Core Category	Core Concepts in Genetics (in bold)/Example Learning Objectives
Nature of Genetic Material	**How is DNA organized?** • Describe the types of DNA regions that do not encode proteins: the general organization, possible function, and frequency of genes and non-gene DNA sequences in a typical eukaryotic genome. • Explain what is meant by single-nucleotide polymorphism (SNP) and short tandem repeat (STR), and explain how SNPs and STRs can be used as genetic markers even if they do not cause phenotypic changes. • Discuss how DNA is packaged in the chromosomes in terms of histones, nucleosomes, and chromatin • Explain the meaning of ploidy (haploid, diploid, aneuploid etc.) and how it relates to the number of homologues of each chromosome. • Describe how the positions of individual genes on a given chromosome are related to their positions on the homolog of that chromosome. • Differentiate between a gene and an allele, including the recognition that genes may have many alleles. • Explain the functional significance of packaging DNA into chromosomes and the lack of correlation between chromosome size and genetic information content.
Nature of Genetic Material	**What are the molecular components and mechanisms necessary to preserve and duplicate an organism's genome?** • Draw a simple line diagram showing a segment of DNA from a gene and its RNA transcript, indicating which DNA strand is the template, the direction of transcription and the polarities of all DNA and RNA strands. • Describe the process of mitosis, transcription, and translation. How are mistakes in these processes identified and corrected?

Core Concepts and Competencies in Genetics. Revised and approved by the Genetics Society of America Education Committee in November 2015

Transmission/Patterns of Inheritance	**What are the mechanisms by which an organism's genome is passed on to the next generation?** • Distinguish between somatic and germline cells; listing similarities and differences. • Compare and explain the inheritance of germline and somatic mutations. • Describe, using diagrams, the sequence of events involving DNA in meiosis from chromosome duplication through chromosome segregation. Explain how meiosis is different from mitosis. • Describe the difference between meiosis in mammalian males and females. • Distinguish between sister chromatids and homologous chromosomes. • Discuss how errors in chromosome number can arise during meiosis, and why such alterations can be detrimental • Calculate the probability of a particular gamete being produced from an individual, assuming independent segregation. • Calculate the probability of a particular genotype, given independent segregation and random union of gametes between two individuals. • Contrast the mechanisms of inheritance of nuclear and organellar genetic information • Explain how independent assortment of alleles during meiosis can lead to new combinations of alleles of unlinked genes.
Transmission/Patterns of Inheritance	**How can one deduce information about genes, alleles, and gene functions from analysis of genetic crosses and patterns of inheritance?** • Interpret pedigree information to determine the suitability of a DNA marker for tracking a disease trait in a family. • Draw a pedigree based on information in a story problem. • Using pedigrees, distinguish between dominant, recessive, autosomal, X-linked, and cytoplasmic modes of inheritance. • Predict the transmission of phenotypes associated with maternal effect genes. • Explain why the terms "dominant" and "recessive" are context dependent and may

differ at the cellular level or at the level of a pedigree.
- Calculate the probability that an individual in a pedigree has a particular genotype (using Bayesian inference if appropriate for course).
- Design genetic crosses to provide information about genes, alleles, and gene functions.
- Interpret the results of experiments comparing the phenotypes that result from single mutations in two different genes with the phenotype of the double mutant, contrasting epistatic and additive interactions.
- Explain how continuous traits are the result of many different gene combinations that can each contribute a varying amount to a phenotype.
- Evaluate how genes and the environment can interact to produce a phenotype.

Transmission/Patterns of Inheritance

How does the phenomenon of linkage affect the assortment of alleles during meiosis?
- Use statistical analysis to determine how well data from a genetic cross or human pedigree analysis fits theoretical predictions including an explanation of the appropriate statistical test.
- Explain the meaning of a LOD score.
- Diagram the process of homologous recombination during meiosis and explain how it can lead to new combinations of linked alleles.
- Explain the role of homologous recombination in ensuring proper segregation of homologs in meiosis I
- Explain how a specific combination of linked alleles (haplotype) can persist through many generations (linkage disequilibrium).
- Calculate gene linkage and genetic map distances and interference from the frequencies of progeny with recombinant phenotypes from genetic crosses.
- Explain how genetic distance is different from physical distance.
- Calculate the probability of a particular gamete being produced from an individual, provided map distance.

Molecular Biology of Gene Function

How is genetic information expressed so it affects an organism's structure and function?
- Explain how the genetic code relates transcription to translation
- Describe how expansion or retraction of triplet repeats can alter gene function and create a phenotype
- Discuss how various factors might influence the relationship between genotype and phenotype (e.g. incomplete penetrance, variable expressivity, and sex-limited phenotype)
- Explain how abnormalities in gene dosage can affect phenotype.

