**BSC2005 Exam #2 Study Guide**

In general, material from the book (and other readings), material from the lecture videos and other videos, and material from the assignments (especially those which reinforce a concept from the reading and/or lectures) is all fair game for the exam.

This is not an exhaustive list of everything you may need to know, and not every single thing from this list will necessarily be asked about on the exam, but this study guide is meant to help you focus in your studying effort on the most important concepts from the modules covered by this exam.

**Module 5: DNA and Genetics**

* What does DNA stand for? How does this name correspond to its molecular structure (also review from Module 2)?
  + Deoxyribonucleic acid. Double strand of nucleic acids. Run in opposite directions
* What are chromosomes? What do they look like in prokaryotes vs. eukaryotes?
  + A diagram of a cell

    Description automatically generated with low confidenceA chromosome is a single, large DNA molecule wound around proteins. DNA in prokaryotes is double-stranded and circular. Eukaryotic DNA, on the other hand, is double-stranded and linear

A diagram of a dna

Description automatically generated with medium confidence

* How many types of chromosomes do humans have? How many total chromosomes?
  + 23 pairs of chromosomes, 46 total chromosomes
  + 22 pairs of numbered chromosomes (autosomes), one pair of sex chromosomes
* What are homologous chromosomes? What are chromatids?
  + A picture containing diagram, pattern

    Description automatically generatedTwo chromosomes in a pair are homologous. Chromatids are two identical DNA molecules that results from the replications of a chromosome during the S phase
  + When a cell is not dividing, DNA is in loosely gathered strands called chromatin
  + When a call is dividing, chromatin coils up tightly to form chromosomes
* Explain the difference between haploid and diploid organisms / cells.
  + Haploid has no copies of chromosomes. Diploid has pairs of chromosomes
  + A picture containing text

    Description automatically generated
* How is diploidy maintained during reproduction?
  + DNA molecules are replicated prior to separation
* What are the four types of nitrogenous bases? Which ones pair with each other? Be able to use Chargaff’s rule to determine the nucleotide composition of a segment of DNA.
  + There are four possible nucleotide bases: adenine (A), thymine (T), guanine (G), and cytosine (C)
  + A-T (2xH bonds), G-C (3xH bonds)
  + Chargaff's rules state that in the DNA of any species and any organism, the amount of guanine should be equal to the amount of cytosine and the amount of adenine should be equal to the amount of thymine. (A + G = T + C)
* What was Rosalind Franklin’s role in determining the structure of DNA?
  + Produced pictures of DNA using X-ray crystallography that guided and eventually supported Watson and Crick’s hypothesis on the structure of DNA
* What does it mean to say that DNA is a double helix? What is the structure of each of the strands?
  + Each subunit—called a nucleotide—has three parts: a sugar, a phosphate group, and a base. In each DNA strand, the phosphate group of one nucleotide binds to the sugar of the next nucleotide to form a chain of linked nucleotides. The two strands of linked nucleotides pair up and twist around each other to form a spiral-shaped double helix. The sugars and phosphates form the outside “backbone” of the helix, while the bases point toward its center, forming internal “rungs,” like steps on a twisting ladder. The bases in one strand associate with bases from the other strand through hydrogen bonds, which hold the two strands of the double helix together.
* What are the chromatids? When during cell division do they form, and how? When do they separate?
  + Chromatids are two identical DNA molecules that results from the replications of a chromosome during the S phase
* What happens a) to the chromosomes, and b) to the rest of the cell during each of the following phases of cell division? Interphase, Prophase, Metaphase, Anaphase, and Telophase, Cytokinesis?
  + Interphase: prepares for division. 3 phases:
    - G1: cell enlarges, produces additional cytoplasm, produces new organelles
    - S: DNA replication
    - G2: Cell prepares for division
  + Mitosis: sister chromatids separate and move towards the end of the cell. 4 phases:
    - Prophase nuclear membrane begins to disassemble. Chromosomes coil up (form X). Mitotic spindle begins to form
    - Metaphase: Spindle fibers from opposite ends of the cell attach to sister chromatids. Chromosomes are aligned in the center of the cell
    - Anaphase: Sister chromatids are separated
    - Telophase: Chromosomes reach each pole. Spindle fibers disassemble. Nuclear membrane begins to reform around the separated chromosomes
  + Cytokinesis: cytoplasm divides into two separate, complete cells
* What are the products of mitosis? How many cells? How does their DNA compare to the original cell?
  + Two cells, each cell has 23 pairs of chromosomes

