Genetic aspects of populations, Hardy-Weinberg equilibrium

Population

What is a population from genetic point-of-view? It is a group of **inbreeding individuals of the same species** that inhabit the same space (prescribed geographical area) and time. The key to a population is that they must be able to interbreed.

Within any given population there is variation (differencies) of/in:

- **phenotypes;** the proportion of individuals within a population that are of a particular phenotype is phenotype frequency
- **genotypes;** the proportion of individuals within a population that are of a particular genotype is genotype frequency
- **alleles;** the proportion of all copies of a gene in a population that are of a particular allele type is allelic frequency

The gene pool is the sum total of all of the alleles (of one locus) present and carried by the population.

- Gametic gene pool – sum of all alleles in gametes.
- Zygotic gene pool – sum of all alleles in zygotes.

For a gene with 2 alleles, A and a:

- N_{AA} is the number of AA homozygotes
- N_{Aa} is the number of heterozygotes Aa
- N_{aa} is the number of aa homozygotes
- $N_{AA} + N_{Aa} + N_{aa} = N$, number of individuals in population

Estimating/calculating of allele frequencies in a population

- **with three distinct phenotypes for a trait (e.g. in MN blood group system)**

  Let p = frequency of allele A, and q = frequency of a. Then:
  $pA = (2N_{AA} + N_{Aa}) / 2N$
  $qa = (2N_{aa} + N_{Aa}) / 2N$
  Variant procedure: Calculating allele frequencies from (known) frequencies of genotypes AA, Aa, aa
  $pA = f_{AA} + ½ f_{Aa}$
  $qa = f_{aa} + ½ f_{Aa}$
  $p + q = 1$

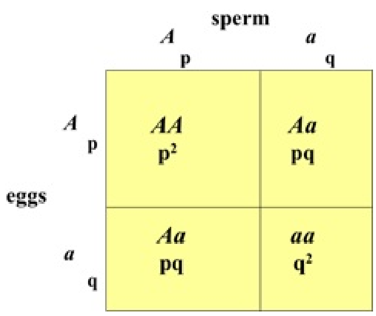
- where (only) two distinct phenotypes exist (i.e., in allelic relation of full dominance/recessivity); estimate of frequency of unfavourable (mutant, deleterious, recessive) allele (Rh blood group system, tasting of PTC or AR diseases)

$$q_{(a)} = \sqrt{\frac{\text{number of recessive homozygotes}}{\text{number of all individuals in the sample}}} = \sqrt{\text{frequency in population}}$$

Genotype frequencies

If frequency of allele A in a population is p, frequency of allele a in a population is q:

- the probability that both the egg and the sperm contain the A allele is $p \times p = p^2$
- the probability that both the egg and the sperm contain the a allele is $q \times q = q^2$
- the probability that the egg and the sperm contain different alleles is $(p \times q) + (q \times p) = 2pq$



Hardy-Weinberg Equation

$$p^2_{(AA)} + 2\,pq_{(Aa)} + q^2_{(aa)} = 1$$

Hardy-Weinberg law

A population that is not changing genetically is in Hardy-Weinberg equilibrium (1908). It comes if these 5 assumptions are correct:

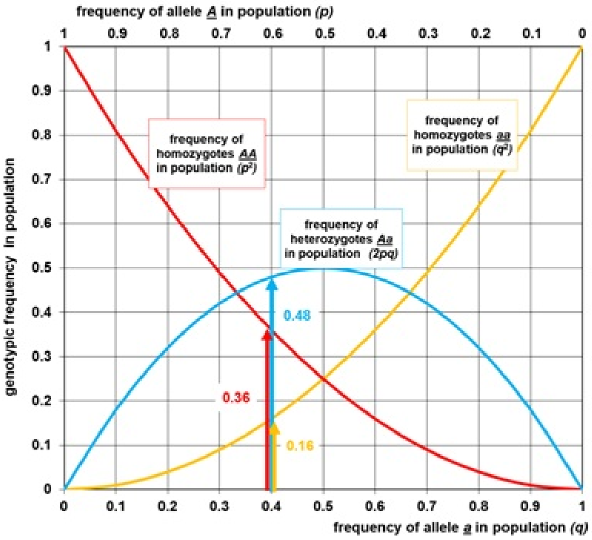
- Random mating (panmixia)
- Large population size (N approaching infinity)
- No migration between populations
- No (or negligible) mutations
- Natural selection does not affect alleles being considered

If these assumptions are true, it follows that:

- Allele frequencies remain constant from one generation to the next
- After one (or more) generations of random mating (breeding), the genotype frequencies (for a 2-allele gene with allele frequencies p, q) are in the proportions: $\mathbf{p^2_{(AA)} : 2pq_{(Aa)} : q^2_{(aa)}}$, and population will be in H-W equilibrium. Var.: H-W equilibrium in a large population will be reached after one generation of (random) breeding.
- For a population to be in Hardy Weinberg equilibrium, the observed genotype frequencies must match those predicted by the equation $p^2 + 2pq + q^2$.

Graphic demonstration of H-W equilibrium

(relation between frequencies of alleles and frequencies of genotypes)



Multiple alleles

Multinomial expansion for two alleles a and b with frequencies p and q $p^2 + 2pq + q^2$ is a binomial expansion of $(p + q)^2$

$$p^2(AA) + 2pq(Aa) + q^2(aa) = (p + q)^2 = (1)^2 = 1$$

For three alleles a, b and c with frequencies p, q and r, the multinomial expansion is $(p + q + r)^2$ which expands into: $p^2 + q^2 + r^2 + 2pq + 2pr + 2qr$, where the first 3 terms being homozygotes and the remaining three heterozygotes.

$$\mathbf{p + q + r = 1} \quad p^2 + q^2 + r^2 + 2pq + 2pr + 2qr = 1$$