

Population Genetics Problem Set 5 Key

1. A population of 200 cattle is surveyed at a locus with two codominant alleles. The genotype A_1A_1 is found to be present in 70 cattle, A_1A_2 is present in 40 cattle, and A_2A_2 is present in 90 cattle.

a. What are the allele frequencies?

$$\Pr(A_1) = p = (70 + 40/2) / 200 = \mathbf{0.45} \quad \Pr(A_2) = q = (90 + 40/2) / 200 = \mathbf{0.55}$$

b. Estimate the level of inbreeding (inbreeding coefficient) in the population.

$$F = 1 - H/2pq = 1 - ((40/200)/(2 \times 0.45 \times 0.55)) = 1 - (0.2/0.495) = \mathbf{0.596}$$

Hint: verify using $f = 0.596$ on unit square

2. A population of plants undergoes partial selfing, with 90% of individuals self fertilizing and 10% outcrossing. Assuming the frequency of alleles at a locus with two alleles is $p = 0.3$ for A_1 and $q = 0.7$ for A_2 .

a. What is the inbreeding equilibrium frequency of heterozygotes?

$$H_e = 4pq(1-S) / 2-S = (4 \times 0.3 \times 0.7)(1 - 0.9) / 2 - 0.9 = \mathbf{0.0764}$$

b. At equilibrium, what would be the frequency of homozygotes for A_1A_1 and for A_2A_2 ?

$$Q = q - (H_e/2) = 0.7 - (0.0764/2) = \mathbf{0.6618}$$

$$P = 1 - H - Q = 0.0764 - 0.6618 = \mathbf{0.2618}$$

Alternatively, equation 5.5b could also be used for P and Q

c. What would be the inbreeding coefficient at equilibrium?

$$f_e = S / (2-S) = 0.9/1.1 = \mathbf{0.818}$$

Alternatively, equation 5.6d could also be used with $T = 0.1$

3. A population of sparrows is found to carry a biallelic locus in the MHC that is subject to overdominance, with selection coefficient $s_1 = 0.2$ for genotype A_1A_1 , and $s_2 = 0.3$ for genotype A_2A_2 (assume that $\omega_{12} = 1$).

a. For allele A_2 what is the equilibrium frequency q_e ?

$$q_e = s_1 / (s_1 + s_2) = 0.2 / 0.2 + 0.3 = 0.2/0.5 = \mathbf{0.4}$$

b. Would the equilibrium frequency be stable? Why or why not?

Yes, overdominant systems are stable because fitness is at a maximum in the equilibrium state. Thus deviations away from the equilibrium frequency lead to selection effects on allele frequency that move them back to the equilibrium frequency.

c. What is the genetic load at the equilibrium frequency?

Genetic load is the reduction in fitness from the maximum possible, which would occur if all individuals were heterozygous (which is not possible).

Mean fitness at the equilibrium state is: $1 - [s_1 s_2 / (s_1 + s_2)]$

Thus genetic load is $[s_1 s_2 / (s_1 + s_2)] = (0.2)(0.3) / (0.2 + 0.3) = \mathbf{0.12}$

d. Currently $p = q = 0.5$. Is the genetic load now higher or lower than it would be at equilibrium?

Since fitness is at a maximum at the equilibrium state, and $q = 0.5$ is not the equilibrium state for this locus, the genetic load is now higher.

(You can also verify this using unit squares and calculating fitness at the equilibrium and non-equilibrium allele frequencies).

4. Determine the frequencies at generation $t+1$ for the following:

Hint: the following general formula (3.12a) can be used in each of these cases:

$$q_1 = \frac{q_0 [1 - s(hp_0 + q_0)]}{1 - 2hs p_0 q_0 - s q_0^2}$$

However, in the solutions below, the new frequencies were calculated using the original frequency and the change in the frequency of q (3.12b):

$$\Delta q = - \frac{s p q [h - (2h - 1)q]}{1 - 2h s p q - s q^2}$$

Alternatives: shorter specific equations are available for each type of fitness. Or use unit squares to help in calculating.

a. At a biallelic autosomal locus, a lethal recessive allele is found at a frequency of 0.2. Assuming random mating, what will be the frequency of the allele in the next generation?

