Student Number___________________ FULL NAME____________________________

Grade_____ /40__

# Population Genetics Midterm

BIO 3119 Fall 2014

## Question 1:

How many generations of genetic drift are required to reduce the expected heterozygosity to 10% of its original value in a diploid, randomly mating population of size 20? [4 marks]

$$H_t = \left(1 - \frac{1}{2N}\right)^t H_0 \approx H_0 e^{-t/2N}$$

$0.1H_0 = H_0e^{-t/2N}$
$0.1 = e^{-t/2N}$
$\ln(0.1) = -t/2N$
$t = -2N \ln(0.1)$

when $N = 20$, $t = -2(20)\ln(0.1) = 92.1$ generations
when $N = 100$, $t = -2(100)\ln(0.1) = 460.5$ generations

## Question 2

In a randomly mating population of size $N = 50$, what is the probability that a neutral allele present in a single copy (e.g. a new mutation) will be lost in the next generation? What is the probability that this allele eventually fixes? [4 marks]

Answer:

$$P(j\ alleles of\ type A) = \binom{2N}{j} p^j q^{2N-j} = \frac{(2N)!}{j!(2N-j)!} p^j q^{2N-j}$$

$$P(0\ alleles of\ type A) = \binom{2(50)}{0} (0.01^0) 0.99^{100} = \frac{(100)!}{0!(100)!} 0.99^{100} = 0.366$$

Probability it eventually fixes is equal to its current frequency = 1/100 = 0.01.

## Question 3

Allozyme genotype data has been collected from a single population of rare middle earth Hobbits. The observed genotype frequencies are shown for one polymorphic locus with three alleles (F = fast, M = medium and S = slow for band migration rates in starch gels).

| **Genotypes** | **Observed counts** | **Observed genotype freq.** | HW expected freq. | HW expected count | $(O-E)^2/E$ |
|---|---|---|---|---|---|
| FF | 320 | 0.32 | $(0.4225)^2 = 0.179$ | 1000(0.179) = 179 | 111.07 |
| MM | 120 | 0.12 | $(0.22)^2 = 0.0484$ | 1000(0.0484) = 48.4 | 105.92 |
| SS | 235 | 0.235 | $(0.357)^2 = 0.127$ | 1000(0.127) = 127 | 91.84 |
| FM | 80 | 0.08 | (2)(0.4225)(0.22) = 0.186 | 1000(0.186) = 186 | 60.41 |
| FS | 125 | 0.125 | (2)(0.4225)(0.357) = 0.302 | 1000(0.302) = 302 | 103.74 |
| MS | 120 | 0.120 | (2)(0.22)(0.357) = 0.157 | 1000(0.157) = 157 | 8.72 |
| Total | 1000 | | | | 481.697 |

a) Are genotypes at this locus at Hardy-Weinburg expected frequencies? Show your calculations below and using the table above, state the degrees of freedom, and by referring to the Chi-Square table at the end of the exam, explicitly state whether the test results lead you to accept or reject Hardy-Weinburg equilibrium. [6 marks]

Let $p$ = frequency of F allele, $q$ = frequency of M allele, $r$ = frequency of S allele

$p = [2(320)+80+125)]/2000 = 0.423$
$q = [2(120) + 80 + 120]/2000 = 0.22$
$r = 1 - p - q = 0.357$
$df$ = 6 classes of data – 2 allele frequencies estimated from the data – 1 = 3
$\chi^2 = 481.7$, which is MUCH greater than the critical value of 7.815.
Therefore, we reject HW for this locus ($P < 0.05$; actually $P < 0.001$)

b) Calculate the fixation index, $F$. [2 marks]

$H_{exp} = 0.186 + 0.302 + 0.157 = 0.645$; $H_{obs} = 0.080 + 0.125 + 0.120 = 0.325$

$F = (H_e - H_o)/H_e = (0.645 - 0.325)/0.645 = 0.496$

c) Is heterozygosity lower or higher than expected under HWE? Suggest a likely reason for the observed deviation. [2 marks]

Heterozygosity is reduced by almost 50%. Inbreeding (i.e., mating between relatives more closely related than expected by chance) is a likely cause.

**Question 4**

A forensic investigator is evaluating two new loci from a reference population in terms of their utility in DNA fingerprinting. Each locus had two alleles with the following observed frequencies of the four possible gametes:

A1B1 = 0.6
A1B2 = 0.1
A2B1 = 0.2
A2B2 = 0.1

a) What are the frequencies of the four alleles in this population? [2 marks]

$p_{A1} = 0.6 + 0.1 = 0.7$; $p_{A2} = 0.2 + 0.1 = 0.3$; $p_{B1} = 0.6 + 0.2 = 0.8$; $p_{B2} = 0.1 + 0.2 = 0.2$

b) What are *D* and *D'* for this set of gamete frequencies? [6 marks]

$D = (0.6)(0.1) – (0.1)(0.2) = 0.04$
Since $D > 0$, use the smaller of $p_{A1}*p_{B2}$ and $p_{A2}*p_{B1}$
$\mathrm{Min}\{(0.7)(0.2), (0.3)(0.8)\} = \min\{0.14, 0.24\}$
So $D_{max} = 0.14$
$D' = 0.04/0.14 = 0.286$

c) In one or two sentences, explain what your values of *D* and *D'* mean and what this says about their usefulness for DNA fingerprinting. [2 points]

*D* indicates a surplus of coupling over repulsion gametes. *D'* indicates that this disequilibrium is 28.6% of its maximum value given the observed allele frequencies. In DNA fingerprinting, the probability of observing a particular multilocus genotype is calculated as the product of the observing the particular genotype at each locus, which assumes the loci are independent (i.e. in linkage equilibrium). These loci should not be used (or else the probability of observing a particular multilocus genotype must take into account this disequilibrium).

**Question 5**

Alport Syndrome is a **dominant** X-linked disease that causes progressive hearing loss and kidney malfunction. The frequency of the gene in the intermountain region of the western United States was recently estimated to be 1/1000. In a population of 100,000 males and 105,000 females, calculate the expected number of each sex exhibiting the disease. Assume the population is at HWE. [4 marks]

Females: $p^2 \times 105{,}000 + 2pq \times 105{,}000 = 0.105 + 209.79 = 209.895$ (or 210)
Males: $p \times 100{,}000 = 100$

**Question 6**
a) Compare and contrast the effects of genetic drift and inbreeding on both allele and genotype frequencies within a single, finite population. Point form answers are fine. [6 marks]

*Inbreeding*

- occurs when matings between relative are more common than expected by chance
- alters genotype frequencies, decreasing heterozygosity and increasing homozygosity
- causes no change in allele frequencies

*Genetic drift*

- occurs due to random sampling in finite populations which, as population size decreases, results in an increased frequency of mating between relatives *even when mating is random* (because in smaller populations randomly chosen mates are more likely to be related
- causes changes in allele frequencies but no deficiency of heterozygotes within a single generation relative to HWE, as long as the population mates randomly.

b) When average effects are considered across many replicate subpopulations, how are genetic drift and inbreeding similar? [2 marks]

When considered across many replicate subpopulations, drift has the same average effect as inbreeding in that it causes no expected change in allele frequency and reduces heterozygosity.