A picture containing text, web page, font, website

Description automatically generatedA diagram of cell division

Description automatically generated with low confidence

* How does DNA replicate? What are the steps involved?
  + During DNA replication, the two strands of DNA separate, with each strand then serving as a template for the creation of a new complementary strand
  + First, an enzyme called helicase unwinds the helix, and the two strands “unzip” from each other.
  + A diagram of a dna structure

    Description automatically generated with low confidenceThen, the enzyme DNA polymerase builds a new strand of DNA along each unzipped strand. Free nucleotides floating inside the cell’s nucleus are added to each new strand in a sequence that is complementary to the nucleotide sequence on the original template strand, with A pairing with T and C with G
* What does it mean to say that DNA replication is semi-conservative?
  + Each of the two new DNA molecules consists of one old and one new DNA strand, with the old strand serving as a template
* What is the job of DNA polymerase?
  + Polymerase attaches new complementary nucleotides to the split DNA molecules.
* How many base pairs does the average human have?
  + 3 Billion
* What does STR stand for, what are they, and how are they used in DNA fingerprinting?
  + A picture containing text, human face, screenshot

    Description automatically generatedShort tandem repeats, found in non-coding areas. Combinations of the length of various STR regions are unique
* What does PCR stand for, and how does it work? What materials need to be included, and what is each of them for?
  + Polymerase Chain Reaction
  + Needs:
    - Primers: identifies where polymerase should bind to
    - Polymerase: replicates DNA
    - A diagram of dna sequence

      Description automatically generated with low confidenceNucleic acids: raw materials for building
    - The DNA to be replicated
  + To a small sample of DNA, scientists add nucleotides, the DNA polymerase enzyme, and primers—short segments of DNA that act as guideposts. The primers bind to complementary segments of each DNA template, and their locations flag the section to which DNA polymerase should bind to begin replication. The DNA is first heated to separate the strands, and then cooled to allow the primers to associate with the DNA and to allow the DNA polymerase to add new nucleotides
* How does the number of copies produced by PCR increase over time?
  + Doubles every round
* What is gel electrophoresis? How does it work? How is it similar to the chromatography you did in Module 4?
  + It separates DNA fragments by size.
  + DNA is negatively charged and migrates towards the positive electrode (at the bottom). Shortest STR fragments travel the furthest/fastest from the source. Different STR regions are colored differently.
* Be able to analyze a picture/diagram of gel electrophoresis (or a table of STR lengths) to determine matches between suspects/crime scene samples (or babies / potential fathers).
  + Exact match means same person
  + A picture containing text, screenshot, design

    Description automatically generatedSharing one band per STR region means parent or child
* What other purposes can DNA fingerprinting serve?
* Cell cycle checkpoints ensure accurate progression through the cell cycle; repair mechanisms at each checkpoint can fix mistakes that occur, such as improper base pairing or DNA damage. If a checkpoint protein is impaired, cells may fail to properly repair DNA mistakes, leading to mutations that are passed on to daughter cells.

**Module 6: Inheritance**

* What are the products of meiosis? How many cells? How does their DNA compare to the original cell?
  + 4 haploid daughter cells. No pairs of chromosomes
* A diagram of cell cycle

  Description automatically generated with low confidenceA picture containing text, font, screenshot

  Description automatically generatedWhat happens a) to the chromosomes, and b) to the rest of the cell during each of the phases of meiosis 1? Meiosis 2?

A screenshot of a cell phone

Description automatically generated with low confidence

* What are homologous chromosomes? What are chromatids?
  + Two chromosomes in a pair are homologous. Chromatids are two identical DNA molecules that results from the replications of a chromosome during the S phase
* Compare and contrast mitosis and meiosis.
  + In mitosis, replicated chromosomes (consisting of a pair of sister chromatids) line up along the midline of the cell. The two sister chromatids of each chromosome then separate, becoming independent chromosomes. Each daughter cell ends up with an identical (diploid) complement of 46 chromosomes.
  + In meiosis, two separate divisions occur, producing four daughter cells rather than two, and giving each cell only one copy of each chromosome. The first division separates homologous chromosomes from each other; the second division, like mitosis, then separates sister chromatids. At the end of meiosis, one (diploid) cell has divided into four (haploid) cells, which develop into egg or sperm
* What is the principle of independent assortment? When does it happen?
  + Each chromosome enters a progeny cell independent of other chromosomes
  + Chromosomes from parents do not stick together
* What is recombination? When does it happen?
  + A picture containing text, screenshot, skeleton

