

COLUMBIA UNIVERSITY

DEPARTMENT OF BIOSTATISTICS

P 8149 - HUMAN POPULATION GENETICS

exercise sheet 2 (covers chapter 3 and 4)

Date due: Saturday OCT 18, 2025 (SUBMIT ONLINE ON CANVAS)

Question 1 (3 marks)

If one gene has alleles A_1, A_2 with frequencies p_1, q_1 , and another gene has alleles B_1, B_2, B_3 with frequencies p_2, q_2, r_2 , what are the frequencies of gametes with linkage equilibrium, assuming that $p_1 = .3, p_2 = .2$ and $q_2 = .3$?

Question 2 (3 marks)

Consider a locus L_1 with two alleles A and a having frequencies p_A and p_a , and a second locus L_2 with two alleles B and b having frequencies p_B and p_b . Show that

$$p_A p_B + p_A p_b + p_a p_B + p_a p_b = 1.$$

Question 3 (4 marks)

If it is known that two loci (L_1 and L_2) have two alleles each (A, a and B, b) and that the alleles occur at equal frequencies in a certain population (i.e. $p_A = p_a = 1/2$ and $p_B = p_b = 1/2$). Assume that $g_{AB} = 1/3$.

- Determine the frequency of the remaining three gamete types in the population.
- If mating is at random and the two loci are on different chromosomes, what would be the gametic frequencies in the next generation?
- If mating is at random but the loci are linked with 30% recombination, what would be the gametic frequencies in the next generation?
- What will be the limiting gametic frequencies, allowing random mating for a long period of time.

Question 4 (4 marks)

The body weight of mice is affected by the pygmy (pg) gene; the following shows mean weights of mice with three possible genotypes (A_1 is the wild type and A_2 is the pg allele)

genotype	mean weight (g)
A_1A_1	14
A_1A_2	12
A_2A_2	6

- (a) Find the effect of allele A_1 and of allele A_2 when (i) $q = \text{freq}(A_2) = .1$, (ii) $q = .4$
(b) Find the breeding values when (i) $q = .1$, (ii) $q = .4$
(c) Find the dominance deviations when (i) $q = .1$, (ii) $q = .4$

Question 5 (6 marks)

Let X_p and Y_p be values of some quantitative trait on two first-cousins. A regression of Y_p on X_p gives a slope b . Show that the heritability of the quantitative trait can be estimated by

$$h^2 = 8b.$$

ANSWERS

Q1 $\text{freq}(A_1B_1) = .06$, $\text{freq}(A_1B_2) = .09$, $\text{freq}(A_1B_3) = .15$, $\text{freq}(A_2B_1) = .14$, $\text{freq}(A_2B_2) = .21$, $\text{freq}(A_2B_3) = .35$, Q3: (b) $g_{AB} = 7/24$, (c) $g_{AB} = 37/120$, (d) $g_{AB} = 1/4$; Q4 (a) For $q = .1$, average effect of $A_1 = .24$, average effect of $A_2 = -2.16$; for $q = .4$, average effect of $A_1 = 1.44$, average effect of $A_2 = -2.16$ (b) For $q = .1$, breeding values are $A_1A_1 = .48$, $A_1A_2 = -1.92$, $A_2A_2 = -4.32$; for $q = .4$, breeding values are $A_1A_1 = 2.88$, $A_1A_2 = -.72$, $A_2A_2 = -4.32$; (c) For $q = .1$, dominance deviations are $A_1A_1 = -.04$, $A_1A_2 = .36$, $A_2A_2 = -3.24$; for $q = .4$, dominance deviations are $A_1A_1 = -.64$, $A_1A_2 = .96$, $A_2A_2 = -1.44$