MCDB 2150–Genetics: Course and Topic Learning Goals

After completing this course, students should be able to:

1. Analyze phenotypic data and deduce possible modes of inheritance (e.g. dominant, recessive,
autosomal, X-linked, cytoplasmic) from family histories.
Draw a pedigree based on information in a story problem.
Calculate the probability that an individual in a pedigree has a particular genotype.
Define the terms "incomplete penetrance," "variable expressivity," and "sex-limited phenotype," and explain how these phenomena can complicate pedigree analysis.

2. Describe the molecular anatomy of genes and genomes.
Recognize that a given gene is generally situated at the same chromosomal locus in a species.
Differentiate between a gene and an allele.
Diagram a typical eukaryotic gene and indicate the locations of (a) regions that are genic but are not coding, (b) regions that are transcribed but not translated, and (c) regions that are both transcribed and translated.
Describe the general organization, possible function, and frequency of genes and non-gene
DNA sequences in a typical eukaryotic genome.
Explain the functional significance of packaging DNA into chromosomes and the lack of correlation between chromosome number and genetic information content.

3. Describe the mechanisms by which an organism's genome is passed on to the next generation.
Define somatic and germline cells, and list similarities and differences between them.
Recognize why germline mutations can be passed onto the next generation, whereas somatic
mutations cannot.
Describe, using diagrams, the sequence of events involving DNA in meiosis from chromosome duplication through chromosome segregation.
Describe the phenomena of linkage and independent assortment of alleles during meiosis, and explain why some pairs of alleles exhibit linkage and others do not.
Explain how independent assortment can lead to new combinations of alleles of unlinked genes.
Diagram the process of homologous recombination during meiosis and explain how it can lead to new combinations of linked alleles.
Explain how a specific combination of linked alleles (haplotype) can persist through many generations.

4. Extract information about genes, alleles, and gene functions from genetic crosses and human
pedigree analysis.
Design genetic crosses to provide information about genes, alleles, and gene functions.
Explain why it is advantageous to use true-breeding organisms in crosses.

Predict progeny genotypic frequencies given the genotypes of the parental gametes.
Identify an allele's mode of inheritance from progeny phenotypes.
Place genes in a functional order based on the phenotypes of double mutants, and explain the
assumptions that must be made when interpreting these results.
Determine gene linkage and genetic map distances by analyzing progeny with recombinant phenotypes.
Use statistical analysis to determine how well data from a genetic cross or human pedigree
analysis fits theoretical predictions.
Determine if two mutations affect the same gene using complementation tests, and explain
the requirements and the basis for interpreting results from these tests.

5. Describe the processes that can influence the frequency of alleles in a population.
Determine allele frequencies based on phenotypic data for a population in equilibrium.
Explain how natural selection and genetic drift can affect the elimination or maintenance of
deleterious alleles in a population.

6. Cite examples of gene dosage variation (ploidy), and explain why it affects phenotype.
Discuss why alterations in chromosome number can be detrimental.
Describe the process of X inactivation in mammals, and explain its function.

7. Compare different types of mutations and describe how each can affect genes, mRNA and
proteins.
Explain, using diagrams, how nucleotide changes result in the alteration of protein activity.
Explain why some mutations do not affect protein structure or function.
Describe how deletions, inversions, translocations, and the movement of transpositional elements can affect gene function, gene expression, and genetic recombination.
Describe how mutations arise and how environmental factors can increase mutation rate.
Cite examples of mutations that can be beneficial to organisms.
Explain why some DNA damage does not result in mutation.
Distinguish between a DNA replication error and a mutation.
Explain what is meant by a single-nucleotide polymorphism (SNP) and how SNPs can be used as genetic markers even if they do not affect protein structure or function.

8. Explain the molecular basis at the protein level for allele types with different genetic behaviors.
Describe the differences between loss of function and gain of function mutations and their
potential phenotypic consequences.
Predict the most likely effects on protein structure and function of null, reduction-of-

function,
overexpression, dominant-negative and gain-of-function mutations.

9. Justify the value of studying genetics in organisms other than humans.
Explain why it is useful to investigate functions of many human genes by studying simple model organisms such as yeast, nematode worms, and fruit flies.
Describe the benefits and limitations of using model systems to study human diseases.
Use bioinformatic data to compare homologous genes in different species and infer relative
degrees of evolutionary relatedness.

10. Describe the steps that are taken to determine the molecular identity of a human gene that
when mutated can underlie a disease.
Use information from model organisms to identify candidate genes in humans.
Use pedigree information and DNA markers to track a disease trait in a family.
Explain, correctly apply, and interpret results from molecular genetic tools such as DNA sequencing, SNP analysis, and microarrays.