1. **MUTATION: SELECTION BALANCE.** Genetic diseases arise from the segregation of deleterious alleles. These are usually recessive, but in rare cases they are co-dominant or dominant.

Consider a hypothetical genetic variant, "a", which researchers find is associated with reduced rates of infant survival compared to individuals with the wildtype "A" allele. Studies show that individuals that are homozygous for this allele have only 70% the normal chance of surviving until adulthood. Heterozygotes are unaffected.

(a, 3 pts) Assuming a mutation rate of $10^{-6}$ for this genetic disorder (approximately correct for single locus mutation rates), what is the predicted frequency of this allele in the population? (Provide answer to the 3 significant figures)

\[ f(a) = 1.83 \times 10^{-3} \]

(b, 3 pts) The population of the US is approximately 300,000,000. Assuming this, what is the expected number of afflicted individuals in the US? (round to the nearest integer)

\[ \# \text{ sick} = 1,000 \]

Now consider the situation in which this deleterious allele were dominant instead.

(c, 3 pts) The population of the world is approximately 6,000,000,000. Assuming this and dominance, what is the expected number of afflicted individuals in the world? (round to the nearest integer)

\[ \# \text{ sick} = 40,000 \]

(d, 3 pts) What does your result imply about the observed prevalence of dominant genetic diseases?

Although their allele frequencies are much lower ($3.33 \times 10^{-6}$ vs $1.58 \times 10^{-3}$), the frequencies of sick individuals are roughly similar

\[
\frac{1,000}{300,000,000} = 3.33 \times 10^{-6} \quad \text{vs} \quad \frac{40,000}{6,000,000,000} = 6.67 \times 10^{-6}
\]

\(\text{(A)}\)

\[ q_{\text{eq}} = (10^{-6}/0.3)^{1/2} = 0.000183 = 1.83 \times 10^{-3} \]

\(\text{(B)}\)

\[ 3 \times 10^8 \times q_{\text{eq}}^2 = 3 \times 10^8 \times (1.83 \times 10^{-4})^2 = 1,000 \]

\(\text{(C)}\)

\[ q_{\text{eq}} = (10^{-6}/0.3) = 3.33 \times 10^{-6} \quad 6 \times 10^9 \times 2 \times p_{\text{eq}} \times q_{\text{eq}} = 6 \times 10^9 \times 2(1-3.33 \times 10^{-6})(3.33 \times 10^{-6}) = 40,000 \]
2. EFFECTIVE POPULATION SIZE. (provide answers to nearest 0.01)

(a, 2 pts) What is the mean census population size of a population that cycles between 300, 400 and 500 members on alternate years? N= ______ 400.00

(b, 4 pts) What is the effective population size of a population that cycles between 300, 400 and 500 members on alternate years? Ne= ______ 382.98

(c, 2 pts) What is the census population size of a population that has 300 males and 800 females? N= ______ 1100

(d, 4 pts) What is the effective population size of a population that has 300 males and 800 females? Ne= ______ 872.73

3. EVOLUTIONARY RATES. (provide answers to nearest 0.0001)

Consider a situation in which the homozygote for an advantageous allele (AA) experiences a 4% fitness advantage over the other homozygote (aa) and the frequency of the advantageous allele is p=0.2.

(a, 3 pts) If the "A" allele is dominant, what is the frequency of the allele in the next generation? p’ = _0.2050_

(b, 3 pts) If the "A" allele is co-dominant (h=1/2), what is the frequency of the allele in the next generation? p’ = _0.2032_

(c, 3 pts) If the "A" allele is recessive, what is the frequency of the allele in the next generation? p’ = _0.2013_
4. POPULATION GENETICS.

Consider two populations of the same species which are separated for a time and very little migration occurs between them; they do still easily interbreed and are not becoming distinct species however.

We will consider co-dominant alleles at a locus that determines pelage color in which the allele “A” causes darker coloration: homozygous AA individuals are black, heterozygous individuals are brown, and homozygous aa individuals are pale.

Within each population the frequencies of the genotypes are as expected under Hardy-Weinberg equilibrium. The figure shows the number of individuals of each phenotype.

