COLUMBIA UNIVERSITY

DEPARTMENT OF BIOSTATISTICS

P8149 – HUMAN POPULATION GENETICS

Exercise Sheet 1 (Model Answers)

Question 1

Let the frequency of the genotype IJ (I, J = A, B, C) be P_{ij} and the frequency of the allele I (=A, B, C) be p_i. Then a

$$p_A = P_{AA} + \frac{1}{2}\left(P_{AB} + P_{AC}\right)$$
$$= .096 + \frac{1}{2}\left(.483 + .028\right)$$
$$= .3515,$$

$$p_B = P_{BB} + \frac{1}{2}\left(P_{AB} + P_{BC}\right)$$
$$= .343 + \frac{1}{2}\left(.483 + .050\right)$$
$$= .6095,$$

and

$$p_C = 1 - p_A - p_B$$
$$= 1 - .3515 - .6095$$
$$= .039$$

[.5+.5+.5=1.5 marks]

Question 2

The initial allele frequencies are

$$p_1 = P_{11} + \frac{1}{2}P_{12} = .2 + \frac{1}{2}\left(.4\right) = .4,$$

and

$$p_2 = 1 - .4 = .6$$

Because of random mating, the genotype frequencies in the next generation are in HWE:

$$P_{11} = p_1^2 = .4^2 = .16,$$
$$P_{12} = 2p_1p_2 = 2(.4)(.6) = .48,$$
$$P_{22} = 1 - P_{11} - P_{22} = 1 - .16 - .48 = .36.$$

[.5+1.5=2 marks]

Question 3

Let the recessive allele be denoted by a and its frequency by q. Since the population is in HWE, we have $q^2 = .30$ so that $q = .548$.

[2 marks]

Question 4

Let the recessive allele be denoted by a and its frequency by q. Then $q^2 = 1/20000$ and the frequency of carriers is approximately $2q = 2\sqrt{1/20000} = .014$

[2 marks]

Question 5

(a) The genotype frequencies are

$$P_{MM} = \frac{1787}{1787 + 3039 + 1303} = \frac{1787}{6129} = .2916,$$
$$P_{MN} = \frac{3039}{6129} = .4958,$$
$$P_{NN} = 1 - .2916 - .4958 = .2126.$$

(b) The allele frequencies are

$$p_M = .2916 + \frac{1}{2}(.4958) = .5395,$$

$$p_N = 1 - p_M = .4605$$

(c) The expected genotypic counts under HWE are

$$E_{MM} = (.5395)^2 (6129) = 1783.9$$
$$E_{MN} = 2(.5395)(.4605)(6129) = 3045.4$$
$$E_{NN} = (.4605)^2 (6129) = 1299.7$$

Under $H_0 : HWE$,

$$X_{test}^2 = \sum \frac{(E_{ij} - O_{ij})^2}{E_{ij}}$$

$$= \frac{(1783.9 - 1787)^2}{1783.9} + \frac{(3045.4 - 3039)^2}{3045.4} + \frac{(1299.7 - 1303)^2}{1299.7}$$

$$= .027$$

Under H_0, $X_{test}^2 \sim \chi_1^2$, and the p-value of the test is $p-val = \Pr\{\chi_1^2 > .027\} = .869$. Since p-$val \gg .05$, we cannot reject H_0 at the 5% level of significance.

[1.5+1+2=4.5 marks]

Question 6

Using the fact that random mating is equivalent to the random union of gametes, the frequencies of the different genotypes are

$$freq(A_1 A_1 A_1) = p^3,$$
$$freq(A_1 A_1 A_2) = \frac{3!}{2!1!} p^2 q = 3p^2 q,$$
$$freq(A_1 A_2 A_2) = \frac{3!}{1!2!} pq^2 = 3pq^2,$$
$$freq(A_2 A_2 A_2) = q^3.$$

[Alternatively, the above frequencies can be obtained from the binomial expansion of $(pA_1 + qA_2)^3$]

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