Introductory seminar on
“Mathematical Population Genetics”
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Prof. Joachim Hermisson
Ilse Höllinger

2 Selection and Mutation

2.1 Diploid selection model with multiplicative fitness

We consider an autosomal gene with k alleles with frequencies $p_1, \ldots, p_k$ in a diploid population of individuals. We denote the relative frequencies of A_iA_i homozygotes by P_{ii} and of A_iA_j heterozygotes by P_{ij} and assume full random-mating. We assume that k positive constants $v_1, \ldots, v_k$ exist such that the fitness of the A_iA_j genotype $W_{ij} = v_i v_j$ (*multiplicative* fitness).

1. Express the mean fitness $\overline{W}$ in the population as a function of $\bar{v} = \sum_i p_i v_i$.
2. Express the marginal fitness of allele A_i, and the frequency p_i' of allele A_i in the next generation.
3. What model do you recognize (discuss possible differences with the current model)?

2.2 Selection at a single locus with two alleles and a recessive lethal allele

In a randomly mating population we consider a gene with two alleles A and a where the homozygotes aa and heterozygotes Aa have the same phenotype and fitness, whereas the homozygotes AA die before the reproductive stage. The frequencies of the A and a alleles are denoted p and q $(p + q = 1)$.

1. Express the fitnesses W_{11}, W_{12}, and W_{22}. What are the corresponding values for the selection coefficient s and the degree of dominance h?
2. Express the marginal fitnesses of the A and a alleles, as well as the mean fitness of the population $\overline{W}$.
3. Express the frequency of the lethal A allele at the next generation.
4. Give the frequency of the A allele after t generations, as a function of t and the initial frequency of the A allele.

2.3 Diploid case with two alleles mutation and selection, without backward mutation

We consider the two-alleles case in discrete time, in a randomly mating population of diploid individuals where the fitness values of the genotypes aa, Aa and AA are $W_{11} = 1$, $W_{12} = 1 - hs$, and $W_{22} = 1 - s$, respectively (we assume $0 < s < 1$). The frequencies of a and A are denoted q and p. The mutation rate from a to A is denoted μ, the backward mutation (from A to a) is assumed to be zero.

1. Express the marginal fitnesses of the a and A alleles, as well as the mean fitness in the population.
2. Express the allele frequencies p' and q' at the next generation.
3. Show that the equilibria, $\hat{p}_{1,2} \neq 0$ are solutions of a quadratic equation.
4. Express the values of the equilibrium points and study their stability in the special case when $h = \frac{1}{2}$.

2.4 Limiting cases of the mutation-selection case with two alleles

For the two non-trivial solutions $\hat{p}_{1,2}$ obtained in exercise 2.3 we will now investigate some limiting cases.

1. If $h = 0$ prove
$$\hat{p}_1 = \sqrt{\frac{\mu}{s}}$$
.
2. If $h \gg \sqrt{\frac{\mu}{s}}$ show
$$\hat{p}_1 \approx \frac{\mu}{hs}$$
3. Show that if
$$h > \frac{1 - \frac{\mu}{s}}{1 - \mu}$$
the second equilibrium $\hat{p}_2$ is admissible, but unstable and satisfies
$$\hat{p}_2 \approx \frac{h}{2h - 1} - \frac{\mu}{hs}$$
4. Finally prove for mutiplicative selection coefficients $W_{11} = 1$, $W_{12} = 1 - t$, and $W_{22} = (1 - t)^2$ that one obtains exactely
$$\hat{p}_1 = \frac{\mu}{t}$$