# Genetics: BIO-2306

Final Review For Genetics Covering Biological Concepts

(*Cumulative practice problems may be found on the final page!*)

**This week is the last week of class, and typically in this week (and the surrounding weeks of class) you are reviewing for the final exam. Please use the review below (as well as other resources from the semester) as a general overview of some of the key concepts taught in the course!** Please take a look at all 12 weekly resources listed on our website, as well as the math content review, to help you review for the final exam!

If you have any questions about these study guides, the final schedule of group tutoring sessions, private 30 minute tutoring appointments, the Baylor Tutoring YouTube channel or any tutoring services we offer, please visit our website www.baylor.edu/tutoring or call our drop in center during open business hours. M-Th 9am-8pm on class days 254-710-4135. **The last day of tutoring in the drop in center will be the last day of class.** To learn about additional resources available during Finals Week, please visit CASE in the West Wing basement of Sid Rich! Good luck on your final exam!

## Section 1: Review of Conceptual Genetics

**Chromosomes:** **chromosomes** are bundles of DNA wrapped around proteins

**Sister Chromatid:** 1 chromosome composed of 2 DNA strands joined at the centromere by *cohesin* proteins

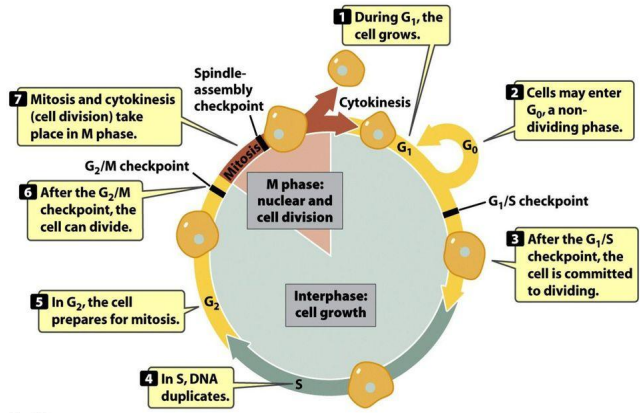
**Locus:** the specific point on a chromosome where a gene is located

**Eukaryotes vs. Prokaryotes:**

https://www.youtube.com/watch?v=RQ-SMCmWB1s

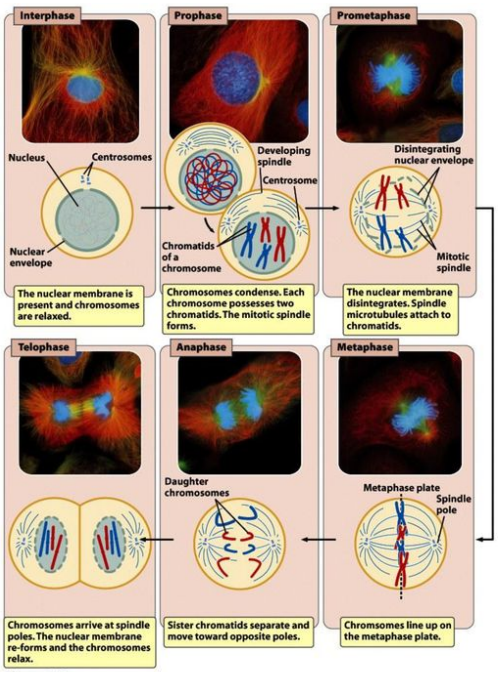
**Cell Cycle:** the cycle of cellular growth and division

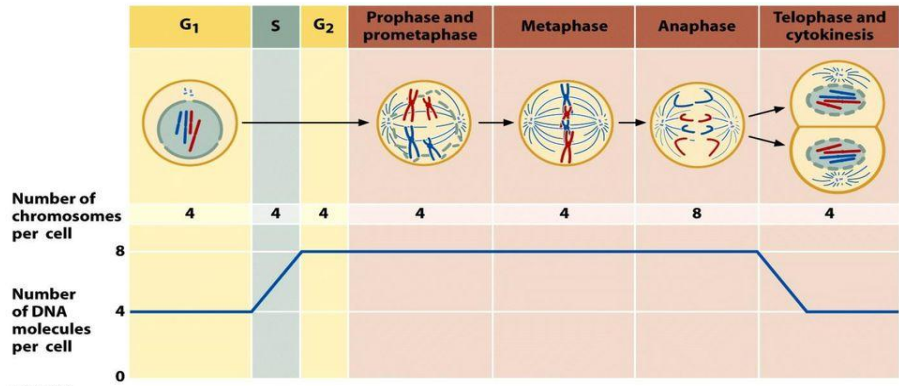
**Interphase:** the part of the cell cycle dedicated to growth/repair, metabolism, and DNA replication




