

1) Briefly (i.e., in a single sentence) define in words all of the following and be very specific. (20 points)

- a) Allele – **alternate form of a gene.**
- b) Evolution – **change in allele or character frequency over time**
- c) Additive Relationship Between Genotype and Phenotype – **Heterozygote phenotype is intermediate between the homozygotes.**
- d) Genetic Drift – **stochastic or random change in allele of character frequency due to sampling errors in finite populations**
- e) Relative Fitness – **The fitness of individual phenotypes relative to the most fit phenotype.**
- f) Transversion – **a point mutation from a purine to a pyrimidine or visa versa**
- g) Allozygous alleles – **two alleles that are identical by state but not descent.**
- h) Homology – **characters that share a common evolutionary origin**
- i) Inbreeding Depression – **the reduction in fitness that accompanies inbreeding and the subsequent expression of recessive, deleterious alleles while in the homozygous state.**
- j) Linkage Disequilibrium – **the observed association of alleles at two loci not in proportion to what is expected based on the independent allele frequencies.**

2 pts each

20 Points

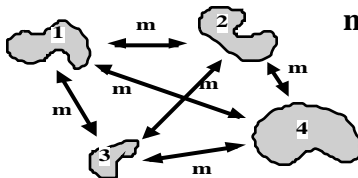
2) Mutation is the primary mechanism by which new variation enters natural populations. In class, I discussed several types of mutations. Name and describe 4 of the 6 types of POINT MUTATIONS that I covered (8 points)

2 pts for 4

- a) **frame shift** – the alteration of the reading frame of a exon due to insertions or deletions. Usually has a large effect (i.e., loss of function).
- b) **Transitions** – the substitution of a purine with a purine or a pyrimidine with a pyrimidine.
- c) **Transversion** – the substitution of a purine with a pyrimidine and visa versa
- d) **synonymous** – substitutions in a coding region that do not change the amion acid specified.
- e) **nonsynonymous** – substitutions in a coding region that do change the amion acid specified.
- f) **nonsense** – substitutions in a coding region that change an amion acid codon to a stop (nonsense) codon.

3) Diagram (i.e., draw a picture of) each migration model listed below. Describe in detail how migration links the populations (9 points)

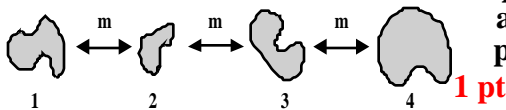
- a) Island Model — **migration is from every population to every other population. Individuals move from population 1 to population 2 as easily as they move from 1 to 3 or any other combinations of populations.**



1 pt

2 pts

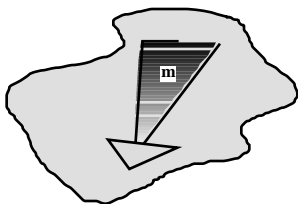
- b) Stepping Stone Model — **each population receives migrants from neighboring populations. The movement of individuals between adjacent populations is higher than between nonadjacent populations**



1 pt

2 pts

- c) Isolation by Distance Model — **in a continuously distributed population, gene flow occurs among local neighborhoods. The frequency of movement between proximate parts of the populations is higher than between distal parts of the population.**



1 pt

2 pts

4) What are the 5 Hardy-Weinberg assumptions? (10 points)

- 2 pts ea**
- infinite population size**
 - random breeding with respect to genotype**
 - no mutation**
 - no migration (immigration or emigration)**
 - no selection**

Genotype	Number Observed
A_1A_1	700
A_1A_2	110
A_2A_2	190
Total	1,000

f) What are the frequencies of the genotypes in the population [i.e., $F(A_1A_1)$, $F(A_1A_2)$, and $F(A_2A_2)$]?

1 pt $F(A_1A_1) = \frac{700}{1000} = 0.700$

1 pt $F(A_1A_2) = \frac{110}{1000} = 0.110$

1 pt $F(A_2A_2) = \frac{190}{1000} = 0.190$

g) What are the allele frequencies in the population [i.e., $F(A_1)$ and $F(A_2)$]?

1 pt $F(A_1) = \frac{700 + 55}{1000}$ or $\frac{1400 + 110}{2000}$ or $0.700 + 0.055 = 0.755$

1 pt $F(A_2) = \frac{190 + 55}{1000}$ or $\frac{380 + 55}{2000}$ or $(0.190 + 0.055)$ or $(1 - 0.755) = 0.245$

h) Is the population in Hardy – Weinberg equilibrium? How could you test this?

