

BIO 135 Study Guide for Exam 3

The 3rd exam will be worth 100 points but will have a possible 110 points available (i.e. there are 10 extra credit points). This study guide is intended to tell you the kinds of information you should know for the first exam, not exactly what will be on it!

Vocabulary

homozygous	trisomy	vector
heterozygous	deletion	plasmid
hemizygous	duplication	bacteriophage
X inactivation	translocation	BAC
Barr bodies	inversion	YAC
Heterochromatin	trisomy 21	restriction enzyme
Euchromatin	turner syndrome	Amp ^r gene
Telomere	triplo-X	LacZ gene
Centromere	Klinefelter syndrome	TA cloning
polyploidy	XYY syndrome	SNP
aneuploidy	origin of replication	STR
monosomy	haplotype	VNTR

What is the X chromosome? What is the Y chromosome? How are they inherited? What genes do they carry? How does this affect sex differentiation? How / where do the X and Y chromosome recombine? What is the NRY (non-recombining Y chromosome)? The NRY is sometimes called the MSY. What is the SRY gene? What does it do?

Be able to work a Punnett Square for X and Y linked traits. How are X linked dominant and X linked recessive alleles inherited? Be able to show this on a pedigree.

What are the parts of a chromosome? What is Trisomy 21? What is the genotype for Trisomy 21? What is the phenotype? Be able to explain (in detail) another chromosomal abnormality (one other than Trisomy 21). What is the genotype? What is the phenotype?

What is the difference between heterochromatin and euchromatin? What is X inactivation? What is the XIST gene? How does X inactivation affect humans? How does this differ from cats? How is X inactivation in humans similar to X cats?

What is recombinant DNA? How can you create a recombinant DNA molecule? How can you select a DNA molecule? What are some of the application of recombinant DNA technology? Name some of the drugs that are produced using DNA technology. How are recombinant DNA molecules replicated, transcribed, and translated *in vitro*? Name some examples.

What is molecular cloning? Be able to list and explain the 4 steps in a molecular cloning experiment. Name two ways that researchers perform the fragmentation step. What is a vector? What is a plasmid? Be able to give examples. What are some ways that plasmids can be used. What is the Amp^r gene and what does it do? What is the LacZ gene? What is the GFP gene? In what organism does the GFP gene occur naturally? What is a multiple cloning site? Name a bacterial species that is commonly used for molecular cloning. Be able to explain when these genes are or are not expressed (i.e. presence/absence of certain chemicals). What is the difference between molecular cloning and organismal cloning?

Name some of the types of genetic mutations. What is the difference between a SNP, STR, and VNTR? What are the typical repeat lengths for each of these? Be able to describe the difference between the following types of mutations: missense, nonsense, frameshift, deletion, insertion, and duplication.

What is a restriction enzyme digest? Be able to draw a digest pattern for a PCR product where there are three genotypes: homozygous allele 1 (e.g. homozygous dominant), heterozygous, homozygous allele 2 (e.g. homozygous recessive).

What is the UCSC Genome Browser? What is a BLAT search? If given a picture of the UCSC Genome Browser page, be able to describe the major features of the main window including chromosome number and which chromosome arm (short or long), DNA sequence, other species that this sequence aligns to, and the amino acid sequence and gene name (if sequence is in a gene region). What are IUPAC codes? Be able to map both the forward and reverse primers onto a single-strand sequence.

What is the difference between microevolution and macroevolution?

What is the definition of a population? What is a gene pool? Be able to describe what the field of population genetics studies. Be able to calculate population allele frequencies. Explain why phenotypic frequencies vary among populations.

What is Hardy-Weinberg? What is the primary assumption made when employing the H-W equation? What is the H-W equation used for? Be able to apply the H-W equation for a biallelic locus. If given the genotype frequencies, be able to calculate allele frequencies. If given the allele frequencies, be able to calculate genotype frequencies. Calculate H-W frequencies if given either an autosomal locus or an X-linked locus.

What is DNA profiling? What types of mutations are used for DNA Profiling? Which types of mutations are the most common and why? What are the applications of DNA Profiling? If given a DNA profiling gel, be able to determine if two samples come from the same individual. If given a DNA profiling gel, be able to determine if an individual could be the parent (or child) of another individual. Given the frequencies of alleles at several loci, be able to calculate the probability that an individual is carrying a particular suite of alleles. For example, if the frequency of allele 1 at locus A is 0.10, allele 1 at locus B is 0.4 and allele 2 at locus C is 0.0001, what is the probability that an individual that has this allelic profile (1-A, 1-B, and 2-C)? What are the CODIS markers? What are they used for?

What is a haplotype? If given 3-4 segregating sites (i.e. polymorphisms) and an outgroup sequence (e.g. chimpanzee), be able to draw a phylogenetic tree representing the relationship among haplotypes. Both the Y chromosome and mtDNA haplotypes trees indicate that the most ancestral lineages are found in Africa and the greatest diversity is found with in Africa. What does this mean in terms of modern human origins? What is National Geographic's Genographic Project?

What are GM foods? Give an example.