation** - This is *building the factory* needed to make the protein.
         1. The **small sub-unit** attaches to the **5’ cap.** This *interaction* signals the large sub unit.
         2. **AUG** (the *start codon* on the mRNA molecule) brings in the tRNA (using the **anticodon**) molecule with Methionine attached. This *starts production* of our protein.
         3. Then the **large sub-unit** is brought in using **initiation factors** (these are enzymes) and uses **GTP** for energy in the process. (Remember, GTP is “like” ATP…both are energy molecules.)
         4. The **large sub-unit** is *aligned* so that Methionine is in the **P site**. The **A site** is *open* for the *addition* of the next tRNA molecule.
      2. **Elongation** - This is the *actual making* of the 1’ sequence of amino acids.
         1. The ribosome **translocates** (“walks”) down the mRNA *one codon at a time* using **GTP**.
         2. This adds a single amino acid, using tRNA, to the *open* **A site** using **GTP** each time.
         3. Another GTP is used to make **peptide bond** between the amino acids of the P and A sites.
         4. The rate of addition is *controlled*by **elongation factors** (enzymes).
      3. **Termination**
         1. This occurs when a termination codon reaches the A site.
         2. A **Release factor** (enzyme) enters the A site causing a **hydrolysis reaction** to occur that *releases* the protein from the last tRNA molecule (which is sitting in the P site).
         3. After the hydrolysis reaction occurs, the ribosome detaches and the sub units separate to be *reused*.
      4. The mRNA may be *reused* to make *more* of that particular protein or it may be broken down and the nucleotides *recycled*, as it is temporary RNA.
         1. **Polyribosomes** (many ribosomes)can also occur on a strand of mRNA.
         2. This allows for a cell to make many copies of the *same* protein very quickly. (Such as might be needed during repair or making antibodies.)
5. **POST (means “after”)** **Translation Modification** (This is the *protein folding* that *must* occur for the protein to be functional.)
   1. If the 1’ sequence enters a **Chaperonin**, the protein *will stay inside* the cell.
      1. Entry is “guarded” by a **Signal Recognition Particle (SRP)** *inside* the bottom piece.
   2. If the 1’ sequence enters the **RER**, the protein *will be exported out of the cell***.**
      1. Signal Peptide on the 1’ sequence. (This acts as a *siren*. It is “like” yelling “Take me to the RER!”)
      2. **S**ignal **R**ecognition **P**article (SRP) - This particle acts as a *guide* leading the 1’ sequence to the RER. It attaches to the Signal.
6. **Proteomics** (Study of Proteins)
   1. The study of genes and the corresponding polypeptide made by that gene segment of DNA. (Remember the one gene one polypeptide hypothesis.)

AP Biology

Protein Synthesis – Part 3

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.22, 2.24, 3.1, 3.4, 3.5, 3.6, 3.18, 3.25, 4.1, 4.2, 4.3, 4.7, 4.24)

Important concepts from previous units:

1. A *change* in the nucleotide sequence is called a mutation.
2. Some mutations can cause cancer (abnormal growth) in organisms.
3. Prokaryotes would have, over millions of years, given rise to Eukaryotes. (Endosymbiant Hypothesis)
4. Types of RNA
5. Prokaryotes vs. Eukaryotes
   1. Prokaryotes DO NOThave **introns** that need to be removed prior to Translation. The 1’ transcript goes *straight to the ribosome* for Translation.
   2. **Genetic engineering**? We can take a 2’ transcript out of a *Eukaryotic organism*. Use the enzyme **reverse transcriptase** to turn the mRNA molecule *back into* a DNA molecule. *Insert* the new DNA strand into bacteria. The bacteria will then be able to Transcribe and Translate off of this new inserted DNA and thus *make that protein*. This has been done for numerous human medicines such as Insulin or Human Growth Hormone.
   3. Eukaryotes DO have **introns**. This allows them to take out the introns and *rearrange the important exon* *pieces* to make an *almost unlimited number of different* proteins. This simple fact is the reason that humans are so vastly more complex than simple bacteria.
   4. The two types of cells basically do the *same process* of Transcription and Translation to make proteins. This *indicates* common ancestry among all organisms. (Unity *again*.)
6. **Mutations** 
   1. *Change in the nucleotide sequence* of DNA or mRNA that code for a protein.
   2. Caused by **Mutagens** (Means to “**gen**erate a **muta**tion”.)
      1. These are a *physical or chemical interactions* that changes the nucleotide sequence of DNA.
      2. Examples of mutagens:
         1. Ultraviolet radiation (UV Radiation) from the sun
         2. Cigarette Smoke
         3. Alcohol in excess
         4. Viruses
         5. Car Exhaust
         6. Chemicals (Laboratory, Pesticides, insecticides, poisons)
   3. Two major typesof Mutations:
      1. **POINT** **mutations** - A *single nucleotide* mutates thus affecting *a single codon*.
         1. **Silent** Point Mutation– The mutation causes *no change in the amino acid* coded for.

(We would never know because it has no effect. This can happen because the codon coding is *redundant,* remember?)

* + - 1. **Missense** Point Mutation – The mutation *changes the amino acid* coded for. (MIStake)

(This is best seen in the mutation that causes Sickle cell.)

* + - 1. **Nonsense** Point Mutation – The mutation *changes from coding for an amino acid to coding*

*for a STOP codon* . NO protein will be made. (NO sense)

* + 1. **READING FRAMESHIFT** **Mutation** (The whole DNA “sentence” is changed)
       1. These mutations alter the *codon sequence*.
       2. **Insertion** – *adding nucleotides* to the sequence.

For Example:THE BIG TAN DOG RAN

with *Inserted* Letter**:** THE B**O**I GTA NDO GRA N

* + - 1. **Deletion** – *taking out nucleotides* from the sequence.

For Example: THE BIG TAN DOG RAN

with *Deleted* Letter: THE BGT AND OGR AN

* 1. Gametes vs. Somatic – Who is affected? If a mutation occurs in somatic cells, the only one affected by the mutation is *the person that the mutation occurred to*. If the mutation occurs in gametes (sex cells), the only one affected will be the organism *“created”* from that sex cell. This is how *future* *generations* may be affected and *this is a cause of evolution*. CHANGE in DNA over TIME.

AP Biology

Eukaryotic DNA Control Mechanisms for Gene *Expression*

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.22, 2.31, 2.37, 3.4, 3.20, 3.21, 3.23, 4.7, 4.17)

**Important** concepts from previous units:

1. During Interphase – The DNA is loose for *easy access* for transcription. (It is “like” a bowl of spaghetti.)
2. During Mitosis or Meiosis – The DNA is tightly wound for *easy separation*. (Look like an “X”.)
3. Chromosome Structure in Eukaryotes
   1. **Histones** - These are *proteins* that are used for DNA to wrap around and thereby helping it to *condense*.
      1. These carry a *positive charge*. (Remember, DNA is negatively charged, so it is like a magnet.)
      2. Evolution? *All Eukaryotes* and a group of Bacteria, Archae bacteria, possess histones. This indicates *common ancestry* among these organisms.
   2. **Nucleosome**  - A *unit* of DNA wrapped around a group of histones. (Nucleotides around histones.)
   3. **Supercoiling** – This is the process of DNA *condensing* from Chromatin to Chromosomes.
   4. **Heterochromatin** - This refers to DNA that *remains condensed* even during interphase. – It is NOT active.
      1. This CANNOT do transcription so it is *inactivated.* (“hetero” means “different”)
   5. **Euchromatin** - This refers to DNA that IS *loose* during interphase. – It IS active**.**
      1. It CAN do transcription and *be expressed*. (“ Eu” means “true”)
4. **Cellular Differentiation** (A.K.A. **Specialization**) - The process of making cells “different” or “special in

function”.

* 1. This process is accomplished by turning certain genes *“on” or “off”.* This is known as **Differential Gene** **Expression**. This accounts for about 1.5% of our total DNA genone. These genes are the Exons.
     1. The genes turned “on” end up making that *protein/enzyme* to make that cell different or special.
  2. Control goes awry? Terrible things may occur such as death or cancer to the cell or organism.