Gene Expression and Regulation

How can gene activity be altered in the absence of DNA changes?
- Discuss the roles of types of RNA other than mRNA in expressing genetic information
- Defend how most cells can have the same genetic content and yet have different functions in the body.
- Contrast the packaging of DNA into euchromatin versus heterochromatin in the context of histone modification, and DNA modification (where applicable)
- Discuss the potential roles of DNA modification, histone modification, and non-coding RNA in epigenetic inheritance, both somatic and germline
- Discuss environmental impacts on epigenetic systems

Gene Expression and Regulation

How do genes and genomes control changes in an organism's structure and function throughout its life cycle?
- Describe how differential histone modification modulates gene activity and is utilized in developmental progression
- Use a model systems to describe investigations of evo-devo
- Describe genetic cascades; use the sex-determination cascade to explain how differential gene expression can result in the development of different sexes.
- Explain how polarity is established in a developing embryo using gene expression gradients.

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Genetic Variation

How do different types of mutations affect genes and the corresponding mRNAs and proteins?

- Describe how duplications, deletions, inversions, and translocations can affect gene function, gene expression, and genetic recombination. Describe the same for transposable elements
- Describe how mutations arise and how environmental factors can increase mutation rate.
- Cite examples of mutations that can be beneficial to organisms.
- Interpret results from experiments to distinguish between different types of DNA rearrangements.
- Distinguish 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.
- Compare the role of both loss and gain of function mutations in the origin of tumors

Evolution and Population Genetics

What are the processes that can affect the frequency of genotypes and phenotypes in a population over time?

- Describe the mechanisms by which variation arises and is fixed (or lost) in a population over time.
- Calculate allele frequencies based on phenotypic or genotypic data for a population, and be able to explain the assumptions that make such a calculation possible.
- Model how random mating yields predicted genotype frequencies in Hardy-Weinberg Equilibrium (HWE), and how non-random mating affects allele and genotype frequencies.
- Test whether HWE has been reached in a population.
- Explain how inbreeding increases the number of homozygotes (and possibly disease) in comparison to HWE.
- Explain how natural selection and genetic drift can affect the elimination,

maintenance or increase in frequency of various types of alleles (e.g. dominant, recessive, deleterious, beneficial) in a population.

- Interpret experiments to determine the relative influences of genes and the environment on a given phenotype.
- Describe how variation can be measured, and what can be done to distinguish genetic and environmental sources of variation.
- Interpret bioinformatics data to compare homologous genes in different species and infer relative degrees of evolutionary relatedness.
- Use comparative data from multiple species to identify which regions of a protein, pathway, regulatory system etc. are critical for function.

Genetics of Model Organisms

How do the results of molecular genetic studies in model organisms help us understand aspects of human genetics and genetic diseases?

- Justify why information on functions of human genes can often be acquired through studies of simple model organisms such as yeast, nematode worms, and fruit flies.
- Compare the benefits and limitations of using model organisms to study human genes and human genetic diseases. Identify specific cases where insights from model organisms have provided crucial insights into human disease.
- Defend the assertion that genetic testing will play a central role in the diagnosis and treatment of cancer in the future.

Methods and Tools in Genetics

What experimental methods are commonly used to analyze gene structure, gene expression, gene function, and genetic variants?

- Explain reverse genetics and compare methods for generating specific mutations in the genome vs. generating phenocopies using techniques such as RNAi or morpholinos.
- Explain the method of SNP/STR mapping and interpret SNP/STR mapping data to pinpoint the chromosomal location of a human disease gene.
- Interpret complementation tests, including an assessment of the molecular interactions that might yield the results observed.

Core Competencies

Students should be able to locate, read, and comprehend primary literature research papers on genetics topics.

Students should be able to implement observational strategies to formulate a question.

Students should be able to generate testable hypotheses.

Students should be able to design an experiment using appropriate controls and appropriate sample sizes.

Students should be able to gather and evaluate experimental evidence, including qualitative and quantitative data.

Students should be able to apply statistical methods when analyzing their data, and use patterns to construct a model.

Students should be able to generate and interpret graphs displaying experimental results.

Students should be able to critique large data sets and use bioinformatics to assess genetics data.

Students should be able to communicate experimental results effectively, including writing research papers and giving presentations.

Students should be able to effectively explain genetics concepts to different audiences.

Students should be able to tap into the interdisciplinary nature of science.

Students should be able to identify and critique scientific issues relating to society or ethics.