## 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 $\underline{\text { A. So }} f_{l}=\left(1+f_{A}\right) / 2=(1+0) / 2=1 / 2$. (Note that this result can be argued directly by applying Mendel's principle of segregation to a selfer.)
(b) Two chains: $\underline{C A D}$ and CBD. $f_{l}=(1 / 2)\left(1+f_{A}\right) / 2(1 / 2)+(1 / 2)\left(1+f_{B}\right) / 2(1 / 2)=(1 / 2)(1+0) / 2(1 / 2)+$ $(1 / 2)(1+0) / 2(1 / 2)=(1 / 2)^{3}+(1 / 2)^{3}=1 / 4$
(c) Two chains: ECADF and ECBDF. $f_{l}=(1 / 2)\left(1+f_{A}\right) / 2(1 / 2)+(1 / 2)\left(1+f_{B}\right) / 2(1 / 2)=(1 / 2)(1+0) / 2(1 / 2)+$ $(1 / 2)(1+0) / 2(1 / 2)=(1 / 2)^{3}+(1 / 2)^{3}=1 / 4$
(c) Two chains: ECADF and ECB. $f_{1}=(1 / 2)(1 / 2)\left(1+f_{A}\right) / 2(1 / 2)(1 / 2)+(1 / 2)(1 / 2)\left(1+f_{B}\right) / 2(1 / 2)(1 / 2)=$ $(1 / 2)^{4}(1+0) / 2+(1 / 2)^{4}(1+0) / 2=(1 / 2)^{5}+(1 / 2)^{5}=1 / 16$
2. Disease frequency $=0.001=q^{2}+0.005 p q=q^{2}+0.005(1-q) q=0.995 q^{2}+0.005 q$. 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 $1^{\text {st }}$ 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. $H_{S}=\frac{2(0.5)(0.5)+2(0.375)(0.625)+2(0.54)(0.46)}{3} \approx 0.488$
$H_{T}=2(0.472)(0.528) \approx 0.498$
$F_{S T}=\frac{H_{T}-H_{S}}{H_{T}}=\frac{0.498-0.488}{0.498} \approx 0.02$
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 $q_{t+1}=(1-u) q_{t}+v\left(1-q_{t}\right)=\left(1-10^{-6}\right)(0.1)+\left(5 \times 10^{-6}\right)(1-0.1)=0.1000044$
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 $)=\pi=3$
11. Nielsen \& Slatkin, p. 58 \#3.8

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

Folded SFS: $E\left(f_{1}^{*}\right)=E\left(f_{1}+f_{n-1}\right)=E\left(f_{1}+f_{3}\right)=E\left(f_{1}\right)+E\left(f_{3}\right)=\frac{1}{\sum_{k=1}^{3} \frac{1}{k}}+\frac{1 / 3}{\sum_{k=1}^{3} \frac{1}{k}}=$ $\frac{8}{11}=0.727$. 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 .