1. Gene control during **transcription** (A through F are associated with *transcription*.)
   1. Is the DNA in a state of Heterochromatin vs. Euchromatin?
   2. **DNA Methylation** of the DNA
      1. This refers to putting a heavy “coat” of methyl (CH3 ) groups of the DNA, thus *preventing* transcription from occurring. The Methyl groups attach to Cytosine or Adenine nucleotides.
      2. This is the source of **Genomic Imprinting** that occurs in gamete production. It essentially *“erases”* information”.
   3. **Histone Acetylation**
      1. This is the attaching of acetyl (COCH3 ) groups to the histones lysine amino acids.
      2. This attaching *breaks the bond* between the DNA and the histones by covering up the positive charges thus creating NO attraction for each other.
      3. This allows for RNA Polymerase and transcription factors to attach to the *“freed”* DNA so that transcription may occur.
   4. Building of the Transcription Initiation Complex (factory). (Remember, this is a *step by step process*. Each

step can be controlled.)

* + 1. **Enhancers** and **Activators** - These help control *the rate* of transcription. They are segments of DNA that basically “grab” the factory, using a bending protein, and move it down the DNA faster thus *enhancing* the process of transcription. They are “Pushers”.
       1. They are always *in front* of gene to be transcribed.
    2. **Repressor** or **Silencer** - These *control* proteins sit on the TATA box – they *prevent* transcription from occurring. This *silences* or *represses* the gene from being expressed.
    3. Both are called **control elements**, because the control the *rate* of transcription**.**
  1. **Coordinated** Control of gene families
     1. The *same chemical signal* causes the simultaneous expression of multiple copies of the *same* gene These multiple copies of the SAME gene are referred to as a **gene** **family**. (Hemoglobin, for red blood cells is an example. We need hundreds of copies of this gene to make the trillions of Red Blood cells our bodies need to deliver oxygen through our body. Coordinated control is essential. It would also be like the bell at the end of the period *signaling* all classrooms to move to the next class at the *same* time)
  2. **Micro RNA (miRNA)** and small **interfering RNA (siRNA)**
     1. These are *little pieces of RNA* that attach to **mRNA** and thus *control transcription* of the mRNA.

1. *Post*Transcription Regulation
   1. **Alternative RNA Splicing** using **Spliceosomes (snRPS).** (Primary becoming a Secondary transcript is controlled.)
   2. Cytoplasmic Degradation - This occurs because of enzymes in the cytoplasm.
      1. This refers to the removal of caps and tails on mRNA molecules, followed by nucleotide sequence catabolism, so they may be recycled. The more As in the Poly A tail, the *longer* the mRNA will last in the cytoplasm.
2. **Translation** Control Mechanisms
   1. Building of the Translation Initiation Complex (Ribosome Factory) This is *also* a step by step process.
   2. If a Faulty 5’ cap (signal) is attached, it will prevent Translation from occurring.
3. *Post* Translation Control Mechanisms
   1. **Chaperonin** or **SRP** for RER. (Where does the 1’ sequence go for folding to occur?)
   2. **Phosphorylation** of the protein/enzyme. (Remember, this is *activating* the molecule by using ATP to *add* a phosphate.) On vs. Off basically.
   3. Transport through the inter-membrane system (As the protein moves through the RER and Golgi, controlling the folding and modification of the protein.)
   4. **Proteosomes** (special protein digesting Lysosomes) control HOW LONG the protein lasts.

AP Biology

Biotechnology Part 1 - Viruses and Gene Therapy

(Associated Learning Objectives: 3.14, 1.15, 1.16, 2.24, 3.1, 3.5, 3.22, 3.29, 3.30, 4.5, 4.6, 4.22)

1. Viral Structure
   1. Viral Genome
      1. Viruses possess either a *double or single strand* of DNA or RNA (This is how viruses are classified.)
      2. Viruses contain very small amounts of DNA or RNA– most are 4 to 500 genes total.
   2. Viral Protein Coat (Referred to as the **Capsid**.)
      1. The Capsid serves two purposes:
         1. *Protection* of the DNA or RNA strands inside.
         2. *Attachment* of the virus to a host cell.
      2. It is built from protein units called **capsomeres**. (means “capsid unit”)
      3. Some viruses can also have a **viral envelope**.
         1. This is a “cloak” derived from the *previous host cell* plasma membrane. (It is an example of mimicry. It looks like a normal cell, but it is actually like a Trojan horse. The danger is inside.)
         2. The AIDS/HIV virus has a viral envelope derived from the T-helper white blood cells.
   3. **Bacteriophages** (A.K.A. **Phages**) – These are viruses that attack bacteria.
      1. These are some of the largest and most complex viruses.
   4. Viruses are NOT living organisms. They cannot be “killed”. They *can* be **denatured** using chemicals though. Some of these chemicals are in anti-viral products you may use, like hand soaps or Kleenex.
2. Viral Reproduction
   1. Viruses *must* have a host cell in order to reproduce. They are considered **Obligate Intracellular** **Parasites**. As the name indicates, viruses *must get inside the host cell* in order to reproduce.
   2. Viruses *need* to use the host cells ribosomes and enzymes to make new DNA or RNA strands and new capsomeres to form new viruses.
   3. **Host Range** – Refers to what organisms a virus can attack. It is determined by recognition of certain *glycoproteins or glycolipids on the host cell membrane.* (Sounds like cell signaling again.)
   4. **Restriction enzymes** – These enzymes, found in bacteria, act as primitive *defense* against viruses. These enzymes *cut up the genome* and thus *inactivate* the genes from being transcribed. They are called *restriction* enzymes because they *only* cut at *certain nucleotide sequences*. In other words, they are *restricted*  in where they can cut.
3. Basic virus life cycles
   1. **Lytic** Life cycle- This cycle *destroys (lysis) the host cell* when the virus leaves. (A.K.A. **Virulent.** Sounds like violent. )
   2. **Lysogenic** Life cycle – In this type of cycle, the virus *permanently incorporates* its DNA into the host genome, but does not immediately kill the host. (A.K.A. **Temperate**. sounds like Temper.) The virus “lives” inside the host cell. As the host cell *reproduces by mitosis*, so does the virus. When the virus becomes aggravated, it pulls out its genome, reproduces, and the leaves the cell by lysis and thereby killing the host in the process.
      1. **Prophage** or **Provirus** (These terms both refer to the inserted viral DNA in the bacterial DNA.)
      2. Herpes and HPV are both examples of viruses that have a lysogenic life cycle. When they get aggravated and cause destruction of host cells, blisters form the destroyed cells. We call them fever blisters or cold sores in the case of a Herpes Type I (around the mouth) infection.
4. **Retroviruses**
   1. Retroviruses are a unique type of viruses. (“retro” means “reverse or backward”)
      1. They use **REVERSE TRANSCRIPTASE,** an enzyme, to turn RNA into DNA (It. does transcription *backwards*. It turns “mRNA” into double stranded DNA, so that it can incorporate into the host DNA.
   2. AIDS/HIV and the common cold virus are both retroviruses.
5. **Gene Therapy**
   1. Genes that are coding for proteins or enzymes are inserted into viral capsids. The viruses are then injected into individuals possessing genetic diseases associated with *missing or non-functional* proteins or enzymes in an effort to *treat* the person suffering from the condition. The DNA is hopefully taken up by the cells.

AP Biology

Biotechnology – Part 2 -Bacterial Genomes

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.22, 2.31, 2.33, 2.36, 2.37, 3.1, 3.18, 3.23, 3.24, 3.26, 3.27, 3.29, 3.30, 4.1, 4.17, 4.22,)

**Important** concepts from previous units:

1. Bacterial DNA has circular chromosomes.

2) Bacteria (Prokaryotes) do NOT possess **introns.**

3) Bacteria reproduce by binary fission.

