Topic 10 Study Guide

**Note: This is a GUIDE, not an all inclusive list of what will be on the exam. This is meant to help facilitate the studying process.**

**Processes to be able to describe:**

* **How meiosis promotes variation**
  + (in prophase I) crossing over/chiasmata formation between homologous chromosomes
  + random alignment of homologues/bivalents in metaphase I - independent assortment of homologues / chromosomes
  + second division of meiosis separates alleles further
  + combinations of alleles in gametes is unlimited (2n)
* **What is happening during each phase of meiosis I (or II)**
  + Prophase, metaphase, anaphase, telophase
* **How chiasmata form during crossing over (Explain synapsis)**
  + Homologous chromosomes pair
  + Synaptonemal complex
  + Chiasma (X-shaped structure) forms
  + Alleles exchanged
  + Draw diagram
* **DNA Replication happens in interphase before cells go through mitosis or meiosis. Look back at enzymes/process of replication**
* **The process of natural selection**
  + Genetic variation, overproduction, competition, survival of the fittest
  + Alleles for beneficial traits spread in population and become more frequent
  + Selective pressures: predation, lack of food/water/shelter, changing environment, etc.
* **Antibiotic Resistance as an example of natural selection (Review SL concepts on this, or bioninja)**
  + Antibiotics themselves DO NOT CAUSE MUTATIONS in bacteria. Bacteria already have to have aquired a mutation randomly in that gene to survive in the presences of antibiotics, and this can spread through population via conjugation/binary fission.
* **The process of speciation**
  + Population/gene pool separated into two populations
  + Reproductive isolation/lack of interbreeding occurs
  + Isolation that caused this could be temporal, behavioral, or geographic
  + Polyploidy can cause isolation too
  + Differences in types of selection favoring different traits in separate populations/random mutations that are spread through the populations cause gene pool to diverge
  + Changes in gene pool lead to formation of new species
* **Gradualism vs Punctuated Equilibrium**
  + Differences in pace/evidences to support them
* **Sympatric vs Allopatric Speciation**
  + Causes
  + One occurs in same location (population not separated), other occurs with geographic isolation
  + Examples
* **Evidences for evolution (Review SL on this)**
  + Homologous vs vestigial vs analogous structures
  + Fossils
  + DNA/Protein similarity
  + Embryonic Development
* **How polyploidy can lead to speciation**
  + Polyploids made by errors in meiosis where chromosome numbers increase, can no longer breed with parents – more common in plants
  + New species in one generation!
  + Hybrid vigour
* **Human Genome Project (review SL on this)**
  + Outcomes (40,000 genes in humans, only know function of half, mapped where they are located on chromsomes, etc.)
  + Implications (ex: identifying genes involved in genetic disorders/gene therapy)
* **Explain nondisjunction and give examples of disorders caused by it** 
  + ex: Down Syndrome, Turners, Klinefelters, etc.
  + Identify normal vs abnormal karyotypes
  + Identify sources of cells for genetic testing (ex. chorionic villi and amniocentesis)
* **Continuous vs Discrete (discontinuous) variation**
  + Polygenic traits (examples)
  + How environment can influence polygenic traits
* **Analyze Pedigrees (dom vs rec and autosomal vs x-linked)**
* **Crosses:**
  + blood types (codominant), sex-linked, monohybrid, dihybrid, test cross (linked genes especially), codominant/incomplete dominance
* **Descriptions genetic disorders**
  + Especially sickle cell, hemophilia, color blindness, cystic fibrosis
* **Causes and implications of sickle cell anemia.** 
  + Point mutation (substitution) that changes glutamic acid to valine
  + Changes protein structure (Review this from Topic 7 – primary, secondary, tertiary, quaternary)
  + Shape change in red blood cells
  + Link to malaria
  + Link to natural selection

**10.1 Meiosis**

**Essential idea:**

* Meiosis leads to independent assortment of chromosomes and unique composition of alleles in daughter cells.

