Study Guide for Exam IV

A study guide is not a substitute for reading the text and studying the notes and practicing problems. It’s a handy checklist that should help you focus on the most important aspects of what we’re covering on this exam. If you can’t find it in the text or notes…Google it! You’ll be amazed at what you learn.

Mutations at the Molecular Level

**Know** the meaning/significance of: mutation (process and effect), evolution, organic evolution, microevolution, macroevolution, physiological adaptation vs. evolutionary adaptation, mutagen, carcinogen

**Understand** the meaning/significance of: germline vs. somatic mutations, determinate vs. indeterminate growth

**Understand** what is meant by the various types of mutations: point mutation, transition, transversion, silent vs. synonymous, missense, conservative vs. nonconservative, nonsense, frameshift, suppressors, neutral, forward vs. reverse, exact vs. equivalent reversion, pseudo wild type

and know the various causes of mutations.

**Understand** the nature of various types of mutations such as: depurination, deamination, oxidative damage, tautomer-induced mispairing/tautomeric shift, trinucleotide repeats (and genetic anticipation, premutation)

**Be familiar with** the nature and operation of various types of mutagens, including: chemical (based modifying agents, base analogs, intercalating agents, mycotoxins, ionizing radiation.

**Know** the meaning/significance of: free radical, pyrimidine dimer, xeroderma pigmentosum, dimer repair mechanisms (e.g., photolyase), damage prevention vs. excision repair, damage reversal

**Know** how endonucleases and exonucleases can cooperate with polymerase I and ligase to effect excision repair

**Know** the meaning/significance of: essential gene, lethal mutation, morphological mutation, conditional mutant, restrictive vs. permissive conditions

The Genetics of Cancer

**Know** the meaning, significance, stages (and traditional pronunciation) of apoptosis and cell proliferation,

**Know** what contributes to the generation of a cancer cell from a normal cell.

**Know** how normal cells and cancer cells respond to death cues, survival cues, proliferation cues, and growth inhibition cues.

**Know** the precise meanings of: totipotent, pluripotent, and multipotent cells, and be able to recognize examples of each.

**Understand** the roles of each of the following in cell proliferation: M, G1, G0 checkpoint, S, and G2 phases of mitosis, protein kinases (cyclins and CDKs and their complexes), phosphatases, Rb, E2F, p53, p21, cyclin-Cdk2. Be able to figure out what would happen if any of these mutated.

**Understand** the roles of each of the following in apoptosis: caspases, zymogens, scavenger cells, cyclin-CDK complex. Be able to figure out what would happen if any of these mutated.

**Know** the meaning/significance of: proto-oncogene, oncogene, tumor suppressor, and be sure you understand what happens when a protein with a particular function mutates and either suffers loss-of-function or gain-of-function (Table 15-1 in your notes is a good place to review this!) Examples such as Retinoblastoma and the Philadelphia chromosome will help you understand these things.

Chromosomal Mutations

**Recall** the various types of chromosomal mutations, including deletions, duplications, translocations, inversions. Know the cytogenetic and possible phenotypic consequences of each.

**Understand** the mechanism of semi-sterility in the inversion and translocation heterozygotes, and how an individual meiocyte may divide to produce either viable or non-viable gametes.

**Understand** the possible consequences of single and double breaks of one chromosome or chromatid.

**Know** the meaning/significance of: inversion heterozygote, paracentric inversion, pericentric inversion, translocation heterozygote, reciprocal translocation, alternate vs. adjacent segregation at meiosis in a translocation heterozygote, position effect, cancers as they relate to these mutations, Robertsonian fusion, centromere fission, NF (fundamental number) of chromosomes

**Understand** what is meant by gene balance, and its significance with respect to aneuploidy vs. polyploidy

**Know** the meaning/significance of: chromosomal rearrangements (deletions, duplications, inversions, translocations) synteny, aberrant euploidies (allopolyplody, autopolyplody), and their significance to evolution of genomes in animals, plants, and fungi.
Genomes and Transposable Genetic Elements

Much of this will sound familiar, so it’s basically a review/checklist. But from your notes and readings in the Genomics chapter, know the meaning/significance of: bioinformatics, comparative genomics, functional genomics, conserved sequence, consensus sequence, genomic library, proteome, ORF (open reading frame), pseudogene, DNA barcode.

Under the Comparative Genomics (or via a dictionary search), be sure you know the definitions of: genes that are homologs, orthologs, or paralogs.

Under the Functional Genomics section, know the meaning/significance of: transcriptome, proteome, interactome.

Know what is meant by “reverse genetics”: inferring a [gene’s, mRNA’s, or protein’s] normal function by intentionally disrupting the molecule in a model organism

Be able to recognize the names of the various basic types of transposable genetic elements. Know why these elements disrupt normal gene function.

Know the contributions of Barbara McClintock and Marcus Rhoades to our knowledge of TGEs.

Understand the relationship between the Ds element and the Ac element in corn. Which one is autonomous? Which is nonautonomous? What do autonomous and nonautonomous mean, in terms of TGE tranposition?

Know the meaning/significance of: unstable phenotype, autonomous vs. nonautonomous TGEs, mutable allele, inverted repeats, insertion sequence, polar mutation, plasmid, stem loop, R factor episome (in Shigella bacteria in Japan), resistance transfer region

Know the difference between and significance of replicative and conservative transposition, and between precise and imprecise excision of transposons.

Understand how P elements cause hybrid dysgenesis in fruit flies with mutant M type cytoplasm.

Know the meaning/significance of: target gene, receptor element, regulator gene, controlling element.

What is the C-value paradox? What do long interspersed and short interspersed elements in the human genome tell us about this? What is the significance of these elements, and their relationship to particular medical problems?