1. Bacterial Genome
   1. They possess *one circular* strand of DNA that is located in the **nucleoid** region.
   2. **Plasmids**
      1. These are small, circular, *exchangeable pieces* of DNA.
      2. These are *in addition* the main large circular DNA strand.
      3. These help to *increase variation and survival*.
   3. Bacterial Replication
      1. *100% Identical clones* are produced through binary fission. Allows them to reproduce *very quickly*.
   4. Ribosomes are needed to create proteins; but they do NOT possess any organelles.
2. Bacterial Variation Processes (Remember, variation *increases survival* chances in a *changing* environment.)
   1. **Transformation** (This means simple change.)
      1. A bacteria took in DNA from an external source. (Recombination of DNA occurred.)
      2. Biotechnology? This is what we do to make bacteria “learn” new tricks, such as eat crude oil.
   2. **Transduction** (This is new DNA has been *carried in* by a virus thus creating the “change”.)
      1. Phage introduced the new DNA into the bacterial DNA.
   3. **Conjugation** (This is “Bacterial sex”.)
      1. Bacteria exchange plasmids through a conjugation tube from the “male” to the “female” (Bacteria DO NOT have sexes like humans do.)
      2. **F factor** (If a bacteria possess this gene, they are considered “male”; Shown as F+); (F- are “female”. They do NOT possess the F factor gene.)
         1. **Pili** – This structure is a protein “sex whip” for pulling the “female” close so that a conjugation tube can be made between the two bacteria. The pili is created by *expressing* the F factor gene.
      3. **R Plasmids**
         1. These plasmids exchange *antibiotic* ***R****esistance genetic information*. This *helped* in the evolution of MRSA (Methicillin-Resistant Staphylococcus aureus)
3. Transcription *Control Mechanisms* (Remember, these are ways to *control* Gene Expression.)
   1. **Transposons** “Jumping Genes” (These DNA *segments* act as *“Blockers”* to transcription.)
      1. Barbara McClintock discovered this control mechanism in the1940’s. She worked with Maize. She won a Nobel Prize for this work.
      2. Two types of transposons that exist:
         1. **Basic Insertion**

i. This is the *simplest* form.

ii. **Transposase** – enzyme that allows the DNA *to “jump” from location to location.*

* + - 1. **Composite (means** “complex”)

i. Transposase on *both sides of a resistance gene* are “jumping” as a unit.

* + 1. These *also* occur in Eukaryotes. They also are control mechanisms in these cells too. Another example helping to show *common ancestry* among the life forms on Earth.
  1. **Operon System** “operator”
     1. Francois Jacob and Jacques Monod discovered this control mechanism.(1961)
     2. **Operon** “operator” *controls* RNA Polymerase *access* to the DNA strand.
     3. *Operon* is part of the **promoter** sequence. It is located between the TATA box and Start codon.
     4. **Repressor** and **co-repressor** - These *molecules* act as an “off “switch.
     5. **Inducer** - This *molecule* acts as an “on” switch.
     6. These are *both* Negative Feedback loops. (They stop a process that is occurring, and gets it going in the opposite direction.)
     7. These are considered *regulatory genes* as well.

AP Biology

Biotechnology Part 3

(Associated Learning Objectives: 1.5, 1.14, 1.15, 1.16, 2.24, 3.1, 3.5, 3.6, 3.13, 3.18, 3.21, 3.26, 3.28, 4.1, 4.22)

**Important** concepts from previous units:

1. Nucleotides *always* base pair the same way – A with T and C with G
2. *All* living organisms, and some viruses, have DNA as the inheritable form of information transfer.
3. **Genetic Engineering** - The field of science dealing with *manipulating* genomes.
   1. **Recombinant DNA** is the major focus of genetic engineering.
      1. In this process, DNA from *two different sources* is combined into *one* molecule of DNA.
   2. **Biotechnology** - This term refers to the use of *computers and other devices* to help in performing science.
4. **Bacterial Cloning** Process
   1. The first step in this process uses **restriction enzymes** to create “Sticky Ends” on a **plasmid** *and* DNA from another source.
      1. These are enzymes that cut DNA at *specific nucleotide sequences*.
         1. This specific DNA sequence is referred to as the **restriction site**.
      2. These enzymes create **restriction fragments** as the DNA source is cut up into *fragments*.
      3. The *same restriction enzyme* must be used on *both* the bacterial plasmid and the DNA source.
   2. The second step is to *introduce the fragments* to the “open” plasmids for *recombination* to occur.
      1. The “sticky ends” base pairs will match allowing for recombination to occur.
   3. The third step uses the enzyme **Ligase** to *seal* the DNA fragments together.
   4. The fourth step is to introduce the *recombined* plasmids *back into* the bacteria. The bacteria are also called a **Cloning Vector**. A **vector** is a *carrier* organism.
   5. The fifth step is to allow the bacteria to *reproduce*, by binary fission, to achieve *a large working population*.
   6. The sixth step is to *identify* the bacteria of interest (the bacteria containing the recombined plasmid of importance inside) using by **Nucleic Acid Hybridization**.
      1. First, *create* a *radioactive* nucleic acid probes using radioactive Phosphorus. This will have the complimentary nucleotide sequence to the gene of interest.
         1. Remember the Hershey- Chase Experiment.
      2. Then *denature* the DNA double helix using *heat*. (The DNA double strand *separates*.)
      3. The radioactive probe *seeks out* the gene of interest and attaches to it, as the nucleotide sequences *match.*
      4. The next step is to use film filter paper to identify radioactive colonies of bacteria.
         1. The radioactivity will cause a *color change* on the film. This will tell where within the Petri dish the important bacteria are located.
      5. Now *separate the colonies* of interest from “trash” colonies. These bacteria *will make* our protein of interest. (For example, making human insulin or human growth hormone.)
   7. The last step is to culture (grow) the bacteria for experimentation and perform protein screening to *verify* the protein is being produced by the bacteria.
   8. Reproduced recombinant plasmids will *be stored* in **Genomic Libraries** for future use.
5. Problems going from eukaryotes 🡪 prokaryotes in making proteins.
   1. The **introns** *must be removed* from the eukaryotic DNA first. (Remember, Prokaryotes *do not* have introns.)
      1. Scientists have to collect the *modified mRNA* that *exits* the nucleus first.
      2. Then they need the enzyme **reverse transcriptase** to turn the single stranded m RNA molecule *back into* a double stranded DNA molecule.
         1. The “new” DNA molecule is known as **cDNA. (Complimentary DNA)** A copy of this cDNA molecule will be stored in a **cDNA library**.
   2. Need to then attach a **promoter** sequence (**expression vector**) at the *beginning* of the c DNA molecule so that a transcription complex (“factory”) can be build.
   3. Then attach “sticky end” sequences and insert into the bacteria to start production.
6. Yeast Artificial Chromosomes (YAC’s) - Process for “building” a chromosome with *multiple genes* for cloning.
   1. Yeast are single celled *fungus*. (These are Eukaryotic organisms.)
   2. They *will* recognize introns; therefore scientists can use *straight DNA from the source*. They *do not* have to acquire mRNA and perform the above procedure.
   3. Then they *recombine* all the DNA segments using **Ligase**.
   4. Attach a **Centromere** for Mitosis. (Remember, this is the *spindle fiber attachment point* on chromosomes.)
   5. Attach *numerous* **telomeres** using the enzyme **telomerase.** These are for *replication* during the S phase.
   6. Introduce the *artificial chromosome* to the yeast cell by **Electroporation** (Electrical Shock).

