

BIOL 403 EVOLUTION Exam 2 Spring 08

- 1 (12 pts). List four assumptions of the Random Mating (Hardy-Weinberg) Model. For each assumption, give an example of an evolutionary model developed in lecture which relaxes or removes this assumption.

Assumption	Model
1. No selection	HW model plus selection
2. No mutation	selection-mutation balance model drift-mutation balance
3. No migration	island migration model, migration- selection balance
4. Infinite pop'n size (No chance change in allele frequency)	Genetic drift, drift-mutation balance

2. (4 pts) The Hardy-Weinberg model does not allow evolution (of allele frequency). What good is a population genetic model without evolution? List one application of the H-W model.

As a null model

3. (8 pts) Given the following (human) genotype frequencies for a genetic polymorphism affecting resistance to HIV infection, is there significant evidence for deviation of genotype frequencies from random mating expectations? Show your work.

# Obs'd	Genotype		
	+/+	+/ Δ 32	Δ 32 / Δ 32
	80	20	0

	Exp'd #s	Obs'd #s
$p^2 = 0.81$	81	80
$2pq = 0.18$	18	20
$q^2 = 0.01$	1	0

chi -square (1 df) = 1.23, N.S.

The genotype frequencies do not deviate significantly from expectation assuming random mating.

4. (4 pts) Does this example resemble any real world population in terms of genetic variation for the Δ 32 allele? If so, which one?

Yes, a northern European population.

5. (4 pts) For genotypes with the following fitnesses and frequencies at birth

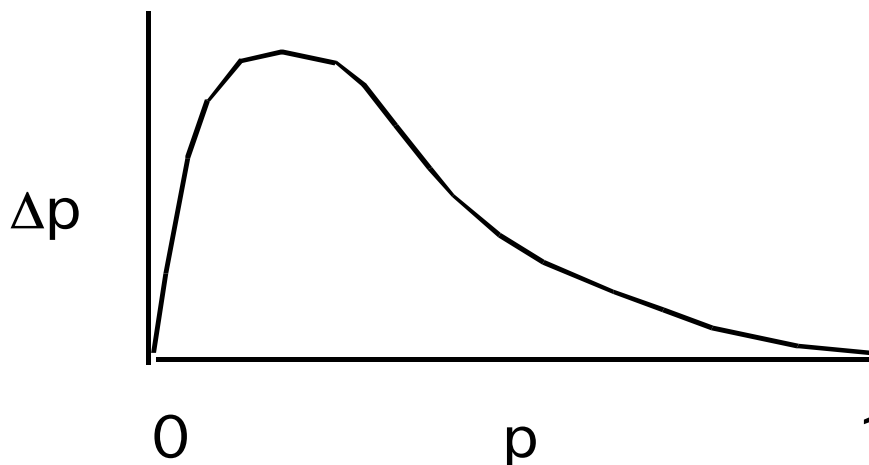
Genotype	A_1A_1	A_1A_2	A_2A_2
Birth Freq'cy	p^2	$2pq$	q^2
Fitness	1	1	$1-s$

Write down an expression for the frequency of the A_1 allele after selection:

$$\text{Freq}(A_1) = \frac{(p^2 + pq)}{[p^2 * 1 + 2pq * 1 + q^2(1-s)]}$$

$$= \frac{p}{(1-sq^2)}$$

6. (4 pts) The plot below depicts the rate of progress of directional selection for a dominant favored allele with frequency p , as a function of the allele frequency. Explain the leftward skew of the plot.



Selection is less effective for $p > 0.5$, because the disfavored allele is found disproportionately in heterozygotes of high fitness.

7. (4 pts) Assuming overdominant selection where the fitnesses of the three genotypes A_1A_1 , A_1A_2 and A_2A_2 are 0.8, 1 and 0.2 respectively, calculate the average fitness of the population when $p = f(A_1) = 0.8$.

$$\bar{w} = 0.64 * 0.8 + 0.32 * 1 + 0.04 * 0.2 = 0.84$$

8. (4 pts) Refer to the preceding problem. For $p = f(A_1) = 0.8$, calculate the average excess of the A_1 allele in this case as $p = w_{11} + q w_{12} - \bar{w}$ (where \bar{w} is the average fitness of the population). Explain your result; what is the utility of this equation?