    Description automatically generatedWhen homologous chromosomes line up during meiosis 1, they trade segments of DNA
* Nondisjunction
  + Chromosomes do not separate normally during meiosis
  + Occurs in meiosis 1 if both homologous chromosomes end up in same cell
  + Occurs in meiosis 2 if the two sister chromatids do not separate properly
* What is aneuploidy? When does it happen?
  + Individuals have too many or too few chromosomes
* What is a gene? What is an allele?
  + Genes are chunks of DNA that contribute to particular traits or functions by coding for proteins that influence physiology. Alleles are different versions of a gene, which vary according to the nucleotide base present at a particular genome location. An individual's combination of alleles is known as their genotype.
* What does it mean if an organism is heterozygous? Homozygous?
  + Homozygous: the same genes on homologous chromosomes have the same allele
  + Heterozygous: the same genes on homologous chromosomes have different alleles
* What’s the difference between an organism’s genotype and its phenotype?
  + Genotype: allele make up
  + Phenotype: physical expression of the genotype
* What does it mean for a phenotype to be dominant? Recessive?
  + Dominant: expressed as along as there is at least once copy of the dominant allele
  + Recessive: only expressed if individual is homozygous for the recessive allele
* What is blending inheritance? How did Mendel prove it wrong?
  + A picture containing text, screenshot, font, number

    Description automatically generatedAll offspring will be intermediate between their parents. The law of segregation, which states that the two copies of a gene separate when producing gametes, so each gamete only has one allele
* Given an individual’s genotype, be able to list all of the possible types of gametes that individual could make. Be able to do this for one gene at a time, but also be able to list all of the combinations of alleles in the gametes for more a genotype containing more than one gene.
* Be able to use Punnett squares to get from parental genotypes (or phenotypes) to predicted frequencies of offspring genotypes (or phenotypes). Be able to do this for monohybrid AND dihybrid crosses.
* What’s the difference between autosomes and sex chromosomes?
  + Autosomes (non-sex chromosomes, 1-22)
  + Sex (determine sex, #23), XX = F, XY = M. Y chromosome contains the SRY gene which cause testes to develop
* What does it mean to be hemizygous? Which individuals (in humans) are hemizygous? For which genes?
  + Males are hemizygous for alleles on the X chromosome, as they only have one X. Whatever allele appears on the X will appear in the phenotype
* What are sex-linked/X-linked traits? Which sex (in humans) is more likely to express the phenotype of these traits? Which is more likely to be a carrier? Be able to use this to trace the inheritance of these traits through a pedigree.
  + Traits where the genes are located on the X chromosome. Recessive traits are more likely in males since they only have one X
* Be able to use Punnett squares to get from parental genotypes (or phenotypes) to predicted frequencies of offspring genotypes (or phenotypes) for X-linked traits.
* How does genotype relate to phenotype in a trait with Mendelian (typical) dominance? Incomplete dominance? Codominance? Be able to use Punnett squares to give offspring genotype & phenotype frequencies for each.
  + Mendelian: Dominant is expressed if present, else recessive
  + Incomplete: the heterozygous phenotype is intermediate (red + white = pink)
  + Codominance: both alleles are expressed equally in the heterozygote (horse with both black and white hairs)
* How does ABO blood typing work? What kind of trait is this? Be able to use Punnett squares… etc. etc.
  + If A allele is present, A proteins are present on hemoglobin
  + If B allele is present: B proteins are present
  + If genotype does not have A or B genes, it is type O and neither A nor B proteins are present
  + A and B are codominant, O is recessive
* What are polygenic traits? How do they differ from traits with multifactorial inheritance?
  + Traits controlled by more than one gene
  + Often quantifiable/measurable characteristics, which display a wide range of phenotypes (height)
  + Multifactorial: combination of genes and environmental effects affect the phenotype