AP Biology

Biotechnology Part 4

(Associated Learning Objectives: 1.5, 1.14, 1.15, 1.16, 2.8, 3.1, 3.5, 3.13, 3.24, 3.26, 3.28, 4.1, 4.6, 4.22, 4.25)

Important concepts from previous units:

1. DNA has a *negative charge* because of the negatively charged phosphates in the sugar-phosphate “backbone”.
2. Like charges *repel* and opposite charges *attract*.
3. **Polymerase Chain Reaction** (PCR) (Requires *no organism* in the production of *new* DNA molecules.)
   1. The process was developed in 1983 by Kary Mullis. He won a Nobel Prize in 1993 for this.
   2. The process is used to turn a *single* molecule of DNA into a *large, workable sample* of 100% identical DNA molecules.
      1. This is widely used in criminal forensics (Murder cases).
   3. The process:
      1. Put the DNA sample in a PCR **Thermal Cycler** machine.
         1. The machine uses *heat*, DNA Primers, enzymes and a constant supply of nucleosides to build DNA molecules that are *identical* to the original molecule in nucleotide sequence.
         2. First step: *Heat* is used to *separate* the DNA double helix so that replication can occur.
         3. Second step: The attachment of a DNA Primer to the template DNA strand will occur to start replication.
         4. Third step: The **DNA polymerase** enzyme *works* 5’🡪3’ attaching nucleosides to the growing “new” side of the replicated DNA molecule.
         5. Fourth step: *Cool* the mixture to recombine DNA back into a double strand.
         6. *Repeat the cycle many times* to get large, workable sample of the DNA.
4. **Genomics -** The study of *large* amounts of genetic information (genomes).
5. **Gel Electrophoresis**
   1. This process is used to create a “DNA fingerprint”.
   2. Take *different* DNA samples and expose them to the *same* restriction enzyme to cut the DNA into *fragments*.
      1. This creates **Restriction Fragment Length Polymorphisms (RFLP’s)**
         1. These are *fragments* of DNA having *different lengths*. (Can you see that in the term?)
   3. Then take the DNA RFLP’s and load them into the agar gel.
   4. Turn on the *electricity*. (Remember, DNA is *negatively* charged because of the phosphate backbone, so it will be repelled on the negative end [Black] and pulled by the positive end [Red].) Electricity *will* *flow* from the Black 🡪 Red strips.
   5. The RFLP’s will *separate* according to *length/size of the fragments*.
      * 1. Big pieces move *slowly* through the gel.
        2. Small pieces move *quickly* through the gel.
   6. Stain the gel with Carolina Blue to see the DNA fragments *within* the gel.
   7. The DNA Bands *create a unique “fingerprint”* of the individuals DNA.
      1. 1 in 70 *Trillion* genetic possibility of *identical* copy. (There are only 7.5 *billion* people on Earth.)
6. **Human Genome Project (HGP)**
   1. The project was begun in 1990 and ended in 2003.
   2. The project mapped out the *entire DNA genome* nucleotide sequence for *all humans as a species*.
   3. It found we have around 40,000 *different* genes in our genome.
   4. These make up *only* about 3% of the total genome.
   5. **Alternative RNA Splicing** is the *key* to making the hundreds of thousands of *different* proteins and enzymes our bodies need or use.
   6. A “project” is now being done for thousands of different species and comparing using **Bioinformatics**.
   7. Genome nucleotide *sequences* can then be *compared* to establish *relatedness* among species.
7. DNA **Nucleotide Sequencing** processes were involved in the above project.
   1. **DNA Microarray Assay** (This process looks *like* a Light-Brite toy.)
      1. Uses *radioactively labeled and colored* nucleotides to create a *visible* sequence on a monitor.
      2. Follow the colored *sequence* to determine the DNA sequence. (For example, red = adenine)
   2. **Dideoxy Chain Termination Method**
      1. Run like a PCR, but has special Dideoxyribonucleotides *added to the mix*. (This *stops* replication.)
      2. The procedure produces *chains of different length* due to termination by dideoxy nucleotide.
      3. Then the fragments are run through a gel and scanned using a laser to identify the dideoxy.
      4. Pieces are then combined together using the Dideoxy hits to “create” the nucleotide sequence.
8. Using a Gel Electrophoresis to create a plasmid map
   1. This allows scientists to *know the genes and their sizes* found on bacterial plasmids.
9. Uses for DNA Technology
   1. Gene Therapy
      1. This uses a virus to introduce a new gene to the cell’s genome in our body’s cells.
      2. Somatic cells vs. Germ cells (Somatic cells only affect you; germ cells affect future generations.)
   2. Pharmaceuticals
      1. Helps with creating new medicines.
      2. Vaccines against diseases and maybe even cancers in the future.
   3. Criminal Forensics
      1. DNA fingerprints of suspects.
      2. Paternity/Maternity testing.
   4. Environmental Clean-up
      1. Bacteria are used to process human sewage in water treatment plants.
      2. Bacteria that can clean up Oil Spills or breakdown Plastic by eating the oil compounds.
      3. Organisms helping clean up heavy metals (such as Mercury) from mining or waste collection.
   5. Agriculture
      1. Having organisms produce more food.
      2. Having organisms produce “larger” food.
      3. Having organisms produce hardier food for easy transport across the world.
      4. Having organisms produce healthier foods.
      5. Having organisms that can produce food during winter. (Winterized)
   6. Livestock
      1. Organisms that are “meatier”.
      2. Organisms that are “leaner”. (Having less fat.)
      3. Organisms that are disease resistant.
10. **Transgenic** Organisms
    1. DNA from two *different* organisms are *combined* to make one organism that *possess traits* from both “parent” organisms. These traits will be passed on through reproduction, using gametes.
11. **Genetically Modified (GM)** Foods
    1. These are foods that have been produced/altered in the DNA.

AP Biology

Genes and Organism Development

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.22, 2.31, 2.32, 2.33, 2.34, 2.36, 2.37, 3.1, 3.5, 3.9, 3.11, 3.20, 3.21, 3.22, 3.23, 4.1, 4.7, 4.17)

Important concepts from previous units:

1. Genes are segments of DNA that are the “blueprints” for making proteins and enzymes within cells.
2. **Zygote** (One 2n cell that is the result of a n sperm fertilizing a n egg.)
   1. This cell will give rise to over two hundred different human cells all with the 100% identical genome within them during development.
3. **Cell Differentiation** (A. K.A. **Specialization**.)
   1. Expressing *different* genes makes cells *different or specialized*in function and shape.
   2. Specialized functions are the products of *“adult”* cells.
4. **Morphogenesis** (“morph” means “body shape”; “genesis” means “creation of”)
   1. The process of morphogenesis is the *product of cell differentiation* occurring during *development*.
   2. **Apoptosis** (Programmed cell death) is a *crucial* part of development too. (For example, apoptosis helps to “create” the spaces between your fingers and toes by “killing off” those cells in the webbing.)
   3. Morphogenesis in *plants***:**
      * 1. The cells of plants *do not move* as they are restricted by the cell wall. They mature *in place* and *respond* to environmental cues.
        2. Plants display *continual growth* until they die. (The growth occurs at the **Apical Meristem**. These tissues are found at the *tips* of roots and stems.)
   4. Morphogenesis in *animals***:**
      * 1. The cells of animals *move* into their final position during development.
        2. Animal display *limited growth*. (They die after a certain number of years.)
5. **Cloning** and **Clones**

# Cloning is the process of making *100%* *genetically identical* organisms called Clones.

* 1. Animal Cloning
     + 1. First step: Remove an *egg cell* from a *female* organism.

1. This cell has all the enzymes and machinery to make development possible.
   * + 1. Second Step: Remove the *Haploid* nucleus from the egg cell.
       2. Third Step: Take a *somatic cell nucleus (Diploid)* out of a somatic cell and put it in the egg cell.
       3. Fourth Step: Put the “egg” cell in a *surrogate organism (female)* to develop until birth.
   1. Ian Wilmut and Dolly (1997) He was the first to develop this process of cloning. Dolly was the name of the first cloned sheep.
2. **Stem cells**
   1. These animal cells are said to be **Pluripotent**. (They can become any type of cell.)(“pluri” means “many”)
      * 1. These cells have *many possabilities* as to what they will develop into as they develop.
   2. They are said to be “embryonic” in development. They also have no genes “locked up”; therefore they can make *any* protein or enzyme.
   3. Origins of stem cells. (Embryonic vs. Adult) Embryonic are found in *developing embryos* and adult stem cells are found within *developed* tissues. The difference is that adult stem cells have undergone a small amount of differentiation and therefore CANNOT make *every* protein/enzyme and therefore are *limited* in what type of cell they can become. Embryonic stem cells have NOT undergone ANY differentiation. They CAN make *every* protein/enzymes.

# Research? Embryonic stem cells are more valuable in research because of the *unlimited* possibilities. They could cure diseases such as Diabetes or SCIDS, repair spinal cord injuries, or be used to grow new organs for transplants.

