Homework Set #2 Key

See Nielsen & Slatkin for solutions to problems 3, 5, 7, 8, & 10

1.



- (a) The only chain is the common ancestor <u>A</u>. So $f_I = (1 + f_A)/2 = (1+0)/2 = \frac{1}{2}$. (Note that this result can be argued directly by applying Mendel's principle of segregation to a selfer.)
- (b) Two chains: CAD and CBD. $f_1 = (\frac{1}{2})(1 + f_A)/2(\frac{1}{2}) + (\frac{1}{2})(1 + f_B)/2(\frac{1}{2}) = (\frac{1}{2})(1 + 0)/2(\frac{1}{2}) + (\frac{1}{2})^3 + (\frac{1}{2})^3 = \frac{1}{4}$
- (c) Two chains: ECADF and ECBDF. $f_I = (\frac{1}{2})(1 + f_A)/2(\frac{1}{2}) + (\frac{1}{2})(1 + f_B)/2(\frac{1}{2}) = (\frac{1}{2})(1 + 0)/2(\frac{1}{2}) + (\frac{1}{2})^3 = (\frac{1}{2})^3 + (\frac{1}{2})^3 = \frac{1}{4}$
- (c) Two chains: ECADF and ECB. $f_1 = (\frac{1}{2})(\frac{1}{2})(1 + f_A)/2(\frac{1}{2})(\frac{1}{2}) + (\frac{1}{2})(\frac{1}{2})(1 + f_B)/2(\frac{1}{2})(\frac{1}{2}) = (\frac{1}{2})^4(1+0)/2 = (\frac{1}{2})^5 + (\frac{1}{2})^5 = 1/16$
- 2. Disease frequency = $0.001 = q^2 + 0.005pq = q^2 + 0.005(1-q)q = 0.995q^2 + 0.005q$. Solving for q(using the quadratic formula) gives $q = \frac{-0.005 + \sqrt{0.005^2 - 4(0.995)(-0.001)}}{2(0.995)} = 0.029$. In a random mating population, the frequency of the disease would be $q^2 = (0.029)^2 = 0.00084$. Inbreeding increases the prevalence of the disease in this population by a factor of 0.001/0.00084 = 1.2. Although the level inbreeding is very low (an order of magnitude smaller than 1st cousins), this is a fairly substantial increase in disease prevalence.
- 4. Nielsen & Slatkin, p. 74 #4.2

a.
$$p_1 = (2*20 + 20)/(2*60) = 0.5$$

 $p_2 = (2*15 + 15)/(2*60) = 0.375$
 $p_3 = (2*20 + 25)/(2*60) \approx 0.54$
 $\bar{p} = \frac{0.5 + 0.375 + 0.54}{3} \approx 0.472$
b. $H_S = \frac{2(0.5)(0.5) + 2(0.375)(0.625) + 2(0.54)(0.46)}{3} \approx 0.488$
 $H_T = 2(0.472)(0.528) \approx 0.498$
 $F_{ST} = \frac{H_T - H_S}{H_T} = \frac{0.498 - 0.488}{0.498} \approx 0.02$

- 6. Nielsen & Slatkin, p. 33 #2.2 Let q_t be the expected frequency of the mutation in generation t. If $q_t = 0.1$ then $q_{t+1} = (1 - u)q_t + v(1 - q_t) = (1 - 10^{-6})(0.1) + (5 \times 10^{-6})(1 - 0.1) = 0.1000044$
- 9. Nielsen & Slatkin, p. 58 #3.6

$$\hat{\theta}$$
(Watterson) = $=\frac{S}{\sum_{k=2}^{n} \frac{1}{k-1}} = \frac{5}{1+\frac{1}{2}+\frac{1}{3}} = 2.73$

 $\hat{\theta}$ (Tajima) = π = 3

11. Nielsen & Slatkin, p. 58 #3.8

Site frequency spectrum: E(proportion of singletons) = E(f_1) = $\frac{1}{\sum_{k=1}^{n-1} \frac{1}{k}} = \frac{1}{1+\frac{1}{2}+\frac{1}{3}} = \frac{6}{11} = 0.545$. Assuming A is ancestral, the observed value of f_1 = 0, which is much less than the

expected value of 0.545.

Folded SFS: $E(f_1^*) = E(f_1 + f_{n-1}) = E(f_1 + f_3) = E(f_1) + E(f_3) = \frac{1}{\sum_{k=1}^3 \frac{1}{k}} + \frac{1/3}{\sum_{k=1}^3 \frac{1}{k}} = \frac{8}{11} = 0.727$. The observed proportion of folded singletons is $f_1^* = f_1 + f_3 = 0 + 2/5 = 0.4$, which is also less than the expected value of 0.727.