**Nature of science:**

* Making careful observations—careful observation and record keeping turned up anomalous data that Mendel’s law of independent assortment could not account for. Thomas Hunt Morgan developed the notion of linked genes to account for the anomalies. (1.8)

**Understandings:**

* Chromosomes replicate in interphase before meiosis.
* Crossing over is the exchange of DNA material between non-sister homologous chromatids.
* Crossing over produces new combinations of alleles on the chromosomes of the haploid cells.
* Chiasmata formation between non-sister chromatids can result in an exchange of alleles.
* Homologous chromosomes separate in meiosis I.
* Sister chromatids separate in meiosis II.
* Independent assortment of genes is due to the random orientation of pairs of homologous chromosomes in meiosis I.

**Applications and skills:**

* Skill: Drawing diagrams to show chiasmata formed by crossing over.

**10.2 Inheritance**

**Essential idea:**

* Genes may be linked or unlinked and are inherited accordingly.

**Nature of science:**

* Looking for patterns, trends and discrepancies—Mendel used observations of the natural world to find and explain patterns and trends. Since then, scientists have looked for discrepancies and asked questions based on further observations to show exceptions to the rules. For example, Morgan discovered non-Mendelian ratios in his experiments with *Drosophila*. (3.1)

**Understandings:**

* Gene loci are said to be linked if on the same chromosome.
* Unlinked genes segregate independently as a result of meiosis.
* Variation can be discrete or continuous.
* The phenotypes of polygenic characteristics tend to show continuous variation.
* Chi-squared tests are used to determine whether the difference between an observed and expected frequency distribution is statistically significant.

**Applications and skills:**

* Application: Morgan’s discovery of non-Mendelian ratios in *Drosophila.*
* Application: Completion and analysis of Punnett squares for dihybrid traits.
* Application: Polygenic traits such as human height may also be influenced by environmental factors.
* Skill: Calculation of the predicted genotypic and phenotypic ratio of offspring of dihybrid crosses involving unlinked autosomal genes.
* Skill: Identification of recombinants in crosses involving two linked genes.
* Skill: Use of a chi-squared test on data from dihybrid crosses.

**Guidance:**

* Alleles are usually shown side by side in dihybrid crosses, for example, TtBb. In representing crosses involving linkage, it is more common to show them as vertical pairs, for example:  
  http://xmltwo.ibo.org/publications/DP/Group4/d_4_biolo_gui_1402_1/img/10.2.png

**10.3 Gene Pools and Speciation**

**Essential idea:**

* Gene pools change over time.

**Nature of science:**

* Looking for patterns, trends and discrepancies—patterns of chromosome number in some genera can be explained by speciation due to polyploidy.

**Understandings:**

* A gene pool consists of all the genes and their different alleles, present in an interbreeding population.
* Evolution requires that allele frequencies change with time in populations.
* Reproductive isolation of populations can be temporal, behavioural or geographic.
* Speciation due to divergence of isolated populations can be gradual.
* Speciation can occur abruptly.

**Applications and skills:**

* Application: Identifying examples of directional, stabilizing and disruptive selection.
* Application: Speciation in the genus *Allium* by polyploidy.
* Skill: Comparison of allele frequencies of geographically isolated populations.

**Guidance:**

* Punctuated equilibrium implies long periods without appreciable change and short periods of rapid evolution.
* Many crop species have been created to be polyploid. Polyploidy increases allelic diversity and permits novel phenotypes to be generated. It also leads to hybrid vigour.

**3.1 Genes**

**Nature of science:**

* Developments in scientific research follow improvements in technology—gene sequencers are used for the sequencing of genes. (1.8)

**Understandings:**

* A gene is a heritable factor that consists of a length of DNA and influences a specific characteristic.
* A gene occupies a specific position on a chromosome.
* The various specific forms of a gene are alleles.
* Alleles differ from each other by one or only a few bases.
* New alleles are formed by mutation.
* The genome is the whole of the genetic information of an organism.
* The entire base sequence of human genes was sequenced in the Human Genome Project.

**Applications and skills:**

* Application: The causes of sickle cell anemia, including a base substitution mutation, a change to the base sequence of mRNA transcribed from it and a change to the sequence of a polypeptide in hemoglobin.
* Application: Comparison of the number of genes in humans with other species.
* Skill: Use of a database to determine differences in the base sequence of a gene in two species.