1. **Pattern** **Formation**
   * 1. DNA information (genes) that *controls* the development of the species’ “Pattern”.
     2. Each species is *unique* to an extent in the “pattern” and DNA sequences that creates it.

# Positional Information

* + 1. The cell “position” is accomplished through *cell-to-cell communication*.
       1. **Where** in relation to the whole organism?
       2. **What** is next to it?

# Maternal Effect Genes (A.K.A. Egg Polarity Genes)

# Controlling the *polarity of the Zygote* helps to determine the Head and Tail or Root and Shoot.

# This “control” is accomplished by production of cytoplasmic determinant proteins and morphogens (Proteins that affect morphogenesis.)

# They will accumulate on *one* side of the zygote cell. This accumulation *determines* the poles of the cell and what each end will start development of in the organism.

# They are referred to as “maternal” because they are produced in the *female egg cell.*

# Segmentation Genes

# These genes produce proteins that influence what will happen in a particular segment of an organism.

# Best examples are insects and crustaceans.

# Homeotic Genes (A.K.A. Hox genes)

# These genes are the *master control* genes for an organism’s development. These are the *most important genes* in any organism… as the control *total development* from start to finish.)

# They contain the Homeobox (A *unique* DNA nucleotide sequence.)

* + - 1. It is a 180 Nucleotide sequence found in **Hox genes**.
      2. Evolution? The *more similar the sequence* between organisms; the *more closely related* in terms of evolution they are. The *more different the sequence*; they *less related* they are.

AP Biology

The Process of Meiosis - Part 1

(Associated Learning Objectives: 1.5, 1.14, 1.15, 1.16, 2.24, 3.1, 3.4, 3.7, 3.8, 3.9, 3.10, 3.11, 3.12, 3.13, 3.27, 3.28, 4.1, 4.17, 4.23, 4.24, 4.25)

**Important** concepts from previous units:

1. DNA is *replicated* in the **S phase** of the cell cycle.
2. **Mitosis** is normal cell division. It involves *one* division. Mitosis means “division of the nucleus”.
3. **Meiosis** makes gametes, sperm and egg. This involves *two* divisions.
4. **Heredity**
   1. This refers to the *transmission of traits* from one generation to the next by *inheriting* DNA from the parent (for asexual reproduction) or parents (for sexual reproduction).
      1. Most DNA is for *common* information. (*Unity* among species and within a species, like blood.)
      2. Some DNA is for *varied* information. (*Diversity* among species and within a species, such as the fur coloration between a zebra and a horse or freckles on one person and no freckles on another.)
5. **Genetics**
   1. This is the science that deals with the *transmission of information* in the form of DNA. It can range from studying how traits are passed from one generation to the next using Punnet squares or identifying DNA segments (what we call genes) and the proteins or enzymes that they make. It is a huge field of science.
   2. This field has had a tremendous impact on society as a whole. Such things as cloning, to new medicines, to making bacteria and yeast making human hormones, to making biological weapons such as Super Anthrax.
   3. Ethics can be involved. (Ethics is looking at the Good vs. Bad in terms of morality.) It is *always* an issue in science, particularly in this field.
6. **Gene**
   1. A *unit of hereditary information* in the form of a DNA *sequence of nucleotides* found on chromosomes.
      1. Most genes code for some type of protein or enzyme. It is the “million dollar blueprint” for making *one* thing. It would be like the blueprint for making a steering wheel.
7. **Genome**
   1. This refers to an organism’s *entire* genetic make-up. *All* the DNA within a cell. It would be like the “blueprint” for making the *whole* functioning car.
   2. Half of the DNA comes from the mother (“half” is represented by “n”); The other half of the DNA comes from the father ( n ). Therefore, a half *plus* a half equals 2 halves which is equal to 1 organism.

**(** n + n ) = 2n. (“half” is also called **haploid** and “two halves” is called **diploid)**

1. **Locus**
   1. The ***location*** of a gene on a chromosome. This is important when you are talking about autosome vs. sex chromosomes.
2. The two types of reproduction that can occur by living organisms:
   1. **Asexual Reproduction**
      1. This involves only *one* parent. The parent is producing *genetic clones* of itself. The parent and offspring are 100% identical in terms of DNA content and DNA nucleotide sequence.
      2. Benefits – Reproduction can occur very *quickly* (Good for taking over a new area).
      3. Risks – Every organism is *the same*. So if a disease affects one; it can affect *all*. (There is NO variation!) This caused the Irish Potato Famine. Potatoes are originally from South America. One species of potato plant was taken to Ireland. This became the only species that the farmers could plant, as no new species were brought over afterwards. A pathogenic fungus, called Potato Blight, began attacking the plants. Since they were all alike in terms of DNA because they were clones, they fungus wiped them out quickly causing the famine to occur.
   2. **Sexual Reproduction**
      1. This involves *two* parents to contribute DNA. This process *“creates” variation*, which is important in terms of *survival*.
      2. Benefits – It produces *variation.* This is why some organisms have *advantages* over others within the same species in terms of survival and *the ability to reproduce*. Variety means there exists the *possibility* to evolve over time while living in an ever changing environment. For example, Wooly Mammoth. Those with *less* hair survived and passed on those genes for *less* hair to their offspring as the environment became *warmer* over time. This lead to the evolution of our modern elephant, which has very little hair. The mammoths with *more* hair died before they could reproduce; thereby “wiping” out those genes and eventually causing the extinction of the *old* species.
      3. Risks – It takes *two* to be *able* to reproduce and they *must* be of the opposite sex for the physical reproduction to occur. This is not good for an *endangered species*. It also takes more *time*. It also involves a *more complicated process to create the gametes* that have half the DNA content.
3. Three possible types of life cycles, *based on DNA content* within cells
   1. Haploid Majority
      1. This type occurs mostly in Fungi and some Protists (These are single celled organisms mainly).
      2. The *majority* of these organisms life time is spent with cells that are *haploid* in terms of DNA content.
      3. They come together to make *diploid* cells to “create” variation to increase survivability in a *changing environment*. After recombining DNA, they undergo **meiosis** to *return* to a haploid state, but with a *different* DNA composition within the cells.
   2. Diploid Majority
      1. This type of life cycle is seen in Animals.
      2. The *majority* of these organisms life time is spent with cells that are *diploid* in terms of DNA content.
      3. The diploid parent organisms make *haploid gametes (sperm or egg)* by undergoing **meiosis**. These cells are made and *stored* in the reproductive organs. They (sperm and egg) come together during sex to bring the DNA content *back to* diploid, so as to be able to make a *new organism*. The diploid cells undergo **mitosis** to keep making new diploid *cells* and thereby the organism.
   3. Alternation of Generations (half-n-half basically)
      1. This type of life cycle is seen in Plants and most Algae. (These Algae are multi-cellular protists.)
4. Human Life Cycle is Diploid Majority
   1. **Somatic** (“soma” means “body”) cells make up most of our body.
      1. These cells possess 46 chromosomes inside them. They are 2n – diploid.
      2. **Karyotypes** will *display* all 46. A **karyotype** is basically *pictures* of the chromosomes.
      3. **Homologous** (“same”) **Chromosomes** can be seen. These are called **Autosomes.** 44 = 22*pairs*exist in all human cells. (If *female*, the two *sex* are the same too… two X chromosomes.)
      4. **Heterologous** (“different”) **Chromosomes** *may* be seen in males. These may be the 2sex chromosomes. (In *males*, there is one X and one Y chromosome.)
         1. Female (XX); Male (XY)
   2. **Germ** (“germ” means “beginning”) cells (A.K.A. **gametes**)
      1. These are the *sex cells*. They are n – haploid. (egg-female; sperm-male)
      2. **Fertilization**, which is the *fusion of egg and sperm together*, must occur to be able to reproduce.
         1. This fusion between egg and sperm produces a *single* *diploid* cell called a **zygote**.
         2. The **zygote** goes on, through repeated *mitosis*, to produce the new organism.