**M-Phase:** division of the nucleus

**Mitosis:** the division of a parent cell into two *identical* daughter cells (2n → 2n) → Equational division




*All diagrams, tables and figures are the property of Benjamin A. Pierce; Genetics: A Conceptual Approach*

**Meiosis:** The 2 divisions of a single diploid parent cell to 4 genetically *different* haploid daughters (2n→ n)

**Sources of Variation:**

**Random Alignment** of homologs in metaphase 1

**Crossing Over** of homologs in prophase 1 (**chiatisma**)

**Meiosis 1:** reductional division→ separates homologous pairs (2n → n)

***Shugoshin*** prevents separase from lysing cohesins in sister chromatids

**Meiosis 2:** equational division→ divides chromatids as in mitosis (n → n)

**Mendelian Inheritance:** the general pattern of heredity discovered by Gregor Mendel

**Law of Segregation:** each individual has 2 copies of an allele which code for a trait; these two alleles are separated (**Anaphase 1**) of gamete formation

**Law of Independent Assortment:** in a cross involving more than two genes, the alleles segregate independently of each other (*unless they are* ***linked***)

**Chromosomal Sex Determination:** generally, most studied organisms display the **X-Y** system for sex determination, though several others exist

**Sex Linked Gene:** a gene located on a sex chromosome

**X-Linked:** mother to child **or** father to child (dominant or recessive)

**Y-Linked:** father to son **only**

**Hemizygous:** since males only carry one copy of the **X**-chromosome (or the **Y**), they are considered hemizygous (*single* allele carriers)

**Genomic Imprinting:** males and females have different patterns of methylation; for certain genes or structural mutations, whether they are inherited from the mother or father will determine the phenotype of the offspring.

**Lyon Hypothesis:** in all individuals with more than 1 X-chromosome, all but 1 will be *inactivated* (at random) → **Barr Body:** the remnant of an *inactivated* X chromosome

***note:** some sex determining genes are not inactivated, so the 'feminizing' effect depends on X-chromosome dosage *and* whether or not there is an **SRY** gene

**Chapter 6: Pedigrees** (watch this short video!)

https://www.youtube.com/watch?v=Gd09V2AkZv4

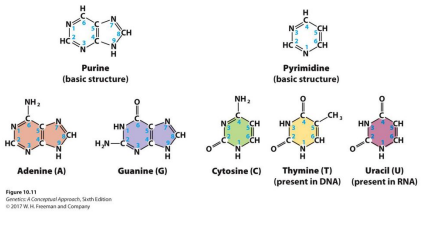
## **Section 2:** DNA and Chromosomal Structure and Discovery

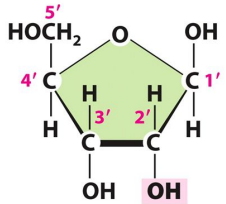
**Deoxyribonucleic Acid: DNA**

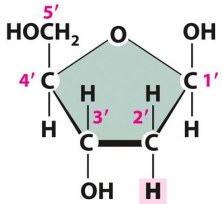
**Ribonucleic Acid: RNA**

**Nucleic Acid Structure** →

**Chargaff's Rules:** the proportion of A&T and G&C are ***equivalent*** in **DNA** and the total proportions add up to 100%

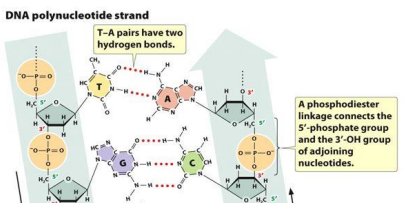



**Griffith's Experiment:**





**Transforming Principle:** some "transforming substance" had to have caused the change from the non-virulent to virulent *S. pneumoniae*… we now know this is **DNA**

**Avery, MacLeod and McCarty Experiment:** proved that DNA is the "transforming substance"; Used a modified version of Griffith's experiment where digestive enzymes were applied to transformed bacteria

