Remember: This study guide is no more than a checklist of what you should know for the exam. It is not a substitute for attending class or reading the lecture notes and text. Anything we covered in class is fair game, even if it is not explicitly listed here on the study guide. Exam II will be given on Wednesday, March 7 and will consist of 50 multiple choice questions.

**Gene Interactions**

Once again, be sure you understand the terminology. Make yourself a list of vocabulary words to double-check yourself.

**Do not freaketh out** at the terms “gain of function” and “loss of function” mutations. They are just what they sound like. They are all around us all the time. The terms are not mysterious code.

**Understand** pleiotropy, and epistasis/hypostasis. Again, these are just TERMS for phenomena that should be intuitively easy to understand. It’s the language of genetics.

**Understand** the very basic idea of “inborn errors of metabolism.” It’s simply when a critical enzyme mutates to be non-functional in the pathway in which it’s supposed to work. Voila!

**Be sure** you understand complementation. Make up some practice examples of your own to see how this works.

**Don’t memorize** the names of the gene loci involved in mammal (including horse) coat color. But be able to apply what you know about how they work if I were to give you information about them in a problem.

**Understand** the different types of suppressor mutations.

**Understand** the concepts of penetrance (a populational thing) and expressivity (an individual thing), and how these two things contribute to population polymorphism.

**Understand** how various environmental influences (sex, age, external factors) can influence (or even mimic) gene expression.

**Understand** how maternal effect works. In short: The **GENOTYPE** of the mother determines (at least in part), the **PHENOTYPE** of the offspring because of very early developmental control.

**DNA Structure and Replication**

These are things you have seen before, though perhaps not in as much detail. They are pretty straightforward, and the notes should serve as the study guide here.

**Please don’t ask** the dreaded question: “Do we have to know...?” Because I’ve already answered that one: If it’s in the notes, then you can consider it important enough to possibly show up on the exam.

**Know** the basic components of the DNA molecule and nucleotide subunits.

**Know** the meaning/significance of: nucleotide, nucleoside-phosphate, phosphodiester bond, 3’ and 5’ direction, the numbering system of the nucleotide carbons,

**Know** the basic steps and enzymatic “players” in DNA replication. (I’m not going to list them here!) **Understand** Chargaff’s Rule and how to apply it.

**Understand** what is meant by semi-conservative (as opposed to conservative) DNA replication as well as continuous vs. discontinuous replication on the leading and lagging strands. **Know** the meaning/significance of: autoradiography, replicon, Okazaki fragment, template strand, coding vs. non-coding strand, sense vs. antisense strand, etc.

**Know** what is meant by a topoisomer, and the significance of topoisomerases, endonucleases, exonucleases.

**Know** the basic differences between prokaryotic and eukaryotic DNA replication. Understand the problem posed by telomeres, and how this is overcome by the cell. Know the names of the people largely responsible for pioneering this work (Greider and Blackburn).

**Know** the basic structure of a eukaryotic chromosome, including what is meant by a nucleosome, histone, etc. (You still need to remember the terms you learned for Exam I)

**Know** the meaning/significance of: genome and the different types of genomes, nucleotide (which are purines, and which are pyrimidines? Which ones bind to each other? Which ones are found in DNA? RNA?)
Know the contributions of Griffiths, Avery, Chargaff, Franklin, Watson & Crick, Meselson and Stahl, Greider and Blackburn

Know the structure of a nucleotide, including its 5’ → 3’ directionality. Know what is meant by a nucleic acid growing from 5’ to 3’ when manufactured.

Know the meaning/significance of: phosphodiester bond, nitrogenous base, introns and exons, regulatory regions of a gene, repetitive DNA, replisome, helicase, gyrase, primase, ligase, destabilizing proteins, bidirectional synthesis, semi-conservative replication (vs. conservative replication)

Understand the phases of DNA replication and how it works. Be able to recognize the enzymes responsible for the various stages.

Know the meaning/significance of: leading vs. lagging strand, Okazaki fragment, Y-junction/replication fork; continuous vs. discontinuous replication, template strand, RNA primer, downstream, upstream, topoisomers of DNA, endonuclease vs. exonuclease activity, processivity, histone, nucleosome

Understand the difficulty of telomere replication, and how the cell manages this task. Know some of the implications of disorders related to telomerase dysfunction.