AP Biology

The Process of Meiosis - Part 2

(Associated Learning Objectives: 1.5, 1.14,1.15, 1.16, 2.22, 2.31, 2.32, 3.1, 3.3, 3.8, 3.9, 3.10, 3.11, 3.12, 4.1)

Important concepts from previous units:

1. Evolution is “change over time”.
2. Sexual reproduction involves *haploid* sperm and egg gametes.
3. The DNA within the egg and sperm *will create* the *next* generation organism.
4. **Meiosis** - means “The process of *gamete* formation”
   1. This process occurs in the cells of the *sex* organs of the organism. These organs are called **Gonads.**
   2. This process has *2 divisions* in the process *after*  the S and G2 phases.
      1. Remember , that the S phase *doubles the number of chromosomes*. In humans 46 🡪 92.
      2. **Meiosis I** - This division is the separation of chromosome *pairs***.** In humans, 92 🡪 46
      3. **Meiosis II** - This division is the separation of *sister chromatids***.** In humans, 46 🡪 23
   3. In this process, males produce **4** haploid sperm; each having 23 chromosomes.
   4. In this process, females produce **1** haploid egg with 23 chromosomes. The other three cells *degrade* into structures called **polar bodies** during the process. These can be seen on the *nucleus membrane* in female cells, not males.
   5. Stages to the process of Meiosis
      1. These stages are *very similar* to the stages of Mitosis.
      2. *Three major differences*, from Mitosis, are present to *increase variation.*

(Remember, Mitosis is *normal* cell division. It basically makes *clones* of the adult. *No variation*.)

* + - 1. **Crossover** (“genetic swapping”) occurs in **Prophase I**. (Creates variation.)
      2. Chromosome *pairs* separate in **Anaphase I**. (Creates Variation.)
      3. *Sister Chromatids* separate in **Anaphase II.** (Creates Variation.)

1. **Crossover (**“genetic swapping”) between *homologous* chromosomes.
   1. This creates *variation from the parent’s genome*. They are then called **Recombinant Chromosomes**.
   2. **Synapsis** – Chromosomes that are in a state of being *intertwined together*. (“syn” means “together”; “sis” means “process of”)
   3. **Tetrad** - *Four* chromosomes *twisted together* (“tetra” means “four”… Like the game Tetris has four different shapes.)
   4. **Chiasmata** – Where the chromosomes physically *overlap* making an “x”. (Chi is the Greek letter for X.)
2. Major *differences* between Mitosis and Meiosis:
   1. The *number of divisions* (Mitosis has 1; Meiosis has 2)
   2. The *final products* of each process (Mitosis – “cloned” daughter cells; Meiosis – haploid gametes)
   3. **Crossover**, in Prophase I, creates variation (No crossover in Mitosis)
   4. Chromosome pairs vs. sister chromatids separating in the *second division* to *reduce* DNA to haploid state.
3. *Sources* of variation creation
   1. *Independent* assortment of chromosomes. ( This happens 2x in Anaphase I and II.)

1. 2 = Total number of possibilities (One goes one way; the other the other way in separation.)

* + - 1. n = number of variables; 23 = number needed to make a haploid set in humans.
      2. 2n = 223
    1. For humans the total is about *8 Million possibilities* for *each* parent *with each* division.
    2. 8Million possible outcomes X 2divisions X 2 parents = 4,096,000,000 *possible combinations* for just 46 chromosomes!
  1. Now add in **Crossover** (“genetic swapping”)
     1. Amount of crossing over varies from tetrad to tetrad. If *little* crossover occurs, the offspring looks *very much like* the adult parent. If *lots* of crossover occurs, the offspring looks *very different* from the adult parent.
  2. *Random* fertilization by a sperm. (There are millions released by the male. Which one will make it to the finish line?)
  3. That makes *you* a 1 in 70 *trillion* *possibility* – YOU ARE PRETTY DARN SPECIAL!

1. Evolution? As organisms became *more complex*, a more complex and more *survival oriented* way of reproducing came into existence over *millions of years*. The addition of a *second division* with a couple of slight changes in the *same* four steps (Prophase, Metaphase, Anaphase, and Telophase) creates the variation. The variation helps with survival and this would be beneficial in *changing environments*. Those that survive long enough get to *reproduce* and keep the species going. Those that don’t do not pass on those *defective* traits for surviving in that environment.

AP Biology

Mendelian Genetics – Part 1

(Associated Learning Objectives: 1.16, 2.31, 3.1, 3.2, 3.9, 3.10, 3.11, 3.12, 3.14, 3.18, 3.19, 3.24, 3.26, 4.1, 4.25)

**Important** concepts from previous units:

1. Genes are DNA segments that are *inherited from parents* during reproduction.
2. A gene is the “blueprint” for making a **polypeptide** (protein).
3. Proteins are made (expressed) by the processes of **transcription** and **translation (Protein Synthesis).**
4. Gregor Mendel (1850) He is considered to be the “Father of Genetics”.
   1. He was a monk who worked with pea plants, this is because he was the cook too.
5. **Character -** An *inheritable physical feature*. (This is a *characteristic* such as eye *color* or hair *color*.)
6. **Trait -** This is a *variation* of a character. (Such as *blue* colored eyes or ***black*** colored hair.)
   1. This *requires* inheriting *two* alleles; one from each parent.
7. **Alleles**
   1. This term refers to *different versions of a gene*. (Remember, a gene is a distinct *DNA nucleotide sequence* that can make one protein or enzyme. (Brown, blue, green eye color. These are three different *versions* or DNA *sequences* of a *single gene*, but they *all* are making the eye *color*.)
   2. Each trait needs *two* alleles. One from each parent to be made or “expressed”.
   3. **Dominant** alleles are given *capital letters*. (These are like books or recipe cards *with information* in them.)
   4. **Recessive** alleles are given *lower case letters*. (These are like books or recipe cards *with blank pages* – no information is on them on *how* to make the protein or enzyme. The “blueprint” is missing.)
8. **True** “pure” **breed** - These organisms *only* have one *type* of **alleles** for that trait.
   1. A.K.A. Homozygous alleles. ( “Homo” means “same”) Such as BB or bb.
9. **Hybridization** - This is the process of “creating” an organism with two *different* types of **alleles** for that trait.
   1. Referred to as **Hybrid** or **Heterozygous** alleles. (“hybrid” and “hetero” mean “different”) Such as Bb.
10. **Phenotype** (“pheno” means “physical”)
    1. This term refers to a *physical trait* that can be “seen”. (Blue eyes or Type A blood, would be examples.)
11. **Genotype** (“geno” means “genetic”)
    1. This term refers to an organism’s *genetic (DNA)* *make-up for a trait*. (Such as BB, Bb, and bb.)
    2. If the **genotype** of an organism is *unknown*, we can perform a **test cross** to *find* it.
       1. Toperform this test, we*must* use a **homozygous** **recessive** to *mate* with our unknown.
          1. This allows for no information to be “covered up” by a*known* dominate allele.
12. **Punnett Square -** a chart showing the *possible genotypic outcomes* for a mating based on parent’s **genotypes**.
    1. **Monohybid** – This chart displays *one trait*. (It has 4squares.) ( 41 = 4 squares)
    2. **Dihybrid** – This chart displays *two traits*. (It has 16 squares.) (42 = 16 squares)
    3. **Trihybrid** – This chart displays *three traits*. (It has 64 squares.) (43 = 64 squares)
13. Mendel’s **Law of** **Segregation** (Segregate means “to separate”)
    1. The *homologous*chromosomes or **sister chromatids** can move *independently* of one another.
    2. This is *supported* by what happens during Anaphase I and II of meiosis.
14. Mendel’s **Law of** **Independent Assortment** - This basically states that *variations* are possible on *sets of*

*chromosomes.* (Assortment means “variety exists”)

* 1. This states that chromosomes line up *independently* of one another on the *midplane* of a cell.
  2. This occurs at Metaphase I and II in Meiosis.

1. **Probability** “Chance”
   1. This refers to the *likelihood* of a *certain* outcome actually happening. (What are the chances of…occurring?)
   2. **Probability** ranges on a scale between 0 and 1.00. (From 0% to 100% essentially.) 0.5 is 50% and so on.
2. Quick way to assess **probability** in a mating cross (Use the worksheet provided by your teacher)
   1. Uses the **Rule of Multiplication**. This applies to the *parents*genetics coming together to make an *offspring.*
   2. A calculation to *determine* the outcome for *a specific* genotype combination in the offspring.