**Module 7: Natural Selection & Evolution**

* What is the definition of evolution? Be able to recognize examples of phenomena that do or do not count as evolution.
  + Biological populations change over generations
  + Evolution is defined as a change in allele frequencies in a population over time. Evolution affects populations. Individuals do not evolve.
* What does “theory” mean in the scientific context. Is the criticism that evolution is “just a theory” valid?
  + System of well-supported ideas based on tested hypothesis. Also allow predications to be made
* Natural selection is one cause of evolution. Evolution by natural selection occurs when individuals in a population vary genetically in ways that affect their ability to survive and reproduce in a particular environment. Some alleles become more common, while others become less common as a result.
* What were some of the evolutionary ideas developed by thinkers and scientists before Charles Darwin? What pieces were they missing?
  + Erasmus Darwin: theorized a common ancestor, missing a viable mechanism for change
  + Jean Baptiste Lamarck: use or disuse (stretching necks); mechanism for inheritance
* Explain how Darwin’s voyage on the HMS Beagle, his reading of Lyell and Malthus, and his work with farmers and pigeon breeders shaped his ideas about evolution by natural selection.
  + Geographic variation among species
  + Lyell: principles of geology; the current geology is the result of small changes over a long period of time
  + Malthus: An essay on the principle of population; exponential growth is keep in check due to limited resources (competition)
  + Farmers/breeders: evidence of selection
* Who is Alfred Russell Wallace and why is he important?
  + Formulated evolutionary theory independently of Darwin
* What four things must be true in order for evolution by natural selection to occur? What would occur if each of these things weren’t true? Which one corresponds to the “natural selection” part?
  + Population must show variation in a trait
  + Some of that variation must be heritable
  + More individuals must be born than survive to reproduce
  + Variation in the trait must affect reproductive success of individuals
* Where does variation in a population come from? How does it relate to natural selection?
  + Mutation. Individuals having different traits, as a result of mutations, is required for species to evolve
  + Bacteria can introduce variations via conjugation and lateral gene transfer
* What is fitness (in the evolutionary biology sense)?
  + Ability to survive and reproduce. Ability to get genes into the next generation
* Distinguish between natural selection and evolution.
  + Evolution is the gradual change in species over time; happens to a population; change in the gene pool from one generation to the next; due to natural selection or random processes
  + Natural selection happens to individuals and is not random; one of the mechanisms that affects evolution
* What is an adaptation?
  + A trait which allows a population to become better suited to their environment as a result of natural selection.
  + Over time, natural selection leads to adaptation: advantageous traits become more common in the population, which as a result becomes better suited, or adapted, to its environment.
* Populations are groups of individuals of the same species living together in the same geographic area.
* For each of the directional, stabilizing, disruptive, positive frequency-dependent, and negative frequency-dependent selection, describe what it means in terms of which phenotypes have high vs. low fitness, and predict what will happen to a) the mean phenotype, and b) the variance in phenotype in the population in the next generation.
  + Directional: environment favors one extreme; population shifts towards that extreme over time (artic rabbits with white fur)
  + Stabilizing: Environment favors middle values (birth size)
  + Diversifying: environment favors both extremes, opposite effect of stabilizing selection
  + Positive-Freq-Dependent: selects most common phenotype (poisonous butterflies looking like each other), decreases variation in population
  + Negative-Freq-Dependent: selects rarest phenotype (right vs left favoring fish, leads to oscillation between favoring of each phenotype), maintains variation in population

A picture containing text, human face

Description automatically generated

* What is sexual dimorphism, and why is it a puzzle given what we understand about evolution by natural selection?
  + Males and females have very different traits (size, color). Males tend to have more elaborate traits.
  + Natural selection should select against these traits that reduce survival (peacock)
* How are sexual selection and natural selection similar? How do they differ?
  + Requires a heritable trait, some variants have a higher fitness than others. Can be directional, stabilizing, disruptive, or freq-dependent
  + Difference is that sexual selection favors traits that helps an individual get laid, rather than traits that help them survive
* How does sexual selection by male-male competition occur? What about sexual selection by female choice?
  + Male-male competition: combat, mate guarding, sneaky males (seals)
  + Female-choice: choice based on characteristics. Ability to provide resources/paternal care. Attractive traits. Good health

