Exam 2 study guide:

You may need a calculator for this exam. A simple scientific calculator will do. You MAY NOT use a cellphone, tablet or laptop as a calculator. It must be a regular, separate, calculator (graphing calculators are ok if that's all you have). Please do not forget your calculator.

Vocabulary, vocabulary, vocabulary!!! Do you know what I mean if I ask you to list the phenotypes from a F1 testcross? Do you know what to look for if you are asked to examine an autosomal recessive pedigree for evidence of linkage? Dominant vs. recessive? Homozygous vs. heterozygous? Genotype vs haplotype? Haplosufficient vs haploinsufficient? Gametic vs. Somatic? Know your vocab! Its your best tool! You should know the keywords from your reading assignments!

Topics to focus on:

Basic cell cycle (G0, G1, S, G2, M) and generally what's going on in the cell in each phase
Mitosis and Meiosis: you should know the steps, how many genome copies there are at each step, what the chromosomes are doing, where they are going, how they are moving.
IMPORTANT: understand the molecular basis of the **independent assortment of chromosomes** during meiosis! This is such an important concept for genetic variation!
Crossing over between homologous chromosomes: generally what happens, what is the associated vocabulary (bivalent, chiasma, etc)

IMPORTANT: Mendelian principles: (know your vocabulary here, too!) know how to do Punnett squares (for one and two genes), calculate genotypic and phenotypic ratios, understand the outcomes of various types of crosses, know what is meant by parental generation, filial generation (F1 etc), testcross.
- calculating the number of genotypes (3^n) or gametes (2^n) from a cross between heterozygotes
-Predicting genotypic and phenotypic outcomes using the product rule and sum rule (pay attention to whether the question is asking for the GENOTYPIC or PHENOTYPIC ratio!!!!!)

IMPORTANT: Pedigrees: be able to identify inheritance patterns and infer genotypes based on pedigree information (Note: Don't over-think these pedigrees! If it seems like the most obvious answer is autosomal dominant, but it is remotely possible under some circumstance that it is sex-linked recessive – **go with the obvious answer!)** Also remember that when we are talking about rare genetic diseases, we assume that non-family members (people that marry into the family) are homozygous for the wildtype condition when determining an inheritance pattern UNLESS there is irrefutable evidence that they are not. You should also be able to draw pedigrees based on a family history, and calculate the probability that new offspring will be affected by a condition using the product rule.

IMPORTANT: Linkage analysis:

- be able to identify linkage based on altered offspring ratios, and calculate the recombination frequency (RF) and theoretical distance in map units (m.u)
- know how recognize linkage in a pedigree, infer genotypes of individuals based on the pedigree, and calculate an RF
- Understand the various types of molecular markers (SNPs, RFLPs, SSLPs: mini and micro) and how they are used to identify the location of genes in the genome.

IMPORTANT: Gene interactions – how can interactions between genes affect the expected outcome of crosses?

- Be able to identify examples of co-dominance, incomplete dominance, lethal alleles, pleiotropy, polygenic inheritance
- epistatic relationships (9:3:4, 9:7, 12:3:1 and 15:1) and be able to understand what those altered ratios tell you about how two genes are likely interacting with each other (ie are they redundant? required in a single pathway in series? jointly required for a phenotype?)
- Understand and explain the difference between penetrance and expressivity, and be prepared to give examples.

Journal Club: there will be one brief question relevant to our Journal Club (I will not ask you to memorize how many cell lines were screened for mutations or any detail like that) but it will be something general to our discussion (Perhaps an ethical consideration we discussed, or I may ask you to explain the results from a figure we discussed - you will be given the figure and legend, etc.) Your attendance and active participation in the discussion will be enough to be able to answer the question.