

7.03 Problem Set 6

Due before 5 PM on Thursday, November 20, 2003

Hand in answers in recitation section or in the box outside the class

1. In humans, albinism (unpigmented skin, hair, and eyes) is due to an enzymatic deficiency, and it is an autosomal recessive trait. Suppose that in a small country of one million people (“Generation 1”), there are 500 *aa* albinos and 9000 *Aa* heterozygous carriers.

(a) Estimate q , the frequency of allele *a*, in Generation 1. Show your calculations.

(b) Estimate p , the frequency of allele *A*, in Generation 1. Show your calculations.

(c) In the next generation of 1 million individuals (Generation 2), what are the expected numbers of *aa* albinos and *Aa* carriers? (Assume random mating and all other Hardy-Weinberg conditions.) Show your calculations.

(d) Has the frequency of allele *a* changed between Generations 1 and 2? Briefly justify your answer.

(e) Was Generation 1 in Hardy-Weinberg equilibrium? Briefly justify your answer.

What is the probability that a child will be albino if:

(f) Both parents are non-albino members of Generation 2?

(g) One parent is a non-albino member of Generation 2, and the other parent is a non-albino member of Generation 1.

(h) Both parents are members of Generation 2, and one parent is albino and the other is non-albino.

2. Consider a heritable autosomal disease with an incidence in the population of 1 per thousand. On average, individuals with the disease have 80% as many children as the population average.

In answering the various parts of this question, assume that mating is random. State any additional simplifying assumptions that you employ, and show your calculations.

- (a) What mutation rate would be required to maintain the observed incidence of the disease in the population if the disease is dominant?
- (b) What mutation rate would be required to maintain the observed incidence of the disease in the population if the disease is recessive?
- (c) Now assume that the mutation rate is zero, that the disease is recessive, and that the disease allele is maintained in the population by heterozygote advantage. Calculate the heterozygote advantage, and restate its meaning in biological terms.
- 3.** In this question we will consider the interaction of selection and inbreeding in determining the incidence of autosomal recessive diseases. Consider a gene in which recessive mutations occur at a rate of 10^{-5} . Assume a selective disadvantage S of 0.4 in homozygotes for the recessive allele.

In answering the various parts of this question, show your calculations (unless none are required), and state any additional simplifying assumptions that you employ.

- (a) Calculate q , the frequency of the recessive allele. Also calculate the incidence of the disease. Assume random mating.
- (b) Now assume that, for thousands of generations, 10% of all children have been products of first-cousin matings (the remaining 90% being products of random matings). Calculate the steady-state value of q . Also calculate the incidence of the disease at steady state. (Hint: first modify the equation $\Delta q_{\text{sel}} = -Sq^2$ [from lecture 26] to reflect inbreeding's effects on the incidence of homozygotes for the recessive allele.)
- (c) Now assume that the population described in part (b) suddenly and completely ceases all inbreeding. Calculate the incidence of disease in the first generation conceived with no inbreeding.
- (d) Would q be expected to rise, fall, or remain unchanged during the first 10 generations after the cessation of inbreeding described in part (c)? Briefly justify your answer. (No calculations needed.) What numerical value would q approach after thousands of generations with no inbreeding?