**Watson and Crick's Discovery of DNA's 3D Structure:** Watson, Crick and Franklin discovered DNA's structure in 1953

**Chromosomal Structure:**

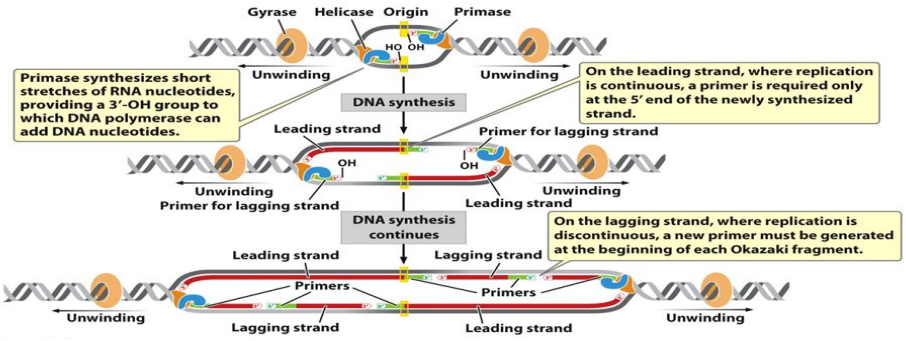
- **Chromatin:** the complex of DNA and proteins
  - ***DNase* Hypersensitive Site:** sites where DNA is less tightly bound
- **Histones:** proteins which associate with DNA (only in eukaryotes and some archaea)
  - **Five Types:** H1, $H2_A$, $H2_B$, H2, H4
  - **Nucleosome:** A DNA-histone complex which DNA wraps around (~146bp)
    - **Core Nucleosome:** an octamer (2 sets of) $H2_A$, $H2_B$, H3, H4
    - **H1 + Linker DNA:** H1 holds the DNA in place on the nucleosome and linker DNA (~50 bp) joins adjacent nucleosomes
  - **Histones** generally tend to express (+) charged residues (**Lys**, **Arg**) to attract the (-) charged phosphate backbone of DNA → adding methyl or acetyl groups decreases affinity of DNA for a histone

## **Section 3:** The Central Dogma

**Meselson-Stahl Experiment:** Proved DNA replication is *semiconservative*

**Stages of Replication:**

- **Initiation**
- **Unwinding**
- **Elongation**




**Enzymes:**

| Eukaryotic | Prokaryotic |
|---|---|
| ***DNA-pol α:*** has primase activity; creates RNA primer followed by a short stretch of DNA; ***DNA-pol δ:*** completes replication of the lagging strand; ***DNA-pol ε:*** replicates the leading strand; ***DNA ligase*:** joins the *Okazaki fragments* | ***DNA primase*** binds to helicase and forms RNA primers; ***DNA-pol I*** replaces RNA with DNA nucleotides (special exonuclease 5'-->3'); ***DNA-pol III*** catalyzes the addition of dNTPs to the growing strands of new DNA |

## **Transcription** (DNA → RNA)

### Initiation

1. Promoter Recognition:
    a. The **core enzyme** of ***RNA-pol*** binds to the **σ factor** to form the ***RNA-pol holoenzyme***. This allows the polymerase to bind
2. Formation of **transcription bubble**: ***RNA-pol holoenzyme*** begins to unravel DNA
3. Synthesize first bonds between rNTPs (**note:** the first nucleotide keeps all *3 phosphates*)
4. Escape of Transcription apparatus from promoter: ***RNA-polymerase*** undergoes a change in shape that causes it to *release* **σ** and 'escape' the promoter to move downstream

### Elongation

***RNA-pol*** acts as a helicase to unwind **downstream** DNA and rewind **upstream** DNA; it also adds rNTPs complementary to the **template/non-coding** strand **5' → 3'**

### Termination (once *RNA-pol* reaches the terminator)

**Rho-Dependent Termination:** a protein (***rho***) causes termination

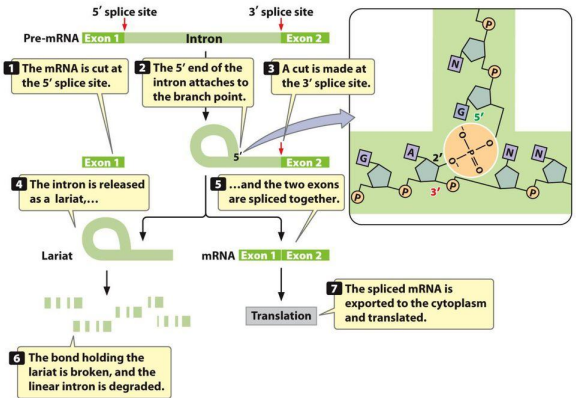
**Rho-Independent Termination:** **inverted repeats** and/or **poly-uracil stretches**

