

ANSC 446 / IB 416
Population Genetics
Exam 3, November 20, 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.

(9) 1. The ostrich farm on Route 57, owned by Mr. Jones, still only has single combed birds. Single comb is recessive to double comb. However, the Jones farm has now decided to double their flock size, by adding individuals from the ostrich farm on Route 47, owned by the Patels. Mrs. Patel has estimated the allele frequency of single comb in their flock as 0.4

(3) a. Estimate the allele frequency of single comb in the new Jones flock.

$$q_1 = (1-m)q_0 + mq_m = (.5)(1) + (.5)(.4) = 0.7$$

(3) b. What would be the migration rate for the first generation?

$$\text{Migration rate} = (1 - 0.7) / (1 - 0.4) = .3/.6 = .5$$

(3) c. Would this represent an example of the continent-island model of migration, or of the general model of migration. Why?

Continent-island model of migration. Gene flow is unidirectional.

(3) 2. What is the approximate probability of loss of a new neutral mutation in the first generation if the number of offspring per family follows a Poisson distribution and averages 2 offspring per family.

$$\text{ANS: } P(\text{loss}) = .368 \text{ (See Hedrick page 367)}$$

(4) 3. What is the difference between orthologous and paralogous genes?

Orthologous genes are separated by a speciation event (eg, most single copy genes in humans will also be found as a single copy gene in the chimpanzee). Paralogous genes are separated by a duplication event (eg, the genes encoding myoglobin and hemoglobin descend from duplication of an ancestral gene).

- (15) 4. Only 40 Javan rhinoceros survive in the island of Java.
Assume that this population size is constant across generations.

(3) a. For a population of this size, how much of the current generation's heterozygosity will be lost in the next generation due to drift?

$H_{t+1} = (1 - (1/(2N))) \times H_t = (1 - 1/(2 \times 40)) H_t = (1 - 0.0125) H_t = 0.9875 H_t$
Since .9875 of heterozygosity is retained each generation, **.0125 or 1.25%** of any remaining heterozygosity is lost due to drift each generation.

(3) b. If drift is the only force operating to alter genotypic frequencies, how many generations will it take for a reduction in heterozygosity to approximately 50% of the initial value in populations of size 40?

ANS: $t = -2 (N) \ln(x) = -2(40) \ln(.5) = \mathbf{55 \text{ generations}}$

(3) c. If drift is the only force operating to alter allele frequencies, how many generations will it take to reach fixation for an allele with an initial frequency of .5 when the population size is 40?

ANS: $T(q) = [-4 (N) (1-q) \ln(1-q)] / q = [-4(40)(1-.5) \ln(1-.5)] / .5$
 $= [-80 \ln(.5)] / .5 = \mathbf{111 \text{ generations}}$

(Note that this twice the value of the answer in part b)

(3) d. Assuming that the effective population size equals the census size of 40, and assuming no selection or migration. How long is it expected to take on average for a new neutral mutant allele to become fixed in this population?

Answer: $T_1(p) = 4 N_e = 4(40) = \mathbf{160 \text{ generations}}$

(3) e. For a new neutral mutation in this population, what is the probability that it will become fixed in the population?

Answer: $1/(2N) = 1/80 = \mathbf{0.0125}$

- (4) 5. Name a type of transposable element present in the human genome

(Example) short interspersed nuclear elements (SINEs) such as Alu sequences

(10) 6. Give the best definition for the following terms:

(2) a. Frameshift mutant: indel that changes the reading frame of subsequent codons in a transcript (and thus affects the amino acid sequence)

(2) b. Metapopulation: subpopulations in discrete habitat patches, that turnover with extinction and recolonization from other patches. The newly colonized patches may display the results of founder effects

(2) c. Cline: a directional change in allele frequencies across (geographic) space or between subpopulations (or between species), potentially due to selection or substructure. May be stable or transient.

(2) d. Mutation load: reduction in fitness due to deleterious mutants not yet removed by selection

(2) e. Admixture: the proportion of gene flow from an outside population.

(12) 7. The Seeing Eye maintains a kennel of approximately 20 female German Shepherds and 20 male German Shepherds in their breeding population.