(a, 1 pt each) What are p and q for each population? (provide answers to nearest 0.001)

Population 1: p = _0.300_ q = _0.700_

Population 2: p = _0.600_ q = _0.400_

(a, 1 pt each) If were to combine all this data and calculate the allele frequencies, what would p and q be for the overall population? (provide answers to nearest 0.001)

Overall population: p = _0.500_ q = _0.500_

(c, 1 pt each) Pooling the data as we just did gives us total observed numbers for each phenotype. In the table to the right, fill in these values. Using the allele frequency data for the combined population we can also predict these numbers assuming Hardy-Weinberg equilibrium (which is true for each separate population). In the table to the right, fill in what we expect these two sets of values to be. (provide answers to nearest 0.001)

<table>
<thead>
<tr>
<th>Observed</th>
<th>Expected</th>
</tr>
</thead>
<tbody>
<tr>
<td>f(AA) = <em>648</em></td>
<td>f(AA) = <em>600</em></td>
</tr>
<tr>
<td>f(Aa) = <em>1104</em></td>
<td>f(Aa) = <em>1200</em></td>
</tr>
<tr>
<td>f(aa) = <em>648</em></td>
<td>f(aa) = <em>600</em></td>
</tr>
</tbody>
</table>

(d, 3 pts) If you did this correctly you will notice that the observed and predicted numbers don’t match. The Wahlund effect is the name for this mismatch and may arise when subpopulations are pooled and their combined data is treated as if the separate populations were a single panmictic population. The Wahlund effect can be mistaken for a different process involving non-random mating however. Just seeing the pattern of mismatch between the observed and predicted phenotype frequencies caused by the Wahlund effect has an alternative non-geographic explanation. What is the important pattern and what type of mating may be easily confused with the presence of relatively isolated subpopulations?

The smaller number of heterozygotes compared to the expected number is consistent with inbreeding or positive assortative mating.
5. MOLECULAR PHYLOGENETICS & THE MOLECULAR CLOCK. Consider four taxa (argostics, blarneys, critters, dermestoads) where argostics and blarneys are a pair of sister taxa and critters and dermestoads are a pair of sister taxa. Root the tree you will draw below accordingly.

Consider their aligned DNA sequences:

<table>
<thead>
<tr>
<th>Taxon</th>
<th>Sequence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Arg</td>
<td>ACTTCGATAGCTAGATGCTAGATGCTAGATGCTAGACCAGATAGCCAGAGAACTAGACTAG</td>
</tr>
<tr>
<td>Bla</td>
<td>........C........C........G........T............</td>
</tr>
<tr>
<td>Cri</td>
<td>.AA......A........G...T........T........C..G.</td>
</tr>
<tr>
<td>Der</td>
<td>........A..A........G...T...A......T.....A.....G.</td>
</tr>
</tbody>
</table>

(a, 2 pts ea) How many transitions and transversions appear to have occurred during the history of these taxa?

# Transitions = ________ 3
# Transversions = _____ 10

(b, 4 pts) In the box, draw the phylogeny of these taxa based on the information described above. The scale indicates each substitution, draw branch lengths accurately with the root on the left side and taxa on the right.

(c, 2 pts) Assume a molecular clock. If the speciation event that split argostics and blarneys occurred 20 million years ago, how long ago did the speciation event that separated critters and dermestoads occur?

____ 30 ____ MYA

(c, 2 pts) Assume a molecular clock. If the speciation event that split argostics and blarneys occurred 20 million years ago, how long ago did the speciation event that separated blarneys and critters occur?

____ 40 ____ MYA
FOR THE REMAINING QUESTIONS USE YOUR SCANTRON FORM,

► MULTIPLE CHOICE: (2 pts each).

(1) Which type of genetic data is the worst to use for distinguishing individuals within a population from one another?
(A) Conservative amino acid polymorphisms.
(B) Frameshift polymorphisms.
(C) Microsatellite repeat variants.
(D) Polymorphisms at wobble positions.
(E) Polymorphisms in introns.

(2) Which of the following terms refers to an amino acid substitution that has a major effect on the size or chemical properties of the amino acid residue?
(A) Conservative
(B) Nonsynonymous
(C) Radical
(D) Silent
(E) Synonymous.

(3) When a nucleotide change in a codon within an exon alters a single amino acid it is termed a(n) ______ mutation.
(A) frameshift
(B) indel
(C) nonsense
(D) nonsynonymous
(E) synonymous

(4) Which of the following causes inbreeding depression?
(A) Inbreeding leads to increased rates of deleterious mutations in populations which lowers the mean fitness.
(B) In inbred populations, for a given allele frequency of a deleterious allele, there are more individuals homozygous for that allele.
(C) In inbred populations, the allele frequencies of deleterious alleles are higher due to increased homozygosity.
(D) Mating with relatives causes genetic incompatibilities that lead to deleterious genotypes.
(E) Mating with relatives too much makes animals sad.

(5) Which of the following is the best description of negative assortative mating?
(A) Individuals preferentially mate with others having different genotypes.
(B) Individuals preferentially mate with others having similar genotypes.
(C) Some genotypes are not preferred by any other genotypes.
(D) This type of mating leads to higher frequencies of homozygotes than predicted by H-W equilibrium calculations.
(E) This type of mating leads to lower frequencies of heterozygotes than predicted by H-W equilibrium calculations.