**RNA Processing:** prevents degradation of mRNA and aids in

**Addition of 5' Cap**

**3'-Cleavage and Polyadenylation**

**Splicing:** see diagram (***right***) snRNP = 1 snRNA + proteins → 5 snRNPs make up a ***spliceosome***

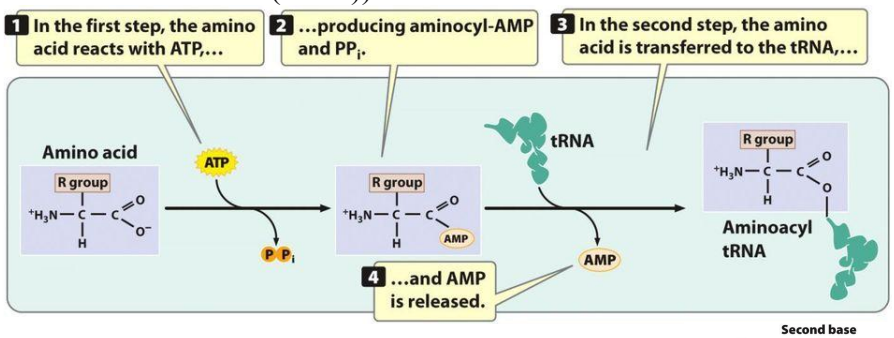


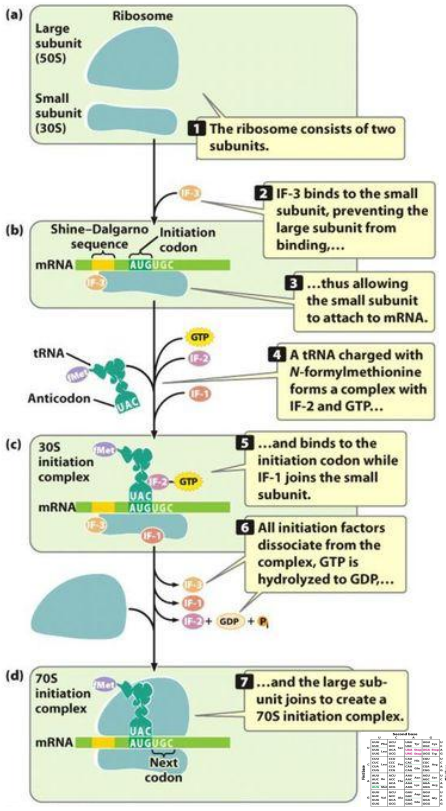

**Note:** *RNA processing* may occur in Euk's or Prok's, but **spliceosomal processing** will *only* occur in eukaryotes.

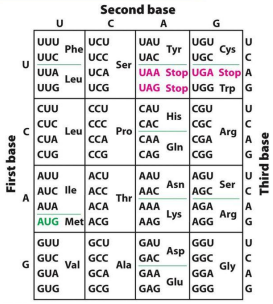
## **Translation** (RNA → Protein)

**Translation:** RNA is copied in the 5' → 3' direction to a protein in the $N_{term}$ → $C_{term}$ direction

**Codons:** units of 3 nucleotides (5'→3') which complimentary bind to a *tRNA* molecule corresponding to an amino acid (Review wobble rules (ch 15))




## **Section 4:** Gene Regulation

### Operons

**Negative Inducible:** the regulator protein is translated in an inactive form, and then is allosterically activated

**Inducer:** molecule that binds to the allosteric site of the repressor, rendering it unable to bind to the operator [allosteric inhibition] (ex. lactose: *Lac operon*)

**NegativeRepressible:** the regulator protein active, then is allosterically inactivated

**Corepressor:** molecule that binds to the allosteric site of the repressor and activates it [allosteric activation] (ex. tryptophan: *Trp operon*)

