ANSC 446 / IB 416 Population Genetics
Exam 2, October 23, 2009

Name _____

(5 pages) Please underline or indicate your answer. If rounding, use 3 significant digits. Show your work or describe your logic to earn partial credit for incomplete answers.

(5) 1. Assume that the initial and final allele frequencies before and after selection are 0.01 and 0.001. How many generations did it take for this amount of change to occur if the allele is a recessive lethal?

Answer: $n = (1/q_n) - (1/q_0) = (1/.001) - (1/.01) = 1000 - 100 =$ **900 generations**

(5) 2. Give an example (from nature or from human medicine) of heterozygote advantage.

Sickle cell disease. Those homozygote for the allele that causes sickle cell disease have reduced life expectancy. Yet in regions where malaria is common, heterozyotes with one copy of this allele are more resistant to malaria than wild-type homozygotes.

(20) 3. The relative fitnesses found for alleles at a locus are 1.2, 1, and 1.3 for alleles A_1A_1, A_1A_2, and A_2A_2, respectively.

(4) a. What are the values of the selection coefficients s_1 and s_2?

$s_1 = 0.2$ $s_2 = 0.3$

(4) b. What is the name given to this type of fitness array?

Heterozygote disadvantage, or underdominance

(4) c. What is the equilibrium frequency for A_2?

$q_e = s_1 / (s_1 + s_2) = 0.2 / (0.2 + 0.3) = 0.4$

(4) d. Would the equilibrium frequency be stable? Why or why not?

Not stable since fitness is at a minimum at q_e, and at local maxima when $q_e = 0$ or $q_e = 1$. Thus after perturbation from the frequency at q_e, the allele frequencies will continue to move further away from q_e in subsequent generations due to selection

(4) e. Four isolated populations have initial allele frequencies for A_2 of 0.0, 0.45, 0.50 and 1.0. At what frequency will allele A_2 stabilize in each of these four populations?

Initial A_2 of q = 0.0 will remain at 0.0
Initial A_2 of q = 0.45 will go to 1.0
Initial A_2 of q = 0.5 will go to 1.0
Initial A_2 of q = 1.0 will remain at 1.0

(14) 4. Give the best definition for the following terms:

(2) a. Gametophytic self incompatibility: Self-fertilization is prevented by requiring pollen to have a different allele from the female parent.

(2) b. Odds ratio (or risk ratio): a way of comparing whether the probability of a certain event is the same for two groups. An odds ratio of 1 implies that the event is equally likely in both groups.

(2) c. Underdominance: heterozygote disadvantage, in which either homozygote genotype has a fitness advantage over the heterozygote genotype

(2) d. Inclusive fitness: fitness of an individual plus his/her effect on the relatives of the individual weighted by their coefficient of relationship

(2) e. Antagonistic pleiotropy: negative correlation of two components of fitness; ie, a single locus affects multiple selection components in opposite directions.

(2) f. Sexually antagonistic genes: genes that have opposite selective effects in males and females.

(2) g. Fisher's fundamental theorem of natural selection: the rate of increase in fitness of any species at any time is equal to its genetic variance in fitness at that time.

(4) 5. Explain why the greatest expected change in allele frequency per generation for a recessive allele occurs around a frequency of 2/3. Explain why the greatest expected change in allele frequency for a dominant allele occurs around a frequency of 1/3?

According to Fisher's fundamental theorem of natural selection, the rate of increase in fitness of any species at any time is equal to its genetic variance in fitness at that time. When a population has two phenotypes and one is very rare

(so variance in fitness is low since almost all individuals have the same phenotype), selection can only change allele frequencies marginally.

By contrast at the indicated frequencies, the recessive phenotype would be present in $(2/3)^2 = 0.44$ of the population, while the dominant phenotype would be present in 0.56 of the population. Since about half of the population would display one phenotype while about half would display the other, variance in fitness is very high, and selection can cause a relatively large change in allele frequencies.

(10) 6. Determine the frequencies at generation t+1 for the following:

(5) a. At a biallelic autosomal locus, an additive deleterious allele with a selection coefficient of 0.3 is found at a frequency of 0.2. Assuming random mating, what will be the frequency of the allele in the next generation?

$$q_1 = \frac{q_0 [1-s(hp_0 + q_0)]}{1 - 2hsp_0q_0 - sq_0^2}$$

$= (0.2 [1 - (0.3((0.5X0.8) + 0.2))]) / [1 – (2X0.5X0.3 X0.8X0.2) -0.3((0.2)^2)]$

$=$ 0.164 / (1 - .048 - .012) $=$ **0.174**

(5) b. For the situation described in (a), and assuming the Basic Selection Model, what was the frequency of the three genotypes among only those progeny that were inviable?