(6) How many generations does it take for a neutral allele to increase from being present in a single heterozygous mutant to becoming the standard wildtype state present in all homozygous individuals in a population of size 3,000?
(A) 3,000 generations.
(B) 6,000 generations.
(C) 12,000 generations.
(D) 6,000 years.
(E) 12,000 years.

(7) Which of the following was not a type of genetic variance discussed when examining phenotypic variance?
(A) Additive
(B) Dominance
(C) Epistatic
(D) Environmental
(E) Total
One of the processes we examined causes genotype frequencies to change consistently while allele frequencies do not. Although the population therefore changes over time, by our restrictive definition of evolution as allele frequency change the population does not "evolve". What process was this?

(A) Epistasis  (B) Genetic drift  (C) Inbreeding  (D) Perturbations

Which of the following best describes what the effective population size represents?

(A) How fluctuating population sizes change the chances of alleles persisting.
(B) The minimum population size needed to effectively maintain genetic diversity.
(C) The number of individuals required in order for the population to evolve effectively instead of going extinct.
(D) The size of an ideal panmictic population that would maintain heterozygosity for as long as the population we are considering.
(E) The total number of adults in a population, not including juveniles since they don’t reproduce.

Fisher’s fundamental theorem is best paraphrased by which of the following?

(A) Higher levels of genetic variation lead to faster rates of evolution.
(B) Higher levels of phenotypic variation lead to faster rates of evolution.
(C) Higher rates of immigration lead to faster rates of evolution.
(D) Higher rates of mutation lead to faster rates of evolution.
(E) Larger population sizes lead to faster rates of evolution.

Which of the following pairs of statements is most CORRECT?

(A) Evolution is development over long time scales; substitution is mutation over long time scales.
(B) Individuals develop and have mutations while populations evolve and have substitutions
(C) Individuals develop and have substitutions while populations evolve and have mutations
(D) Individuals evolve and have mutations while populations develop and have substitutions
(E) Individuals evolve and have substitutions while populations develop and have mutations

For the next two questions consider a large mainland population with two alleles, Bt and Hk, at a locus which are present in equal frequency in a large mainland population.

This mainland begins to send migrants to a nearby island on which the Bt allele is present at 25% of the loci. If after the first generation of this process the new frequency of the Bt allele on the island is 26.5%, which of the following values is closest to the immigration rate to the island?

(A) 0.015%  (B) 1.5%  (C) 3%  (D) 4.5%  (E) 6%

If this migration process were to occur for 6 generations, which of the following values is closest to the frequency of the Bt allele on the island after that amount of time?

(A) 29.2%  (B) 32.8%  (C) 39.5%  (D) 42.3%  (E) 45.8%
For the next two questions consider a cross between an AABbccDD and an aaBbCCdd individual where the alleles at the four loci influence a quantitative trait.

(14) How many different genotypes can result from the cross?
(A) 1    (B) 2    (C) 3    (D) 4    (E) 5

(15) Assume that the effects of the different alleles at each locus are identical with regard to increasing or decreasing the trait and that capital letters indicate alleles dominant to lower case ones. How many different phenotypes can result from the cross?
(A) 2    (B) 3    (C) 4    (D) 5    (E) 6

For the next two questions consider a cross between an AaBbCcDd and an AaBbCcDd individual where the alleles at the four loci influence a quantitative trait.

(16) How many different genotypes can result from the cross?
(A) 4    (B) 16    (C) 36    (D) 81    (E) 96

(17) Assume that the effects of the different alleles at each locus are identical with regard to increasing or decreasing the trait and that capital letters indicate alleles dominant to lower case ones. How many different phenotypes can result from the cross?
(A) 1    (B) 2    (C) 3    (D) 4    (E) 5

The following 3 questions are based upon the videos you watched in preparation for this exam.

(18) Which of the following best summarizes the video.
(A) It discusses genotypic, but not phenotypic, about the color evolution of mice living in light and dark environments.
(B) It discusses phenotypic, but not genotypic data, about the color evolution of mice living in light and dark environments.
(C) It discusses both phenotypic and genotypic data about the color evolution of mice living in light and dark environments.
(D) It discusses how color patterns in mice are related to their palatability to predators.
(E) It discusses how migration between different regions can influence color patterns in mice.

(19) The locus identified as important for the color patterns differences is named which of the following?
(A) LD3X    (C) Melanocortin    (E) Xerxes
(B) Mc1R    (D) Melanoleukin

(20) The researchers also studied other mouse populations in similar environments; what was the interesting result of this?
(A) Despite the same conditions, those populations didn't evolve.
(B) The colors patterns evolve the same way, but via mutations in different genes.
(C) The colors patterns evolve the same way, via mutations in the exact same genes.
(D) There was conclusive evidence of migration.
(E) There was conclusive evidence of a lack of migration.