1 pt $F_{exp}(A_1A_1) = 0.755^2 = 0.57025$

1 pt $F_{exp}(A_1A_2) = 2 \times 0.755 \times 0.245 = 0.36995$

1 pt $F_{exp}(A_2A_2) = 0.245^2 = 0.060025$

Genotype	Number Observed	Number Expected	Difference
A_1A_1	700	570.025	+129.975
A_1A_2	110	369.950	-259.950
A_2A_2	190	60.025	+129.975
Total	1,000		

1 pt **NO, this population is probably not in HWE. As shown in the table above, there clearly are too many homozygotes and too few heterozygotes observed. A χ^2 test will detect significance where**

1 pt
$$\chi^2 = \frac{(\text{observed} - \text{expected})^2}{\text{expected}}$$

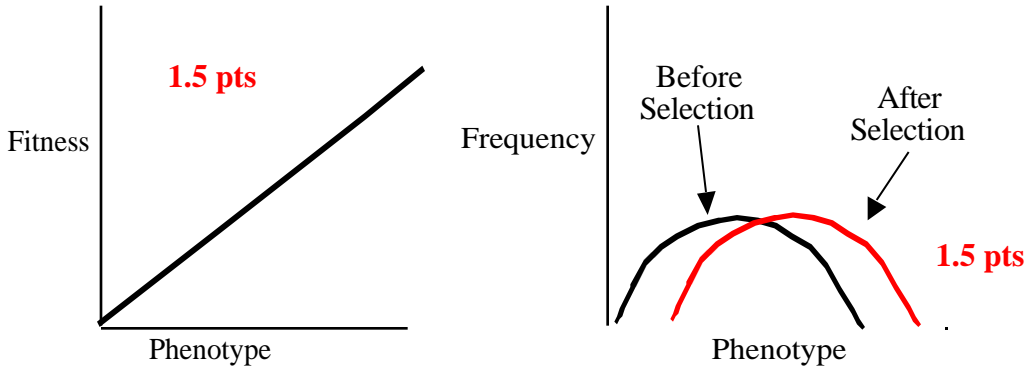
i) Describe a situation that might have resulted the observed genotype frequencies?

2 pts **This is likely a case of underdominant selection, positive assortative mating or inbreeding because there are too many homozygotes and too few heterozygotes. It is possible that selective migration of the heterozygotes is possible, but unlikely to produce such a large deviation.**

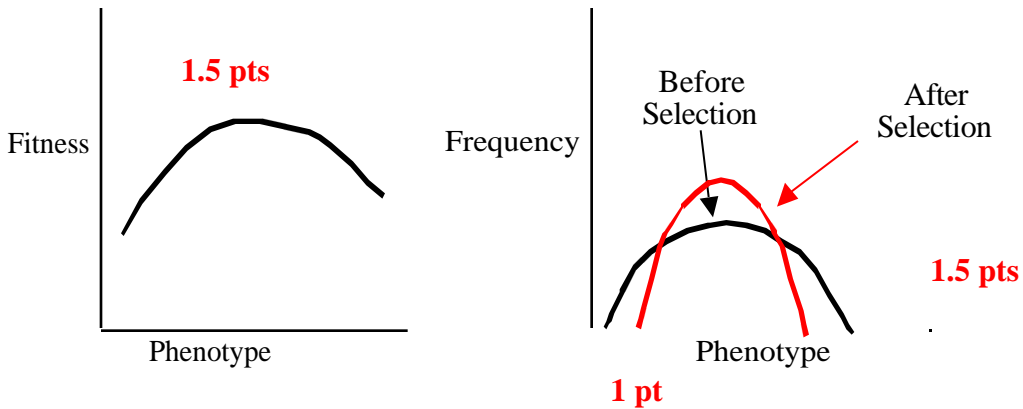
22 Points

- 5) There are three general ways that selection can act on phenotypic variation. In the graph axes below,
 a) Name each type of selection (3 points).
 b) Draw the appropriate fitness functions (4.5 points).
 c) Draw the phenotypic frequency distribution after selection (i.e., how does the curve change?).
 (4.5 points).

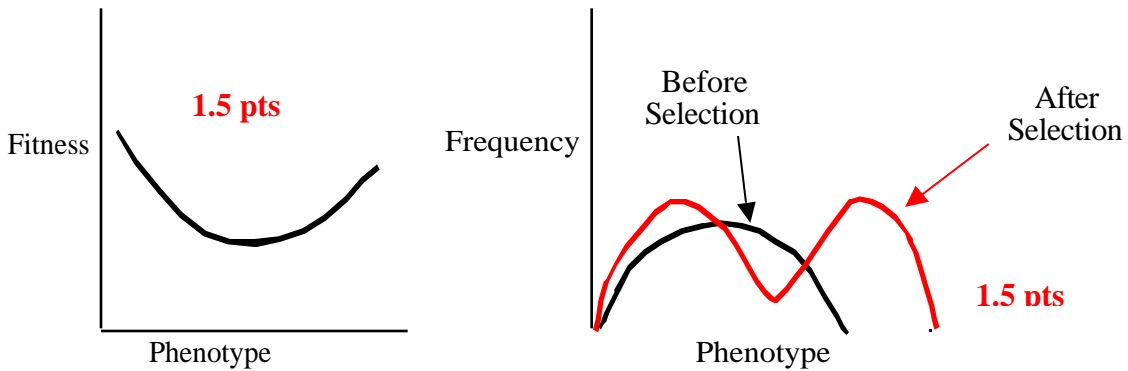
Type of Selection: Directional **1 pt**



Type of Selection: Stabalizing/overdominance **1 pt**



Type of Selection: Disruptive/underdominance



12 points

6) When considering the Neutral Theory of Molecular Evolution and the change in frequency of a strictly neutral allele over time (i.e., generations):

a) What is the rate of neutral evolution? (3 points)

3 pts $k = \mu = \text{mutation rate}$

b) How long does it take this allele to become fixed? (3 points)

3 pts $4N_e$ where N_e is the genetically effective population size.

c) Does the time in question b, above, depend on the population size? How and why or why not? (3 points)

3 pts Yes, drift is faster in smaller population and slower in large populations because the effect of missed sampling of one or even a few individuals is proportionally larger in smaller populations than in large.