**Guidance:**

* Students should be able to recall one specific base substitution that causes glutamic acid to be substituted by valine as the sixth amino acid in the hemoglobin polypeptide.
* The number of genes in a species should not be referred to as genome size as this term is used for the total amount of DNA. At least one plant and one bacterium should be included in the comparison and at least one species with more genes and one with fewer genes than a human.
* The Genbank® database can be used to search for DNA base sequences. The cytochrome C gene sequence is available for many different organisms and is of particular interest because of its use in reclassifying organisms into three domains.

**International-mindedness:**

* Sequencing of the human genome shows that all humans share the vast majority of their base sequences but also that there are many single nucleotide polymorphisms that contribute to human diversity.

**Theory of knowledge:**

* There is a link between sickle cell anemia and prevalence of malaria. How can we know whether there is a causal link in such cases or simply a correlation?

**3.2 Chromosomes**

**Essential idea:**

* Chromosomes carry genes in a linear sequence that is shared by members of a species.

**Understandings:**

* Prokaryotes have one chromosome consisting of a circular DNA molecule.
* Some prokaryotes also have plasmids but eukaryotes do not.
* Eukaryote chromosomes are linear DNA molecules associated with histone proteins.
* In a eukaryote species there are different chromosomes that carry different genes.
* Homologous chromosomes carry the same sequence of genes but not necessarily the same alleles of those genes.
* Diploid nuclei have pairs of homologous chromosomes.
* Haploid nuclei have one chromosome of each pair.
* The number of chromosomes is a characteristic feature of members of a species.
* A karyogram shows the chromosomes of an organism in homologous pairs of decreasing length.
* Sex is determined by sex chromosomes and autosomes are chromosomes that do not determine sex.

**Applications and skills:**

* Application: Cairns’ technique for measuring the length of DNA molecules by autoradiography.
* Application: Comparison of genome size in T2 phage, *Escherichia coli*, *Drosophila melanogaster*, *Homo sapiens* and *Paris japonica.*
* Application: Comparison of diploid chromosome numbers of *Homo sapiens*, *Pan troglodytes*, *Canis familiaris*, *Oryza sativa*, *Parascaris equorum.*
* Application: Use of karyograms to deduce sex and diagnose Down syndrome in humans.
* Skill: Use of databases to identify the locus of a human gene and its polypeptide product.

**Guidance:**

* Genome size is the total length of DNA in an organism. The examples of genome and chromosome number have been selected to allow points of interest to be raised.
* The two DNA molecules formed by DNA replication prior to cell division are considered to be sister chromatids until the splitting of the centromere at the start of anaphase. After this, they are individual chromosomes.

**3.3 Meiosis**

**Essential idea:**

* Alleles segregate during meiosis allowing new combinations to be formed by the fusion of gametes.

**Understandings:**

* One diploid nucleus divides by meiosis to produce four haploid nuclei.
* The halving of the chromosome number allows a sexual life cycle with fusion of gametes.
* DNA is replicated before meiosis so that all chromosomes consist of two sister chromatids.
* The early stages of meiosis involve pairing of homologous chromosomes and crossing over followed by condensation.
* Orientation of pairs of homologous chromosomes prior to separation is random.
* Separation of pairs of homologous chromosomes in the first division of meiosis halves the chromosome number.
* Crossing over and random orientation promotes genetic variation.
* Fusion of gametes from different parents promotes genetic variation.

**Applications and skills:**

* Application: Non-disjunction can cause Down syndrome and other chromosome abnormalities.
* Application: Studies showing age of parents influences chances of non-disjunction.
* Application: Description of methods used to obtain cells for karyotype analysis e.g. chorionic villus sampling and amniocentesis and the associated risks.
* Skill: Drawing diagrams to show the stages of meiosis resulting in the formation of four haploid cells.

