Introductory seminar
"Mathematical Population Genetics"
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1 Hardy Weinberg Law

1.1 Blood types

The ABO blood types can (in the simplest case) be coded by the three alleles A, B, and O at a single locus, where A and B are dominant over O (genotypes AO and BO respectively have phenotypes $[A]$ and $[B]$).

1. Denote with p_A p_B and p_O the frequencies of these alleles. Calculate, under the assumption of Hardy-Weinberg equilibrium, the relative frequencies of the blood types A, B, AB and O (phenotypes $[A]$, $[B]$, $[AB]$, and $[O]$). Why exactly these?
2. Let R_A, R_B, R_{AB} and R_O denote the observed frequencies of the respective blood types within a population. From this calculate the allelic frequencies. Is it possible to do this without Hardy-Weinberg? How could one test wether the population is in Hardy-Weinberg equilibrium?

1.2 Dynamics of phenotype frequencies under assortative mating

Mating can be non-random with respect to some traits, respectively genes. We speak of assortative mating when similar individuals are more likely to mate with each other than expected by chance. Consider the following simple but instructive example. The two alleles A_1 and A_2 occur at a gene locus. Furthermore, the genotype A_1A_1 has the same phenotype as A_1A_2, which is different from the phenotype of A_2A_2. Thus A_1 is dominant. Let ρ denote the proportion of individuals that mate assortatively, *i.e.* only with individuals of the same phenotype. This proportion is assumed to be identical for both phenotypes. Denote the (ordered) genotype frequencies with P_{ij} and the allelic frequencies with p and q, $(p + q = 1)$.

1. The contribution of those individuals that mate randomly to the pool of individuals with genotype A_1A_1, A_1A_2 and A_2A_2 in the next generation is then $(1-\rho)p^2$, $(1-\rho)2pq$ and $(1-\rho)q^2$ respectively (why?).

2. Calculate the contribution of the assortatively mating individuals of genotype A_2A_2 to the different types of offspring and also the contributions of the individuals with the dominant phenotype.

3. Show that the following recursion holds:

$$P'_{11} = (1-\rho)p^2 + \rho\frac{p^2}{1-P_{22}}$$

1.3 Continued: Dynamics of phenotype frequencies under assortative mating

For details see exercise before.

1. Derive also the recursion for the other two genotypes, P_{12}, P_{22}.

2. Which conclusions can you draw from this set of recursions for the allelic frequencies.

3. What recursion follow for the special cases of $\rho = 0$ and $\rho = 1$?

1.4 Dynamics of heterozygosity under assortative mating

We are considering the assortative mating case discussed in exercise 1.2,1.3. The quantity $H = 2P_{12}$ is known as *heterozygosity*.

1. Derive the recursion for H' (which can be expressed as a function of H and ρ).

2. For the case $\rho = 1$, derive the value of $H(t)$ in generation t given e.g. $p(t=0) = \frac{1}{2}$.

3. What general conclusion can be derived for $\lim_{t\to\infty} H(t)$ in the case of $\rho < 1$?

1.5 Dynamics of phenotype frequencies with X-linkage

Consider a gene with two alleles A and a, located on the X chromosome (in mammals, females are XX while males are XY). The A allele is dominant (individuals with genotypes AA and Aa have phenotype $[A]$ while individuals with genotype aa have phenotype $[a]$). The frequencies of the A allele in males and females are respectively denoted p and q, initial frequencies are denoted p_0 and q_0.

1. Express the recursion for the allele frequencies p' and q'.

2. Express the allele frequencies in males and females after t generations.

3. Express the frequency of the $[A]$ phenotype in females and in males.

4. Does this dynamics change if the females:males ratio differs from 1:1 in the population?