AP Biology

Mendelian Genetics – Part 2

(Associated Learning Objectives: 1.5, 1.16, 2.22, 2.23, 3.1, 3.13, 3.14, 3.15, 3.16, 3.17, 3.18, 3.19, 3.22, 3.24, 3.26, 4.17, 4.23, 4.24, 4.25)

**Important** concepts from previous units:

1. Phenotypes occur from proteins or enzymes and they are the result of genes being “expressed” within cells.
2. A genes nucleotide sequence *determines* the **codons** that are used to construct proteins by the ribosomes.
3. **Incomplete Dominance**
   1. This is where the genetic information is *“blended”* together. (For example, Red+ white = Pink). Neither phenotype is *completely* dominating the other. They are*both seen* in a “blended” version.
4. **Complete Dominance**
   1. This is where the *dominate allele* has DNA nucleotide information for a *fully functioning* protein or enzyme and it is “suppressing” the *recessive* allele DNA nucleotide sequence.
5. **Codominance**
   1. This is where *both* alleles are seen but they are*“not blended”* together. They are both *equally* present in terms of phenotype.
6. **Multiple Alleles** - This is where there are *multiple* different versions of the *same basic allele*.
   1. The glycoprotein “hands” of red blood cells would be a great example of this. These “hands” identify the blood types. One type of hand is “A”. Another is “B”. Another is **codominance** “AB”. Then there is the homozygous **recessive** “O”. Since it is recessive, “no blueprint information” was *in* the DNA on how to make the glycoprotein hands “A’ or “B”.
   2. **Hemophylactic Shock** – This occurs when someone is given the wrong blood type. (The “hands” don’t match. So the white blood cells begin killing the new red blood cells.)
   3. **Universal Donor** – Can *give* blood to anyone (Blood Type O –. There cells have NO hands. So they match everyone.)
   4. **Universal Recipient** – Can *receive* blood from anyone. (Blood Type AB. They have BOTH types of hands. So they can match everyone.)
7. **Pleiotropy -** This is where one gene *affects multiple phenotypes*. ( “Pleio” means “multiple”)
   1. Sickle Cell Disease is a great example. This gene *affects* the red blood cells shape, Oxygen carrying ability, Malaria resistance, etc..
8. **Epistasis** - A gene at *one locus* affects a gene at a *second locus***.**
   1. Hair is a great example. Several genes are interacting to “create” hair’s phenotypes – These are: color, shape, thickness, texture. (“epi” means “source”; “stasis” means location) The source is affecting another location.
   2. This usually has a 9:3:4 ratio; not the *normal* 9:3:3:1 ratio as seen with most dihybrids.
9. **Polygenic Inheritance** - This is where there exist *many different degrees* of phenotypic outcomes.
   1. This is due to **Quantitative Characters**. (Quantity -*how many* alleles did you *inherit* from your parents.)
   2. “poly” means “many”; “ genie” refers to “genes”; “inheritance” from your parents
   3. Skin Color is a great example. We have many *different degrees of skin pigmentation* ranging from Albino 🡪 Black, Black. It *depends on how many* *copies of the same gene* for making the skin pigment melanin you inherited from your parents in the sperm and egg.
   4. **Norm of the Reaction** – This refers to where the *majority* of organisms fall on the bell curve for that trait.
      1. Evolution? The norm can tell you about the *type of environment* organisms live in. The norm is because some trait is *beneficial* in that environment.
10. **Multifactorial** 
    1. Many *environmental factors* are affecting the phenotypic display of genes in that organism.
    2. This gives fuel to the argument over Nature vs. Nurture in organisms. (The genetics vs. the environment.)
       1. While there are equally legitimate arguments for both sides. The overwhelming evidence supports a 50/50 reality. 50% of our behavior is innate (in our genetics); after all who teaches a dog to bark. The other 50% comes from our experiences or living environment; such as eating good foods affects the body you “create” over time.
       2. Height, intelligence, and weight can all be considered multifactorial.

AP Biology

Mendelian Genetics – Part 3

(Associated Learning Objectives: 3.1, 3.2, 3.9, 3.11, 3.12, 3.13, 3.14, 3.19, 4.23, 4.24)

1. **Pedigree**
   1. This is a *family history* of trait occurrence in chart form. *Affected* individuals are *shaded in* on the chart.
   2. These help tell the past occurrence and can be useful in *predicting the future occurrence by mating.*
2. **Recessive** Disorders “ no information inherited on *both* chromosomes”
   1. These disorders tend to be very harmful to the organism.
   2. They *only* occur in the *homozygous recessive* *genotype*.
      1. There is nothing to be dominated by, so the disorder is present.
   3. **Carriers** – These are organisms that are *heterozygous* *in genotype*. They are 50/50 in terms of *passing on* the trait. It depends on which allele was present in the *gamete* that was involved in making the offspring. These organisms usually appear *normal* for the trait as the possess one dominant allele.
   4. Human recessive disorders:
      1. **Cystic Fibrosis** (Also referred to as “CF’.)
         1. This is the most common lethal genetic disease.
         2. This disorder affects 1 in 2,500 births.
         3. In Caucasians, 1 in 25 people is a carrier for the disorder.
         4. The disorder creates a faulty Chloride ion (Cl-) protein carrier on cell membranes in the lungs. This causes fluid (water0 to build up in the lung tissues.
            1. People drown in their own fluid.
            2. They are also prone to get multiple infections in the lungs.
         5. Treatment? Since it is *genetic*there is NO cure. Patients have to get the fluid drained from the lungs periodically for their *entire* life. There are medicines to help reduce the number of times this has to occur.
      2. **Tay-Sachs Disease**
         1. This disorder creates a *non-functional lysosome in brain cells*. Brain cells need massive amounts of energy to function properly; therefore, they feed upon *lipids* primarily. The lysomomes break them down using beta oxidation for use in cellular respiration. The lysosomes associated with this disorder are missing an enzyme to be able to do this; so they just fill up with lipids. The cells fill with lipids and then die.
         2. This disorder mainly affects the Jewish Culture because of marrying within the culture. The Jewish culture has a high percentage of carriers.
         3. The children affected, usually die a painful, blind death by age 5.
      3. **Sickle-cell Disease**
         1. This disorder is the most common genetic disorder within the black population. Other populations *can get it* too. It is not exclusive.
         2. It affects 1 in 400 births.
         3. The *6th Amino Acid* is changed (Glutein 🡪 Valine) in the *primary sequence* of one of the proteins needed to make red blood cells. (The easy way to remember this is: 666 is the number of the beast. 6 is the amino acid that changed to create this horrible disease. It went from **g**ood [**g**lutein] to **v**ery bad [**v**aline].)
         4. **Sickle- cell trait** (“trait” is used to refer to individuals that are *carriers*.)
            1. These individuals have *resistance to Malaria* because of the one recessive allele they possess but mainly have normal red blood cells for carrying oxygen.
            2. This is referred to as the **Heterozygous Advantage.** They have an *advantage* over individuals that are homozygous dominant or homozygous recessive. Homozygous dominant are NOT resistant to Malaria. Homozygous recessive are also resistant to Malaria; BUT they have the disease to contend with.
            3. Sickle – cell identification of carriers in individuals is *important to avoid* this disorder from occurring.
         5. These sickle shaped cells have *reduced oxygen carrying ability*. They also are painful when the points of the cell jab into the walls of the blood vessels.
         6. Treatment? There is no cure as it is genetic. Some medicines help with the pain or low oxygen levels.
3. **Dominant** Disorders
   1. *Only* need*one dominant* allele for these disorders to be present or “expressed”.
   2. If an individual is homozygous dominate, then the disease is much worse and usually fatal.
   3. Human Dominate Disorders:
      1. **Achondroplasia** - This is referred to as *Genetic Dwarfism*.
         1. This disorder affects 1 in 10,000 births.
         2. Most people are homozygous recessive and there for much taller than these individuals.
      2. **Huntingdon’s Disease**
         1. This disorder affects 1 in 10,000 births.
         2. It has a late life onset – usually in the 40-50 age range. Usually *after* children are born.
         3. The dominate gene has a locus on tip of Autosome 4.
         4. Family history is important in diagnosis of this disorder. Pedigree can help diagnose.
         5. It is a *slow degenerative* disorder affecting the brain that is almost always fatal.
4. **Multifactorial** Diseases (“Factorial” refers to “the environment”)
   1. Heart Disease - Genetics, diet, alcohol, and smoking are all factors that *contribute* to the development.
   2. Diabetes - Genetic, diet, and race are all factors that *contribute* to the development of the disease.
   3. Cancer - Genetics, life style habits in general are all factors that *contribute* to development of the disease.
   4. Alcoholism - Genetics, lifestyle, and mental state are all factors that *contribute* to development.
5. **Penetrance –** The proportion (percentage) of individuals in a group with a given genotype that *actually* show the expected phenotype.
   1. For example, Huntington’s Disease – 95% of the people who get the dominant allele *actually* express the disease. 5% do not express. So we say there is 95% penetrance for that *allele*.
6. **Expressivity** – the *degree* to which a genotype is expressed in an *individual*. Tends to be more severe in homozygous states, where there are TWO copies of a gene.