**3.4 Inheritance**

**Nature of science:**

* Making quantitative measurements with replicates to ensure reliability. Mendel’s genetic crosses with pea plants generated numerical data. (3.2)

**Understandings:**

* Mendel discovered the principles of inheritance with experiments in which large numbers of pea plants were crossed.
* Gametes are haploid so contain only one allele of each gene.
* The two alleles of each gene separate into different haploid daughter nuclei during meiosis.
* Fusion of gametes results in diploid zygotes with two alleles of each gene that may be the same allele or different alleles.
* Dominant alleles mask the effects of recessive alleles but co-dominant alleles have joint effects.
* Many genetic diseases in humans are due to recessive alleles of autosomal genes, although some genetic diseases are due to dominant or co-dominant alleles.
* Some genetic diseases are sex-linked. The pattern of inheritance is different with sex-linked genes due to their location on sex chromosomes.
* Many genetic diseases have been identified in humans but most are very rare.
* Radiation and mutagenic chemicals increase the mutation rate and can cause genetic diseases and cancer.

**Applications and skills:**

* Application: Inheritance of ABO blood groups.
* Application: Red-green colour blindness and hemophilia as examples of sex-linked inheritance.
* Application: Inheritance of cystic fibrosis and Huntington’s disease.
* Application: Consequences of radiation after nuclear bombing of Hiroshima and accident at Chernobyl.
* Skill: Construction of Punnett grids for predicting the outcomes of monohybrid genetic crosses.
* Skill: Comparison of predicted and actual outcomes of genetic crosses using real data.
* Skill: Analysis of pedigree charts to deduce the pattern of inheritance of genetic diseases.

**Guidance:**

* Alleles carried on X chromosomes should be shown as superscript letters on an upper case X, such as Xh.
* The expected notation for ABO blood group alleles is:

*Phenotype*  *Genotype*  
    O  ii  
     A  |A| A or | Ai   
     B  |B| B or | Bi   
     AB   |A| B  
      
      
      
**5.1 Evidence for Evolution**

**Essential idea:**

* There is overwhelming evidence for the evolution of life on Earth.

**Nature of science:**

* Looking for patterns, trends and discrepancies—there are common features in the bone structure of vertebrate limbs despite their varied use. (3.1)

**Understandings:**

* Evolution occurs when heritable characteristics of a species change.
* The fossil record provides evidence for evolution.
* Selective breeding of domesticated animals shows that artificial selection can cause evolution.
* Evolution of homologous structures by adaptive radiation explains similarities in structure when there are differences in function.
* Populations of a species can gradually diverge into separate species by evolution.
* Continuous variation across the geographical range of related populations matches the concept of gradual divergence.

**Applications and skills:**

* Application: Development of melanistic insects in polluted areas.
* Application: Comparison of the pentadactyl limb of mammals, birds, amphibians and reptiles with different methods of locomotion.

**5.2 Natural Selection**

**Nature of science:**

* Use theories to explain natural phenomena—the theory of evolution by natural selection can explain the development of antibiotic resistance in bacteria.

**Understandings:**

* Natural selection can only occur if there is variation among members of the same species.
* Mutation, meiosis and sexual reproduction cause variation between individuals in a species.
* Adaptations are characteristics that make an individual suited to its environment and way of life.
* Species tend to produce more offspring than the environment can support.
* Individuals that are better adapted tend to survive and produce more offspring while the less well adapted tend to die or produce fewer offspring.
* Individuals that reproduce pass on characteristics to their offspring.
* Natural selection increases the frequency of characteristics that make individuals better adapted and decreases the frequency of other characteristics leading to changes within the species.

**Applications and skills:**

* Application: Changes in beaks of finches on Daphne Major.
* Application: Evolution of antibiotic resistance in bacteria.

**Guidance:**

* Students should be clear that characteristics acquired during the lifetime of an individual are not heritable.

**5.4 Cladistics**

**Understandings:**

* A clade is a group of organisms that have evolved from a common ancestor.
* Evidence for which species are part of a clade can be obtained from the base sequences of a gene or the corresponding amino acid sequence of a protein.
* Sequence differences accumulate gradually so there is a positive correlation between the number of differences between two species and the time since they diverged from a common ancestor.
* Traits can be analogous or homologous.
* Cladograms are tree diagrams that show the most probable sequence of divergence in clades.
* Evidence from cladistics has shown that classifications of some groups based on structure did not correspond with the evolutionary origins of a group or species.

**Applications and skills:**

* Application: Cladograms including humans and other primates.
* Application: Reclassification of the figwort family using evidence from cladistics.
* Skill: Analysis of cladograms to deduce evolutionary relationships.