

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 A₁, A₂ with frequencies p_1, q_1 , and another gene has alleles B₁, B₂, B₃ 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 L₁ with two alleles A and a having frequencies p_A and p_a , and a second locus L₂ 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 (L₁ and L₂) 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 $g_{AB} = 1/3$.

- (a) Determine the frequency of the remaining three gamete types in the population.
- (b) If mating is at random and the two loci are on different chromosomes, what would be the gametic frequencies in the next generation?
- (c) If mating is at random but the loci are linked with 30% recombination, what would be the gametic frequencies in the next generation?
- (d) 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 (A₁ is the wild type and A₂ is the pg allele)

genotype	mean weight (g)
A ₁ A ₁	14
A ₁ A ₂	12
A ₂ A ₂	6

- (a) Find the effect of allele A₁ and of allele A₂ when (i) $q = \text{freq}(A_2) = .1$, (ii) $q = .4$
- (b) Find the breeding values when (i) $q = .1$, (ii) $q = .1$
- (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 freq(A₁B₁) = .06, freq(A₁B₂) = .09, freq(A₁B₃) = .15, freq(A₂B₁)=.14,freq(A₂B₂) = .21, freq(A₂B₃)=.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 A₁ = .24, average effect of A₂ = -2.16; for $q = .4$, average effect of A₁ = 1.44, average effect of A₂ = -2.16 (b) For $q = .1$, breeding values are A₁A₁ =.48, A₁A₂ = -1.92, A₂A₂ = -4.32; for $q = .4$, breeding values are A₁A₁ = 2.88, A₁A₂ = -.72, A₂A₂= -4.32; (c) For $q = .1$, dominance deviations are A₁A₁ = -.04, A₁A₂= .36, A₂A₂= -3.24; for $q = .4$, dominance deviations are A₁A₁ = -.64, A₁A₂ = .96, A₂A₂ = -1.44