AP Biology

Mendelian Genetics – Part 4

(Associated Learning Objectives: 3.1, 3.2, 3.9, 3.11, 3.12, 3.13, 3.15, 3.16, 3.17, 3.19, 3.24)

**Important** concepts from previous units:

1. Genes are located on chromosomes.
2. There are two types chromosomes associated humans – **autosomes** and **sex chromosomes.**
3. Chromosomes are*inherited* from the parents.
4. **Linked Genes** 
   1. These are usually inherited as a *linked* unit because they are found on the *same chromosome*.
   2. This term usually is associated with genes on *Autosomes (1-22)*.
5. Alfred Sturtevant
   1. He was the pioneer of **genetic mapping** – locating the *loci of genes*.
   2. He used *crossover rates* to determine the loci on chromosomes.
      1. The finished product is called a **Linkage Map.**
      2. The *smaller the rate*; the *closer* they are to each other on the *same chromosome*.
      3. The *higher the rate*; the *farther apart* they are from each other on the *same chromosome*.
      4. The loci are measured in **Centimorgans** or **map units**.
6. **Sex-Linked Genes** 
   1. This term refers to genes found on the *sex* chromosomes; 95% of the time it mainly refers to the X chromosome. (Think X when it is seX linked.)
      1. This is because *both sexes* have at least one X chromosome in their genome.
      2. XX (Female and homologous) ; XY (Male and heterologous)
   2. Sex chromosomes undergo *very little crossover* during Prophase I of Meiosis.
   3. Sex of the organism will be *determined* at conception. This is when egg is fertilized by the sperm. You *will* either get a sperm containing an X chromosome or a sperm containing a Y chromosome.
   4. Everyone starts out female. (This is why we all have nipples.)
      1. At about two months of age in the womb, the Y chromosome’s **SRY gene** goes *active* to make testosterone, from estrogen, to finish development of the male. (Remember, **functional groups**.)
      2. *After* development is complete, testosterone production is turned off until puberty. At puberty it is turned back on so as to make the *secondary* sexual characteristics, such as facial hair.
   5. *Patterns*of Inheritance and some Human **Sex-Linked** Genetic Disorders: (NO cure exists, because the problem is in the DNA.)
      1. **Color Blindness**
         1. This is the result of a faulty gene *(recessive)* on the X chromosome for making a particular type of *light wavelength (color) absorbing protein* in cones of the retina of the eye.
         2. The most common type is Red/Green Colorblindness. (Red and Green appear gray.)
      2. **Hemophilia** (Means “love of bleeding”)
         1. These individuals CANNOTmake *(recessive)* **Anti-hemolytic Factor**. (**AHF** for short.)
         2. They may experience problems with possible bleeding to death.
         3. This was a disorder associated with the “Royal Blue-Bloods of Europe” – They were *inbreeding* to keep the crown “In the Family”.
         4. Treatment? These individuals have to keep AHF with them at all times in case they get hurt. If they do get hurt and start to bleed, they will *require* a shot of AHF to stop the bleeding. Even a bruise (bleeding under the skin) can possibly lead to death.
      3. **THE PATTERN ON A PEDIGREE**: It will *appear to mainly affect males* (as they only have *one* X chromosome). This is because if the inherited X chromosome has a recessive gene on it; it will NOT be covered up by a dominant one on another X chromosome (as is the case in most females). Females *can still get these disorders*, but they must inherit *two* recessive X chromosomes. The females tend to be **carriers**, so they appear unaffected. So they tend to pass the recessive X on to their sons. The son will be a *sufferer*, if he gets the recessive X, of the disorder. It *appears to skip* a generation, because the mother is a carrier and the sons are showing the disorder.
7. Genes associated with these two terms *do not* follow Mendel’s Laws of Inheritance and normal ratios.
   1. This is because these terms are mostly referencing *one* chromosome and not inherited *pairs* of chromosomes.
   2. Variation on linked chromosomes is associated with crossover frequency with its homologous mate.
   3. Sex –linked is referencing the X chromosome only. Males have 1 and females two *copies*.

AP Biology

Mendelian Genetics – Part 5

(Associated Learning Objectives: 1.14, 1.15, 1.16, 2.31, 2.32, 2.33, 3.1, 3.9, 3.11, 3.12, 3.13, 3.17, 4.1)

Important concepts from previous units:

1. There are also *gene* errors – point mutations and reading frame mutations.
2. **Chromosomal Errors** than can occur:
   1. These could occur during Mitosis *or* Meiosis.
      1. They would occur during the *Anaphase* Stages where *chromosomes are moving.*
      2. They could also occur during *Crossover* where *gene DNA segments are moving.*
   2. Two *types* of errors can occur:
      1. **Chromosomal Number** (**Aneuploidy** means “Abnormal *number* of chromosomes”)
         1. This is the result of **non-disjunction**. “Failure to separate**”** during Anaphase.
         2. **Trisomic** - *Three* of 1 kind of chromosome.)
         3. **Monosomic** - *Missing one***,** the other half of the pair. (It is located in the Trisomic gamete.)
         4. **Polyploidy** - Many *extra sets* *of chromosomes*.

i. **3n (triploid)** - Three “halves” are in this cell.

ii. **4n (tetraploid)** - Four “halves” are in this cell.

iii. Deadly in most animals; Plants not really affected because they are simple organisms.

* + 1. **Individual Chromosome Structure**
       - 1. These occur because *of faulty crossover*
         2. **Deletion** – Chromosome segment is “missing”. It got stuck *on the other* homologous chromosome during crossover.
         3. **Duplication** – A chromosome segment was “copied” twice. Two genes on one chromosome. It is “missing” from the other homologous chromosome.
         4. **Inversion** – A chromosomal segment is “backwards”. It was *inverted* (“backwards”) during crossover.
         5. **Translocation** – A chromosomal segment is attached to *a different autosome***.** It accidentally broke loose and ended up on another chromosome.

1. **Syndrome**
   1. This term refers to an organism “possessing” the *identifying traits* of a particular genetic disorder.
   2. Human Genetic Disorders due to two *abnormal chromosomal number*:
      1. **Down’s Syndrome** 
         * 1. This affects about 1 in 700 births.
           2. This individuals possess an *extra 21 Autosome* (A.K.A. **Trisomy 21**)
           3. General syndrome features – mental retardation, flat face, squinted eyes, small.
           4. Mainly the result of women of *advanced age* having babies.
2. **Extranuclear DNA** (“extra” means “outside of”)
   1. DNA that is located *outside the nucleus* in organelles. (These are Mitochondria, Chloroplasts, and Plastids.)
   2. Mitochondria inherited from the *mother inside the egg*. They are exactly alike. (Helps in Criminal Forensics cases trying to establish relationship between individuals.)
   3. **Mitochondrial Myopathy** - These mitochondria *lack the ability* to make large quantities of ATP because they posses faulty DNA for making some of the enzymes or proton pump proteins in the cellular respiration process.)
      1. General Characteristics: These individuals are very tired *all* the time.