Exam 1 Study Guide BB2920

Format:

The exam will consist primarily of open response questions and problems in the style of the problem sets. A sample exam from last year has been posted.

General Advice:

- **READ THE QUESTION CAREFULLY!!** This was (and always is) a major place where students lose points. Eg, Does the question ask for the leading strand or lagging strand? Template or coding? Does it say you are looking at a prokaryotic or a eukaryotic gene? If you are stuck on a question, often you can get unstuck by reading the question again carefully and taking note of these types of details.
- **REMEMBER YOUR RULES!** Nucleotides can only be added to open 3' ends! Complementarity: A pairs with T and G pairs with C! Base paired sequences must be antiparallel! This applies to all nucleic acids, DNA and RNA, replication, transcription and all techniques (PCR, sequencing, etc!)
- Anything in the final posted version of the lecture notes is fair game.
- **I will not ask anything from the textbook which I did not cover in class.**

Topics to Study (The following may be an incomplete list. As I said above, anything in the final posted version of the notes is fair game.):

Lecture 1: Genes, Heredity and Biological Variation

- Understand the concept of a gene
- Relationship between genotype and phenotype
- Identify sources of variation
- Identify types of variation (continuous vs. non-continuous)
- Understand approaches to studying genetics (forward and reverse)

Lecture 2: Genomes, Chromosomes, and the nature of DNA

- Be able to define and understand: genome, ploidy, haploid vs. diploid, autosome vs. sex chromosome, karyotype, homologous chromosomes, somatic vs. gametic
- Understand and explain differences between prokaryotic and eukaryotic genomes
- Chromosome structure: chromatin, nucleosomes, histones, levels of chromatin packing
- the nature of DNA: biochemical features of the four nucleotides (phosphate, sugar, nitrogenous base), features of the nucleic acid strand (antiparallel, directionality [5' and 3'], complementarity, hydrogen bonding)

Lecture 3: DNA replication

- Origin of replication

- Functions and order of operation of the following enzymes: helicase, gyrase, single-stranded binding protein, DNA polymerase III, primase, beta-clamp, DNA polymerase I, ligase.
- Direction of synthesis, leading strand vs. lagging strand/okazaki fragment, and be able to explain WHY replication takes place in this way (remember DNA templates can ONLY be READ 3'->5', and DNA can ONLY be SYNTHESIZED 5'->3'!) If you follow this rule first and foremost, you will always be able to properly identify your strands!!!

Lecture 4: Transcription (DNA -> RNA)

- Biochemical nature of RNA, differences between RNA nucleotides and DNA nucleotides
- Components of a Gene: transcriptional unit (5'UTR, Exons, Introns, 3'UTR) and regulatory unit (promoter)
- Upstream vs. Downstream
- Promoters: Location of promoter sequences, proteins that bind promoter, numbering of gene and promoter region
- Identifying direction of transcription, template=noncoding, coding=nontemplate (remember the DNA template strand can ONLY be READ 3'-> 5'. RNA can ONLY be SYNTHESIZED 5' -> 3'!)
- RNA processing (capping, splicing, and polyadenylation), when and *why* each of these are done
- Differences in transcription and RNA processing between prokaryotes and eukaryotes (promoter sequences, presence of introns, processing steps)

Lecture 5: Translation (RNA-> protein)

- Understand the features of a mature (fully processed) mRNA: cap, 5'UTR, ORF, start codon, stop codon, 3' UTR, polyA tail
- Important features of the genetic code: unambiguous, degenerate, non-overlapping, universal
- Note: you will NOT be asked to memorize codons EXCEPT you should be able to recognize AUG (start codon, = Met). If you need to do a translation you will be provided with a codon chart.
- Ribosome and tRNAs, anticodon, wobble pairing
- Directionality of proteins, N -> C, established by peptide bonds between amino and carboxy groups
- Establishing the reading frame, based on identification of start codon

Lecture 6: Mutations

- Physical mutations: Point mutations (transition, transversion, insertion, deletion), Frameshifts (insertion, deletion), missense (conservative vs. non-conservative, you do not need to memorize amino acid groups, if you need to make this determination you will be given an amino acid chart), nonsense, silent.
- Functional mutations: loss-of-function (hypermorph, amorph or null), gain-of-function (hypermorph, neomorph, antimorph), be able to give examples of each.

Lecture 7(+): Tools of molecular genetics (cloning)

- cloning, vectors and inserts, function and use of restriction enzymes, predicting the size of DNA fragments based on a vector map (like the break-out question)
- PCR amplification of a gene or gene region using primers and purified DNA polymerase, creating mutations, designing primers for PCR
- Reverse transcription and why cDNA is useful.
- DNA sequencing, how it works, why you need ddNTPs
- Blots, and their uses for interpreting genetic information
- Southern, Northern and Western, and how to deduce information about mutations