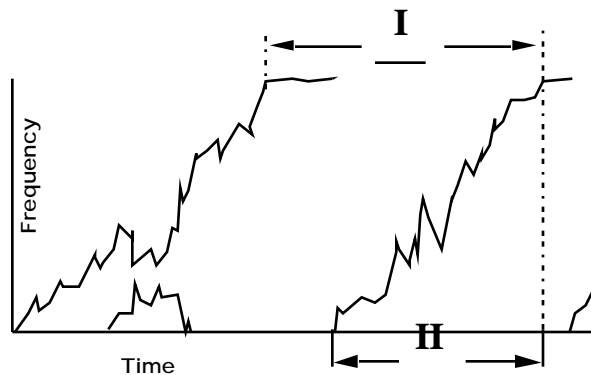
The rate of neutral evolution does not change with population size (i.e., $k = \underline{2N_e\mu} * (\underline{1/2N_e}) = \mu$)

d) Explain why this is true by describing in words the meaning of the underlined variables in the equation above (i.e., what do $\underline{2N_e\mu}$ and $\underline{1/2N_e}$ represent and why?). (3 points)

1 pt $2N_e\mu$ — in a population there are $2N_e$ number of alleles and each one of them can mutate at a rate of μ . That means that new alleles are being generated by mutation at a frequency of $2N_e\mu$.

1 pt $\frac{1}{2N_e}$ — This is the frequency of a new allele in the population. This is also the probability that this allele will go to fixation.

1 pt Taken together, the rate at which new mutation go to fixation in a population is the combined probabilities of them being generated and the probability of fixation or $2N_e\mu * 1/2N_e$



7) Considering a population that is in Hardy-Weinberg Genotype Frequency Equilibrium at birth, answer the following questions.

a) For Overdominance Selection (aka Stabilizing or balancing selection), fill in the following table with the appropriate variables. (4 points).

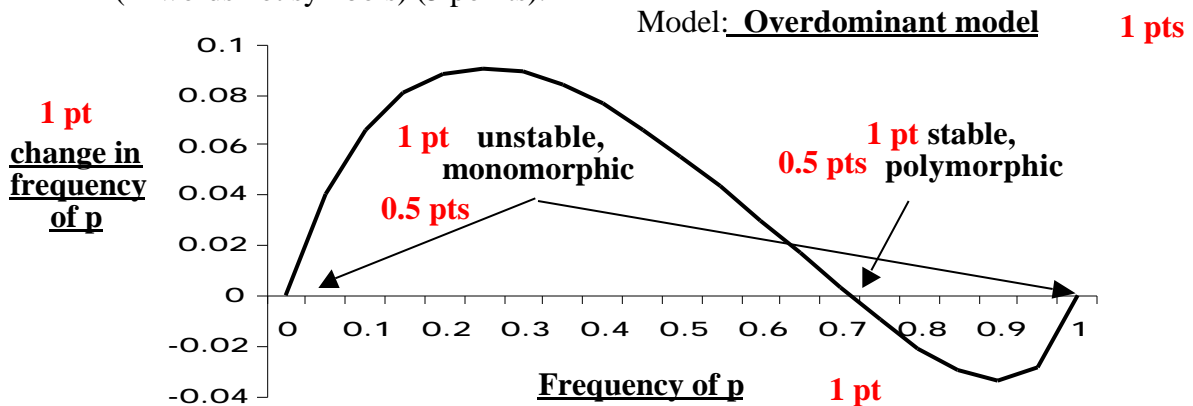
Genotype		<u>AA</u>	<u>Aa</u>	<u>aa</u>
2 pts	Frequency at Birth	p²	2pq	q²
2 pts	Relative Fitness	1 + s	1	1 + t

s and t above are the selection coefficients and are negative

What are the equilibrium allele frequencies? (2 points)

1 pt $\hat{p} = \frac{t}{s+t}$, **0.5 pts** $\hat{p} = 0.0$, and **0.5 pts** $\hat{p} = 1.0$

For the graph below, what model of selection does it describe? Please label ALL equilibrium points. Indicate whether these points are stable or unstable and whether the population is polymorphic or monomorphic. Be sure to label the axes of the graph (in words not symbols) (5 points).



b) For Directional Selection with dominant, deleterious allele “a”, fill in the following table with the appropriate variables (4 points)

Genotype		<u>AA</u>	<u>Aa</u>	<u>aa</u>
2 pts	Frequency before Selection	p²	2pq	q²
2 pts	Relative Fitness	1	1 - s	1 - s

What are the equilibrium allele frequencies? (2 points)

2 pts $\hat{p} = 0.0$ and $\hat{p} = 1.0$