For a recessive allele, dominance (h) is zero, and the selection coefficient (s) is 1. In this example, q = 0.2. Since p + q = 1, p = 0.8. Using the equation above:

$$\Delta q = - \frac{spq[h - (2h - 1)q]}{1 - 2hspq - sq^2} = \frac{-(1 \times 0.8 \times 0.2)(0 - ((2 \times 0) - 1) \times 0.2)}{1 - (2 \times 0 \times 1 \times 0.8 \times 0.2) - (1 \times 0.2^2)}$$

$$= -((0.8 \times 0.2) \times 0.2) / (1 - 0.2^2) = -0.032 / 0.96 = -0.0333$$

$$q_1 = q_0 + \Delta q = 0.2 - 0.0333 = 0.1667$$

Alternatively, equations 3.4a or 3.4b could be used; these give the same answers.

b. At a biallelic autosomal locus, a deleterious recessive allele with a selection coefficient of 0.3 is found at a frequency of 0.2. Assuming random mating, what will be the frequency of the allele in the next generation?

For a recessive allele, dominance (h) is zero. In this example, the selection coefficient s = 0.3 and q = 0.2. Since p + q = 1, p = 0.8. Using the equation above:

$$\Delta q = - \frac{spq[h - (2h - 1)q]}{1 - 2hspq - sq^2} = \frac{-(0.3 \times 0.8 \times 0.2)(0 - ((2 \times 0) - 1) \times 0.2)}{1 - (2 \times 0 \times 0.3 \times 0.8 \times 0.2) - (1 \times 0.2^2)}$$

$$= -((0.3 \times 0.8 \times 0.2) \times 0.2) / (1 - (0.3 \times 0.2^2)) = 0.0096 / 0.988 = -0.00972$$

$$q_1 = q_0 + \Delta q = 0.2 - .00972 = 0.19028$$

Additionally, given that h = 0, and p = 1 - q, the above formula becomes

$$\Delta q = - \frac{sq^2(1-q)}{1 - sq^2} \text{ which is equation 3.6b (3.6a could also be used)}$$

c. At a biallelic autosomal locus, a deleterious additive allele with a selection coefficient of 0.3 is found at a frequency of 0.2. Assuming random mating, what will be the frequency of the allele in the next generation?

For an additive allele, dominance (h) is 0.5. In this example, the selection coefficient s = 0.3 and q = 0.2. Since p + q = 1, p = 0.8. Using the equation above:

$$\Delta q = - \frac{spq[h - (2h - 1)q]}{1 - 2hspq - sq^2} =$$

$$= \frac{-(0.3 \times 0.8 \times 0.2)(0.5 - (((2 \times 0.5) - 1) \times 0.2))}{1 - (2 \times 0.5 \times 0.3 \times 0.8 \times 0.2) - (0.3 \times 0.2^2)}$$

$$= -(0.048 \times 0.5) / (1 - 0.048 - 0.012) = -0.024 / 0.94 = -0.0255$$

$$q_1 = q_0 + \Delta q = 0.2 - x = 0.1745$$

Additionally, given that $h = 0.5$ for an additive allele, and $q = 1-p$, the original formula becomes

$$\Delta q = - \frac{sq(1-q)}{2(1 - sq)} \quad \text{which is equation 3.7b (3.7a could also be used)}$$

d. At a biallelic autosomal locus, a deleterious allele with a dominance level of 0.4 and selection coefficient of 0.3 is found at a frequency of 0.2. Assuming random mating, what will be the frequency of the allele in the next generation?

In this example, level of dominance (h) is 0.4, the selection coefficient $s = 0.3$ and $q = 0.2$. Since $p + q = 1$, $p = 0.8$. Using the equation above:

$$\Delta q = - \frac{spq[h - (2h - 1)q]}{1 - 2hspq - sq^2}$$

$$= \frac{-(0.3 \times 0.8 \times 0.2)(0.4 - (((2 \times 0.4) - 1) \times 0.2))}{1 - (2 \times 0.4 \times 0.3 \times 0.8 \times 0.2) - (0.3 \times 0.2^2)}$$

$$= -(0.048)(0.44) / (1 - 0.0384 - 0.012) = -0.02112 / 0.9496 = -0.02224$$

$$q_1 = q_0 + \Delta q = 0.2 - 0.0222 = 0.1778$$

e. For the situation described in (d), and assuming the Basic Selection Model, what was the frequency of the three genotypes among only those progeny that were inviable?