(3) a. What is the effective population size for an autosomal gene?

$$\text{Answer: } N_e = \frac{4 N_f N_m}{N_f + N_m} = \frac{4(20)(20)}{20 + 20} = \frac{1600}{40} = 40$$

(3) b. What is the effective population size for an mtDNA gene?

$$\text{Answer: } N_e = \frac{N_{ef}}{2} = \frac{20}{2} = 10$$

(3) c. What is the effective population size for a Y-chromosome gene?

$$\text{Answer: } N_e = \frac{N_{em}}{2} = \frac{20}{2} = 10$$

(3) d. What is the effective population size for an X-chromosome gene?

$$\text{Answer: } N_e = \frac{9 \times N_f N_m}{2N_f + 4N_m} = \frac{9 \times (20) \times (20)}{2(20) + 4(20)} = \frac{3600}{120} = 30$$

(4) 8. Why is the hypothetical fitness curve of mutations said to be bimodal?

There is one peak of frequency near neutrality (relative fitness near ω -bar) and a smaller peak near lethality (fitness at or near $\omega = 0$).

(4) 9. According to Dr. Stanley Ambrose, what event occurring 70,000 years ago may have shaped human evolution through bottlenecks or founder effects?

The eruption of Toba

(4) 10. What are the similarities and differences between the stepping-stone model of gene flow, and isolation by distance?

The stepping-stone model assumes gene flow occurs spatially in one dimension between separated populations; by contrast, isolation by distance can occur across continuous habitat and in two dimensions. Both are similar in generating distance-dependent gene flow, in which subpopulations separated by greater geographic distance also tend to display greater genetic distance (due to limited gene flow).

(6) 11. Describe 3 tests of selection (or neutrality)

1. Ewens-Watterson test: compares homozygosity under HWE to that expected under Neutral Theory.
2. Tajima's D: calculates theta using nucleotide diversity and also using segregating sites; the two estimates should be similar under neutrality
3. Hudson-Kreitman-Aguade test: compares within- and between-species variation; they should be similar under neutrality
4. McDonald-Kreitman test: Compares the synonymous to non-synonymous ratio in differences fixed between species to the ratio in polymorphisms within species. Under neutrality, the ratios should be similar.
5. Non-synonymous vs. silent substitutions: within species, are used to identify purifying and Darwinian selection. Non-synonymous will be less common after purifying selection, but more common after Darwinian selection.

(5) 12. Two populations are similar except in terms of inbreeding coefficient. The first population has no inbreeding, while the second population has an inbreeding coefficient of $f = 0.2$. The first population has an effective population size of 100. What is the effective population size of the second population?

$$N_e = N / (1+f) = 100 / (1 + 0.2) = 100/1.2 = 83.3$$

(20) 13. Assume that for two isolated subpopulations of the same size, frequencies of alleles A_1 and A_2 are 0.4 and 0.6, respectively, in the first subpopulation; and frequencies of alleles A_1 and A_2 are 0.6 and 0.4, respectively, in the second subpopulation.

(4) a. Assuming Hardy-Weinberg proportions, what is the frequency of heterozygotes in the first subpopulation?

$$2pq = 2 \times 0.4 \times 0.6 = 0.48$$

(4) b. Assuming Hardy-Weinberg proportions, what is the frequency of heterozygotes in the second subpopulation?

$$2pq = 2 \times 0.6 \times 0.4 = 0.48$$

(4) c. If the two subpopulations were merged into a single panmictic (freely interbreeding) population, what would be the frequency of heterozygotes after Hardy Weinberg equilibrium was reached in the combined population?

$$2 \times p\text{-bar} \times q\text{-bar} = 2 \times 0.5 \times 0.5 = 0.50$$

(4) d. What is the term used in cases where subpopulation structure results in a reduction in overall heterozygosity, even though the subpopulations themselves are in Hardy-Weinberg equilibrium?

Wahlund effect.

(4) e. Calculate G_{ST} between the two subpopulations (before they were merged)

$$G_{ST} = (H_T - H_S) / H_T = (0.50 - 0.48) / 0.5 = 0.02/0.5 = 0.04$$