

## Exam 4 review Sheet

- Exams in this class ask you to demonstrate your knowledge of the subject matter that we have been discussing in class and you have been working on by going over the material and doing practice problems.
- The exam will consist of short answer questions and problems similar to the suggested problems/homework. None of the problems will be a surprise. They are all similar to things you have already worked on. *If you have prepared for the exam, the 55 minutes provided will be ample time to finish the exam. If you are not prepared, you may not finish the exam.*
- The practice exam questions have come from previous exams. They are good practice questions.
- This is not an exhaustive review sheet. It only outlines ideas that you should be familiar with.
- The best way to prepare is:
  - Do and understand all of the suggested problems in the book (this is especially important)
  - Do and understand the homework assignment
  - Do and understand questions from the old exam
  - Understand the concepts listed below

### Pharmacogenetics

- The Cytochrome P450 Enzymes
- Poor vs Rapid metabolizer genetic variants
- Estrogen Receptor positive vs Estrogen Receptor Negative breast cancers
- Her2 and Breast Cancer

### Sequence Variation and Forensics

- Allelic Frequency and Genotypic Frequency
- How PCR works
- Y linked loci for male paternal line, Mitochondrial DNA for maternal lines
- What are Minisatellites and Microsatellites? How can you use PCR to identify an individual's genotype at these loci?
- What are the similarities between non-coding loci and protein coding genes?
- The use of VNTR typing in forensics both in being able to tell people apart and distinguishing different bacteria

### Mapping Mendelian Traits in Humans

- Linkage and types of markers used (microsats, etc.)
- What is heterozygosity in regards to marker loci and why is it important for mapping?
- Informative vs. Uninformative Meioses
- Phase Known vs. Phase Unknown
- LOD Score Method
  - Calculating LOD scores at different recombination distances
  - Reading a Graphical Representation of LOD scores for a Particular Locus
  - Problems that can arise with mapping by linkage analysis
- The concept of Linkage Disequilibrium
- What is a haploype?
- What is a Disease Haplotype? How is it formed?
- Homozygosity mapping and recessive traits
  - Identifying the Disease Haplotype in families (examples of Familial Dysautonomia and Achromatopsia)

\*Make sure to try the example human genetics problems I posted on webCT. Do these to test your understanding of the material.