***Lac* Operons:** negative inducible operon

Prokaryotes need simple sugars to metabolize (create ATP/survive). When **lactose** (the *substrate* of the product of the *lac Z* gene) is cleaved by *β*-Gal, we produce glucose and galactose. The ***lac* operon** codes for genes that help lactose enter a cell and be cleaved

https://www.youtube.com/watch?v=EjRXz1xAdow

**Chromatin Remodeling:** Pushing histones out of the way in order to allow transcription machinery to bind or chemical modification → **EUKARYOTES**

| Acetylation of Histones: | Histone Methylation: | DNA Methylation: |
|---|---|---|
| Neutralizes positive charge on histone side chains (lys and arg); DNA is less tightly wound (***Acetyltransferase:*** add; induction/***Deacetylase:*** remove; repression) | Can either repress of induce transcription( ***Methyltransferase:*** add/***Demethylase:*** remove) | DNA methylation represses transcription because it attracts deacetylase enzymes (DNA to wraps more tightly) **CpG islands:** consensus sequences for methylation near promoters (**cytosines** are methylated) |

**Eukaryotic Initiation:** rate is *highly* regulated by the interaction between TAPs and repressor proteins which act like a foot on and off the accelerator for the rate of *basal transcription apparatus* (**BTA** aka *holoenzyme*) assembly at the **Core Promoter**.

**Gene Regulation at the Chromatin Level:** Tightly wound DNA around histones prevents transcription

- ***DNase-I* Hypersensitive Sites:** **Tightly** packages area around histones were not broken down by *DNase*, so they could not be easily transcribed
  - **Less tightly** compacted regions are more open, more readily transcribed, but are also more readily broken down by *DNase*

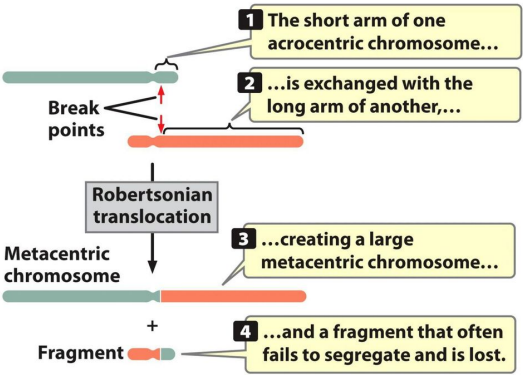
**Epigenetics:** phenotypic differences transmitted without genetic variation due to structural variation of chromatin (**environmental** impact on gene expression) *see ch. 17 and 21*!

## **Section 5:** Mutation and Cancer

**Chromosomal Mutation:** changes that vary the number and/or structure of chromosomes within an individual

**Aneuploidy:** change in the number of *individual* chromosomes (**Robertsonian Translocations or Nondisjunction**)

- **Down Syndrome:** trisomy 21; developmental and physical delays: https://www.youtube.com/watch?v=eruPJS_guNE
  - **Primary:** caused by nondisjunction in **Anaphase II** (2n+1 = 47)
  - **Familial:** caused by a *robertsonian translocation* between chromosomes 14 and 21 (2n = 46)




**Cancer:** cells unable to respond to normal controls to cell division which proliferate (divide) indefinitely

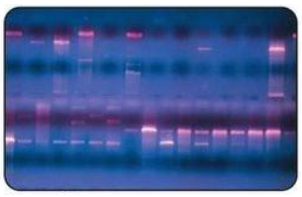
**Clonal Evolution:** mutations which increase the ability of a tumor to survive and reproduce will be 'selected for' in a growing tumor as it moves towards malignancy.

https://www.youtube.com/watch?v=UopUxkeC4Ls

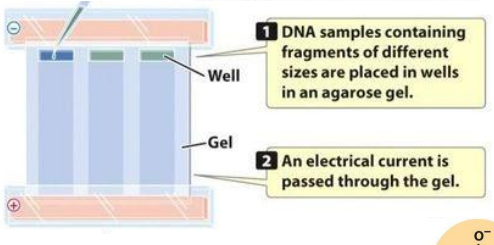
# **Section 6:** Other Topics in Genetics