Since $p = 0.8$ and $q = 0.2$, the frequency of the three genotypes among progeny were:

$$\begin{aligned} \Pr(A_1A_1) &= P = 0.8^2 = 0.64 \\ \Pr(A_1A_2) &= H = 2pq = 0.32 \\ \Pr(A_2A_2) &= Q = 0.2^2 = 0.04 \end{aligned}$$

The selection coefficient is 0.3, which means that 30% of the homozygotes (A_2A_2) for the deleterious allele were inviable:

$$0.3 \times 0.04 = 0.012$$

The dominance level is 0.4, which means the proportion of inviable heterozygotes is 0.4 times the selection coefficient of 0.3. Among all progeny, the proportion of heterozygote (A_1A_2) and inviable is therefore:

$$0.32 \times 0.4 \times 0.3 = 0.0384$$

For dominant homozygotes, there is no inviability: $0.64 \times 0 = 0$

Thus the frequency of genotypes among inviable individuals was:

$$A_1A_1: 0$$

$$A_1A_2: 0.0384 / (0.0384 + 0.012) = 0.762$$

$$A_2A_2: 0.012 / (0.0384 + 0.012) = 0.238$$

f. At a biallelic autosomal locus, a deleterious dominant allele with a selection coefficient of 0.3 is found at a frequency of 0.8. Assuming random mating, what will be the frequency of the allele in the next generation?

For a dominant allele, the effect on fitness is the same whether the allele is in homozygous or heterozygous state; thus dominance (h) is 1. The selection coefficient $s = 0.3$ and $q = 0.8$. Since $p + q = 1$, $p = 0.2$. Using the equation above:

$$\Delta q = - \frac{spq[h - (2h - 1)q]}{1 - 2hspq - sq^2} =$$

$$= \frac{-(0.3 \times 0.2 \times 0.8)(1 - ((2 \times 1) - 1) \times 0.8)}{1 - (2 \times 1 \times 0.3 \times 0.2 \times 0.8) - (0.3 \times 0.8^2)}$$

$$= \frac{-(0.048)(0.2)}{(1 - 0.096 - 0.192)} = -0.0096 / 0.712 = -0.01348$$

$$q_1 = q_0 + \Delta q = 0.8 - 0.01348 = 0.78652$$

Additionally, given that $h = 1$, and that $p = 1 - q$, the original formula is transformable to

$$\Delta q = - \frac{sq(1 - q)^2}{1 - sq(2 - q)} \text{ which is equation 3.11b; 3.11a could also be used}$$

5. Describe an example of viability differences due to maternal-fetal interactions

One example of viability differences due to maternal fetal interactions is the Rh blood group system in humans. Rhesus factor or the Rh D antigen is present on surface of red blood cells of Rh positive individuals. Fetal mortality may occur (though usually not in first pregnancies) due to production of maternal

anti-RhD antibodies when the mother is Rh negative (genotype rr) while the fetus is Rh positive (i.e. genotype RR or Rr).

6. Define the following terms:

- a. Adaptive or positive Darwinian selection: selection for alleles that are advantageous in the present environment. Fitness **advantage** of s.
- b. Purifying selection: selection that reduces the frequency of deleterious alleles in a population. Fitness **disadvantage** of s.
- c. Number of lethal equivalents: alleles whose summed effect is that of lethality; for example, four alleles each of which would be lethal 25% of the time (or to 25% of their bearers), are equivalent to one lethal allele.
- d. Positive frequency dependent selection: selection favoring common phenotypes, in which the fitness of a phenotype increases as it becomes more common.
- e. Inclusive fitness: fitness of an individual plus his/her effect on the relatives of the individual weighted by their coefficient of relationship
- f. Philopatry: tendency of progeny to remain near their natal (or parental) territory
- g. Coefficient of inbreeding, **f**, is the probability that 2 homologous alleles are identical by descent (IBD)
- h. Red queen process: a coevolutionary “arms race” between 2 or more species, in which continual evolutionary change by a species is necessary to retain its place in an ecosystem because of ongoing co-evolution by other species