$$\text{Average excess of } A_1 \text{ allele} = 0.8 * 0.8 + 0.2 * 1 - \bar{w} = 0$$

The average excess of an allele indicates whether the allele frequency increases, decreases or stays the same. In this case, the allele frequency is not changing because it maintained at a stable intermediate value by balancing selection at $p = 0.8$

9. (4 pts) The maintenance of the HbA/HbS polymorphism is due to overdominant selection for resistance to malaria. The HbC allele provides superior protection against malaria in the homozygous state. Nevertheless, the HbC allele is rare in populations in which the HbS allele is common. Conversely, in other populations the HbC allele is common when the HbS is rare. Assuming that these observations represent evolutionary equilibria, explain both observations.

The HbC allele cannot increase in populations in which the HbS allele is common (chiefly because it often occurs in HbS/HbC heterozygotes of low fitness).

Therefore, in populations in which the HbC allele is common, the HbS allele must have been missing from the population initially.

10. (6 pts) List three evolutionary forces other than selection, and characterize each one as strong or weak, in terms of its potential to change allele frequencies.

Genetic drift weak

Mutation weak

Migration strong

11. (6 pts) Cystic fibrosis is an example of a recessive genetic disease in humans whose frequency was not found to be consistent with selection-mutation balance. List the (three) pieces of information needed to make this assessment.

- 1. incidence of the disease in the general population**
- 2. An independent estimate of the actual mutation rate**
- 3. Estimate of the fitness effects (s) of the mutation in the recessive homozygote**

12. (6 pts) Recall our discussion of gene flow between racial groups in Claxton, Georgia. Draw a picture of the island model of migration. Why was the use of the island model of migration appropriate in this case?

The island model assumes immigration into the focal population only.

This is a reasonable model of gene flow in Claxton, given that individuals of mixed race were treated as individuals of the African American population.

13. (4 pts) Migration is sometimes referred to as the glue that holds species together. Why?

Because migration tends to make populations more similar than they would be otherwise, in terms of their allele frequencies.

14. (4 pts) For a population containing 100 individuals where the frequency of two alternative alleles A_1 vs. A_2 is $p = q = 1/2$, describe the most likely state(s) of the population after 1000 generations when genetic drift is the only evolutionary force acting.

Fixed for either A_1 or A_2 .

15. (6 pts) Explain why we think genetic drift accounts for the prevalence of Achromatopsia in Pingelap Islanders. Why don't we think selection is important? Why not mutation?

Achromatopsia is a severe form of colorblindness, which is unlikely to have been favored by natural selection.

Mutation is a weak force, and the observed increase in frequency of the allele is much greater than could be accounted for by this mechanism.

Finally, it is known that the population experienced a severe reduction in population size in the past, which is expected to greatly increase the importance of drift in changing allele frequencies.

16. (4 pts) What (if anything) does the Neutral Theory assume about the relative prevalence of advantageous, disadvantageous and neutral mutations?

That the neutral mutation rate is \gg advantageous mutation rate

Deleterious mutations are assumed not to contribute to the substitution rate under the Neutral Theory

17. (4 pts) What is the most likely explanation for the observation of a dN/dS ratio close to 1 for a gene coding sequence (not a pseudogene)?

A mixture of negative selection acting at most (non-synonymous) sites, and positive selection acting at the remainder.

18. (4 pts) According to the Nearly Neutral Theory, would a large or small population be expected to show a higher rate of substitution *per generation* (assume the absence of positive selection)? Why?

A small population would evolve faster.

Because genetic drift is stronger in small populations, so a larger fraction of deleterious mutations ($s < 1/2N$) drift to fixation

19. (4 pts) A generalization emerging from the Nearly Neutral Theory is that small populations evolve (largely) by drift, whereas large populations evolve (largely) by selection. Provide a real world piece of evidence suggesting that genetic drift has been an important factor in the evolution of the human genome.

The human genome has experienced a much smaller proportion of adaptive substitutions compared to *Drosophila* species. This is most readily explained as a consequence of smaller (effective) population size in humans. Because drift is stronger in small populations, this makes it harder to fix adaptive substitutions.

Another example is the fact that the HbS allele is common in some human populations whereas the HbC allele is common in others. This is pattern is explained in part by the fact that in some populations the HbS allele was missing initially, probably due to drift.