**Module 8: Non-Adaptive Evolution and Speciation**

* What is a gene pool? For a population of X diploid individuals, how many alleles are in its gene pool?
  + The total of all the genetic variation in a population (allele frequencies). 2X
* For a given population, be able to calculate phenotype frequencies, genotype frequencies, and allele frequencies, and to go back and forth between all three.
  + Total of variant / total in population = freq
* If you take all the allele frequencies for a given population, what do they add up to? What about all of the genotype frequencies? Explain why this is true.
  + 1, this is how ratios work
* Explain where the p^2 + 2pq + q^2 = 1 equation comes from, and what it means.
  + Random mating in a population with two alleles. Identifies the frequency of genotypes based on allele frequency. Hardy-Weinberg equation
* When would you expect to see allele and genotype frequencies be in Hardy-Weinberg Equilibrium (what conditions need to be met)? How could you tell if they weren’t?
  + HWE happens when allele and genotype frequencies in a population do not change across generations
  + Requires:
    - No selection
    - Infinitely large population
    - No migration
    - No mutation
    - Random mating
  + When these requirements are violated, evolution is occurring, because allele frequencies are changing over time
* Given allele frequencies, be able to predict genotype/phenotype frequencies if the population is in HWE, and vice versa.
  + p^2 + 2pq + q^2 = 1
* Lost vs fixed alleles
  + Lost: 0% allele frequency in population
  + Fixed: 100% allele frequency in population
  + Maintained until new allele enters the population through mutation or migration
* What is genetic drift? What are its consequences in terms of allele frequencies in a population? When do you expect it to have a stronger vs. weaker effect?
  + A form of non-adaptive evolution where allele frequencies change randomly in ways that do not affect fitness. More of an effect in small populations.
  + Selection is stronger than drift in large populations, in small populations drift can overwhelm selection
* What is the founder effect? What is the bottleneck effect? How do these relate to genetic drift?
  + Founder effect: Form of genetic drift. New population is founded by a small subset of the original population
  + Bottlenecks: drastic reduction in population numbers that can lead to a reduction in genetic diversity. Alleles are initially lost as individuals die, genetic drift in the remaining small population continues to erode diversity
* What is inbreeding? Which of the HWE requirements does it violate? What effects does it have on a population?
  + Non-random mating where related individual are more likely to mate
  + Violates random mating requirement
  + Doesn’t change allele frequencies, does increase the frequency of homozygotes above what is expected from HWE
  + Causes more recessive phenotypes to be displayed (inbreeding depression)
* What effects do migration and mutation have on genetic diversity?
  + Mutation: ultimate source of all new alleles, very slow
  + Migration: new alleles enter a specific population. Homogenizing effect on populations (reduces difference between separate populations).
* Direction of various forces on genetic diversity
  + Mutation: increases
  + Selection: decreases
  + Non-random mating: none
  + Genetic drift: decreases
* What is the Biological Species Concept? What types of species does it apply to? Are there cases where it isn’t as useful?
  + It defines a species as a group of actually or potentially interbreeding organisms reproductively isolated from other such groups
  + If two individuals can mate and produce fertile offspring, they are the same species
  + Factors that keep two species separate are called isolating barriers
  + Not as useful for species that produce asexually (bacteria, is each individual its own species?). Also, occasionally two distinctly separate species are capable of producing fertile hybrids (trees). Are fertile hybrids their own species (red wolf)?
* What is reproductive isolation and why is it important in speciation?
  + Reproductive isolation allows organisms to evolve to be different enough that they become distinct species that can no longer interbreed if their populations become contiguous again
* Define allopatric speciation. How does it occur?
  + The evolution of new species from a previously interbreeding population that is split by a physical barrier
  + Physical separation (Darwin’s finches)
  + One population becomes separated by a physical barrier and are cut off from each other
* Define sympatric speciation. How does it occur?
  + Two species that are not physically separated, but are still genetically isolated. Isolated by pre and post zygotic isolation
  + May take place without a physical barrier if pre-zygotic mechanisms keep populations from interbreeding
* What types of pre-zygotic reproductive isolation are there? What types of post-zygotic isolation? Be able to identify examples of each.
  + Pre-zygotic isolation: acts before mating
    - Temporal: mate at times of year/day
    - Behavioral: males/females are not recognized as potential mating partners (different mating rituals)
    - Ecological: different environments
    - Mechanical: gametes cannot come into contact (the glove don’t fit)
    - Gametic: egg and sperm (if they meet) do not fuse
  + Post-zygotic isolation: acts after the hybrid offspring forms. A zygote forms but is not viable or it is not fertile