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Population Genetics
ECOL 426/526 2004
Final Exam
(Total points possible for graduate students112/112; for undergraduates112/100)

1. (4 pts) Four sequences of a 1000 bp gene give the following counts of pairwise differences: 5, 8, 4, 6, 8, 5. What is the estimate of nucleotide diversity for this sample?

2. (4 pts) Consider a sibship of four offspring from the mating Aa x aa. What is the probability of obtaining two heterozygous offspring in the sibship?

3. (4 pts) The frequency of one form of recessive X-linked color blindness is 5% among European males. What is the expected frequency of this form of color blindness among females? What fraction of females would be heterozygous carriers?

4. (4 pts) The fitnesses of AA, Aa, and aa are 1.0, 0.8, and 0.7, and the initial frequency of A, p_o, is 0.7. What is the frequency of the A allele after one generation of selection?

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5. (4 pts) Calculate the equilibrium allele frequency with overdominance when the fitnesses of AA, Aa, and aa are 0.3, 1, and 0.7.

6. (4 pts) If the frequency of an autosomal recessive disorder is 1/2500 among unrelated parents, what is the expected frequency among the offspring of first cousins?

7. (4 pts) Calculate the inbreeding coefficient for individual A in the path below.

8. Consider a Wright-Fisher model of random genetic drift with a single di-allelic locus (A and a) in which each subpopulation has 50 diploid individuals. At time t = 0 all subpopulations have $p_o = 0.25$ and $q_0 = 0.75$.

a) (2 pts) What is the variance in allele frequency among subpopulations after 4 generations?

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b) (2 pts) What is the average allele frequency after 4 generations?

9 (4 pts) A population of annual plants is studied for four years, and the following numbers of individuals are counted each year: 1000, 1000, 100, 1000. What is the effective population size over this period?

10. (4 pts) Consider a locus at which two alleles, A and B, are maintained at equilibrium frequencies as a result of overdominance. The mean fitness of the population is 0.9. A new allele, C, arises by mutation. Under what conditions will this new allele be able to invade the population as a result of selection?

11. (6 pts) Briefly compare and contrast levels and patterns of DNA sequence variation in humans and in *Drosophila melanogaster*. Consider the level and pattern of nucleotide variation, and suggest explanations for any differences described.

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12. (4 pts) An autosomal gene in one individual in a population of 500 annual plants undergoes mutation to a new neutral allele. What is the probability that this allele will be lost?

13. (4 pts) Calculate F_{ST} for the populations with the genotype frequencies shown in the following table:

	Population 1	Population 2
AA	0.056	0.672
Aa	0.288	0.256
aa	0.656	0.072

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14. (4 pts) If three populations with allele frequencies 0.2, 0.4, and 0.6 undergo migration according to the island model with m=0.05, what are the expected allele frequencies after 8 generations?

15. (6 pts) Briefly describe (a) how mutation rates have been measured in Drosophila using phenotypic mutants, (b) how mutation rates have been measured using molecular data. (c) Do these estimates agree?

16. (4 pts) What evidence suggests that mutation rates may be adaptive? (Be brief).

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17. (4pts) What is the equilibrium frequency of a recessive allele arising with a mutation rate of 10^{-6} with a fitness of 0.4 in homozygotes?

18. (4 pts) The neutral mutation rate for the F9 locus is 10^{-5} in a mouse species that has an effective population sizeof 2×10^4. On average, how often does a population of these mice become fixed for a new mutation at F9?

19. (4 pts) Derive the expression for steady-state heterozygosity under the infinite alleles model. (You do not need to write out all the alegebraic steps; set-up the equations and explain each term, then give the final answer).

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20. (4 pts) Show why the reduction in the mean fitness of a population due to recurrent deleterious mutation is independent of how harmful the mutation is.

21. (4 pts) In the forward and reverse mutation model, what is the equilibrium frequency, p, of A if $\mu = 10^{-5}$ and $v = 10^{-6}$?

22. (4 pts) What is the equilibrium heterozygosity (infinite alleles model) of a population of effective size 50 if new mutations are introduced at a rate 10^{-5} by mutation and at a rate 10^{-4} by migration?

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23. Consider the frequency distribution of segregating sites in the following dataset.

	1	2	3	4	5	6	7	8	9	10
Consensus	A	C	T	T	C	A	G	G	G	A
Allele 1	G	.	.	.	.	.	.	.	.	.
Allele 2	.	T	.	.	.	.	.	.	.	.
Allele 3	.	.	C	.	.	.	.	.	.	.
Allele 4	.	.	.	C	.	.	.	.	.	.
Allele 5	.	.	.	.	T	.	.	.	.	.
Allele 6	.	.	.	.	.	G	.	.	.	.
Allele 7	.	.	.	.	.	.	A	.	.	.
Allele 8	.	.	.	.	.	.	.	A	.	.
Allele 9	.	.	.	.	.	.	.	.	A	.
Allele 10	.	.	.	.	.	.	.	.	.	G

(a) (2 pts) Is Tajima's D negative, zero, or positive for these data? (You do not need to calculate the exact value of Tajima's D, but you do need to show any calculations that lead you to your answer).

(b) (2 pts) Suggest two hypotheses to explain these data.

(c) (2 pts) Suggest a test that would enable you to discriminate between your two hypotheses.

24. (4 pts) The dystrophin locus, *Dmd*, is one of the largest genes in the human genome, spanning over 2 Mb of DNA. Exon 5 and exon 44 are 1 cM apart. In one isolated population, polymorphic sites in these two exons are in complete linkage disequilibrium. How long will it take D' to decay to 0.5 in this population?

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25. (4 pts) Graph the fixation rate as a function of population size for (a) neutral mutations, and (b) advantageous mutations.

26. (6 pts) What is the neutral theory of molecular evolution? What are the major tenets of this theory?