Transcription and Transcript Processing

Understand the structure and properties of RNA, the types of RNA, the phases of transcription (DNA → RNA),

Recognize the names and roles of the various enzymes involved in transcription and processing.

Know the meaning/significance of: core factor, sigma factor, RNA polymerase, promoter, Pribnow Box, conserved vs. consensus sequences, coding vs. non-coding strand; sense vs. antisense strand, TATA Box/Goldberg-Hogness Box

Understand what is meant by haplo-sufficiency and haplo-insufficiency.

Understand the mechanisms of initiation, elongation and termination.

Know the major differences in Transcription and Translation between prokaryotes and eukaryotes.

Understand the roles of various types of RNA (mRNA, tRNA, rRNA, miRNA, siRNA, dsRNA)

Know the meaning/significance of: self-splicing introns, ribozyme, spliceosome and its components, RNA editing, RNA interference

Know the basic structure, components, and properties of the functional RNA molecules (mRNA, tRNA, rRNA, snRNA). Understand the relationship of DNA to RNA, and how the coding strand may differ in viruses, true bacteria, and eukaryotes.

Know the meaning/significance of: primary transcript, polymer, RNA polymerase, sigma factor, core factor, initiation, elongation, termination, codon, anticodon, stop codon, conserved sequence, consensus sequence, Pribnow Box, upstream vs. downstream, coding vs. non-coding strand (and the synonyms for each), inverted repeats, stem loop, polyadenylation,

Understand the basic relationship between gene expression and changes in organismal response to internal and external change (daily, seasonal, life cycle, etc.)

Understand the basic mechanism of termination in bacteria.

Know the major similarities, and differences in transcription between eukaryotes and bacteria.

For Eukaryotic transcription, know the meaning/significance of: carboxyl tail domain (CTD), general transcription factor (GTF), TATA Box (and synonyms), TATA-binding protein, pre-initiation complex, phosphorylation vs. dephosphorylation, phosphatases, phosphorylases, alternative splicing, proteome,

Understand the post-transcriptional processing of the eukaryotic hnRNA (introns/exons, “capping” of 5’ and 3’ ends of the transcript, etc.) and how it’s done (and by what)

Understand the basic function of a ribozyme and the spliceosome (with the snRNPs).

Know the meaning/significance of RNA editing, RNA interference, interferon’s, double-stranded RNA
Gene Expression/Translation
Know the phases of translation (RNA --> protein), inborn errors of metabolism (malignfunctioning alleles), Dominance and Recessiveness at the protein level, cellular division of labor among genes
Know the meaning/significance of: genome, proteome, pseudogene, peptide bond, globular protein, fibrous protein, prion,
Understand the basic mechanisms of translation, as well as the phases and the enzymes involved.
Know the different levels of protein structure and their significance.
Be able to read the Genetic Code table.
Know the meaning/significance of: Wobble Hypothesis, codon, anticodon, amino acid activation (and the enzymes and components involved)
Know the basic structure and workings of a ribosome.
Know how translation differs between prokaryotes and eukaryotes
Know the meaning/significance of: phosphorylation, kinase enzymes, phosphorylase, phosphataes, ubiquitin, ubiquitination, signal sequences,

Biotechnology
Know the meaning/significance of: recombinant DNA, gene cloning, mosaic, chimera, transgenic organism, vector, transformation, Taq polymerase, probe, vector, reporter gene, “sticky end”, constitutive mutant
Know the meaning/significance of restriction endonuclease/enzyme, restriction fragment, restriction site.
Review the link explaining restriction enzymes and their function at http://www.bio.miami.edu/dana/dox/restrictionenzymes.html
What is the purpose of the Polymerase Chain Reaction? What are the three general possible results of an attempted gene transfer into a new cell?
What is bioinformatics?
What is a DNA library? What are the different types of DNA library, and how are they different from each other? What is the practical use of each type?
Know how the Dideoxy method (Sanger method) of DNA sequencing works, and how to read the resulting gel. (View the video tutorial linked to the notes for a good review)
Read the brief section on Knockout Technology in the notes, and know the purpose of Knockout Technology.