Since p = 0.8 and q = 0.2, the frequency of the three genotypes among progeny before selection would be:

$Pr(A_1A_1) = P = 0.8^2 = 0.64$
$Pr(A_1A_2) = H = 2pq = 0.32$
$Pr(A_2A_2) = Q = 0.2^2 = 0.04$

The selection coefficient is 0.3, which means that 30% of the homozygotes (A_2A_2) for the deleterious allele were inviable:

0.3 X 0.04 = 0.012

The dominance for an additive allele is 0.5, which means the proportion of inviable heterozygotes is 0.5 times the selection coefficient of 0.3. Among all progeny, the proportion heterozygote (A_1A_2) and inviable is therefore:

0.3 X 0.32 X 0.5 = 0.048

For dominant homozygotes, there is no inviability: 0.64 X 0 = 0

Thus the frequency of genotypes among inviable individuals was:

A_1A_1: 0
A_1A_2: 0.048 / (0.048 + 0.012) = 0.8
A_2A_2: 0.012 / (0.048 + 0.012) = 0.2

(10) 7. A population of 300 cattle is surveyed at a locus with two codominant alleles. The genotype A_1A_1 is found to be present in 100 cattle, A_1A_2 is present in 40 cattle, and A_2A_2 is present in 160 cattle.

(5) a. What are the allele frequencies?

$Pr(A_1) = p = (100+(40/2)) / 300 =$ **0.4** $\quad Pr(A_2) = q = (160+(40/2)) / 300 =$ **0.6**

(5) b. Estimate the level of inbreeding (inbreeding coefficient) in the population.

f $= 1 - H/2pq = 1 - ((40/300)/(2 \times 0.4 \times 0.6)) = 1 - (0.133/0.48) =$ **0.722**

Hint: verify using ***f*** = 0.722 on unit square

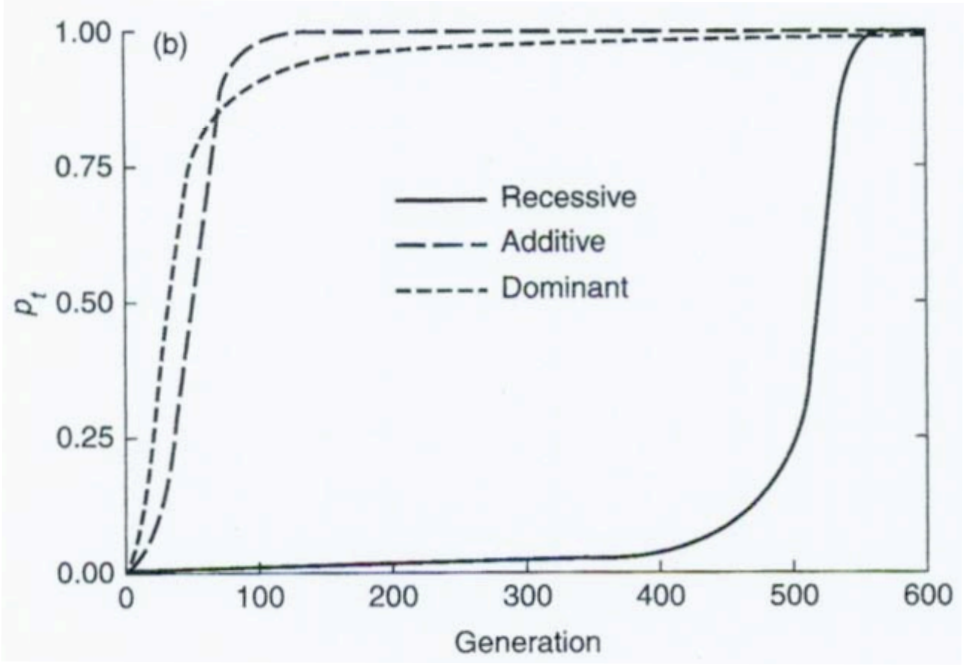
(5) 8. Five alleles (A_1, A_2, A_3, A_4, A_5) at a single locus (A) have been identified on a plant that prevents self-fertilization by self-incompatibility. If the A locus is at equilibrium, what is the expected allele frequency of allele A_3?