**Gel Electrophoresis (GE):** Separation of DNA due to its mass (ie molecular weight)

DNA moves down an electrophoresis gel due to its net negatively charged backbone



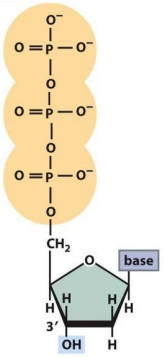
**Gel:** highly porous agarose gel allows DNA to pass through. The largest pieces will travel the furthest and the **smallest pieces** will travel the **furthest** and the **largest fragments** will travel the ***least far***. DNA is dyed to visualize under UV light **(above, right)**




**Cathode (-):** the negatively charged pole will repel the DNA towards the *anode*

**Anode (+):** the negatively charged DNA will be attracted to the positive charge

**Polymerase Chain Reaction (PCR):** DNA amplification using thermocycling (cycles of changing temperatures)




**'Raw Materials':** *buffer solution* (KCl or $MgCl_2$), *Taq Polymerase*, dNTPs, Template DNA, forward and reverse primer (ie free 3'-OH group)

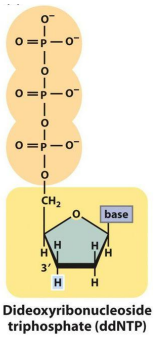
**Reaction:** a process repeated ~20-40 times to amplify DNA exponentially

**Denaturation** (~2min @95°C)**:** separates (*denatures*) DNA strands at high heat

**Annealing** (~1min @60°C)**:** primers bind (*anneal*) to the ssDNA templates

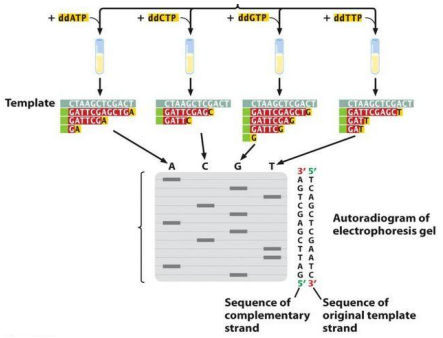
**Elongation** (~1min @72°C)**:** *Taq Pol.* adds dNTPs to ssDNA template

**DNA Sequencing:** determining the primary (nucleic acid) sequence of a DNA molecule




**Sanger 'Di-deoxy' Sequencing:** reaction is <u>similar to PCR</u>, but uses 4 separate containers with one of the four types of *di-deoxy nucleoside triphosphates* (**ddNTPs**) in addition to dNTPs. This gives the sequence *<u>complementary</u>* to each DNA strand

These lack a 3'-OH group, so they **terminate** DNA replication

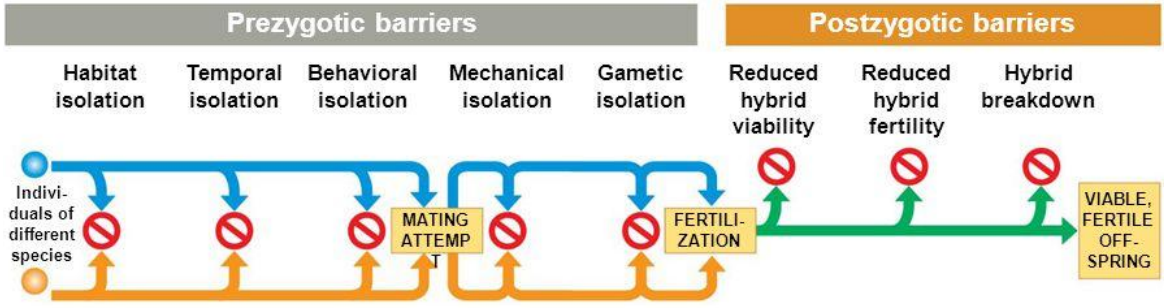



Each of the four reactions (**ddATP**, **ddGTP**, **ddTTP**, and **ddCTP**) are placed into separate gels and run in electrophoresis → each ddNTPs has a fluorescent tag

The *shortest* molecules travel the <u>furthest</u>, so the DNA sequence can be determined by looking at band position from the bottom up [to the wells].

**Biological species concept:** A group of organisms which can interbreed successfully with one another, but are ***reproductively isolated*** by members of other species

**Reproductive isolation:**



## Practice Questions From the Whole Course:

1. Click this link to view the practice problems: https://docs.google.com/document/d/13yQZ0q78hm8ORlilg22ZpWfVuq8dSWo6kEbq9aGfMCk/edit?usp=sharing

**THANK YOU for using these resources this semester! Best wishes on your final exam!**