Answer: $q_{eA3} = 1/(n \text{ alleles}) = 1/5 =$ **.20**

(4) 9. For diploid plants that are self-fertilizing, heterozygosity in one generation is equal to half that of the previous generation. For mice undergoing generations of full-sibling crosses, heterozygosity will be equal to half that of the parental generation, plus a quarter that of the grandparental generation. Why is it necessary to add a quarter of the heterozygosity present in the grandparents' generation for the mice but not for the plants?

In the case of the full-sibling crosses (but not in the case of the selfing plants), the parents may each be homozygote at a locus, but for different alleles (mother A_1A_1 and father A_2A_2, or vice versa). At this locus, their mating would increase heterozygosity among the offspring. Overall, this effect would be equal to ¼ the heterozygosity present in the grandparents.

5) 10. The following Figure from Hedrick's text shows the rise in frequency for an allele at an initially low frequency (p_0 = 0.01) undergoing positive Darwinian selection (s = 0.1), in cases where the allele is recessive, dominant, or additive:



Given that the fitness value for an additive allele is exactly half way between the fitness of a dominant allele and the fitness of a recessive allele, why doesn't the curve showing increase in an additive allele fall exactly intermediate between the curve for a dominant and the curve for a recessive allele?

At low frequencies an allele is found almost entirely in heterozygous genotypes. Selection does not affect the heterozygotes if the allele is recessive, but the heterozygotes *are* subject to selection if the allele is additive. Hence at low frequencies an additive (like a dominant) allele increases quickly in frequency due to positive selection acting on the heterozygotes, while a recessive allele does not.

(9) 11. A rare recessive disease has an allelic frequency of 0.02.

(3) a. Population 1 is in Hardy Weinberg equilibrium. What is the frequency of diseased homozygotes in this population?

Q = q^2 = (0.02)^2 = **0.0004**

(3) b. Population 2 also has an allelic frequency of 0.02 but is inbred so that ***f*** = 0.1. What is the frequency of diseased homozygotes in population 2?

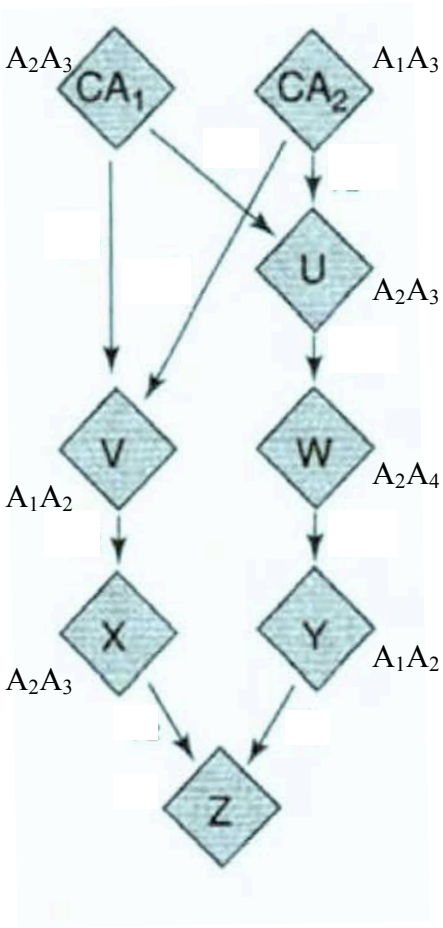
Q = q2 + fpq = (0.02)^2 + (0.1 X 0.98 X 0.02) = 0.0004 + 0.00196 = **0.00236**

(3) c. The allelic frequencies are the same between the two populations, but what is the ratio of the frequencies of diseased individuals between population 2 and population 1?

0.00236:0.0004

or **5.9 to 1**

(9) 12. In the pedigree on the right, CA1 and CA2 are outbred.



(3) a. What is the inbreeding coefficient for individual X?

f = 0

(3) b. What is the inbreeding coefficient for individual Z?

f = (0.5)^6 + (0.5)^6 = 1/64 + 1/64 = 2/64 = 0.03125

(3) c. Genotypes are shown for the A locus for individuals included in the pedigree. What are the possible genotypes for individual Z to have at the A locus? Which of these would be identical by descent, and which have identity in state?

Four possible genotypes for Z:

A_1A_2 the two alleles are not identical
A_2A_2 identical by descent
A_1A_3 the two alleles are not identical
A_2A_3 the two alleles are not identical