

# Population polymorphisms and their causes

- A **Population** where the gene frequency of the most common allele is less than or equal to **0.99** (99%) is **polymorphic for a given trait**.
- Of course, this stated value is not an objective limit, but was only **determined by agreement**.
- It is most convenient to determine the degree of polymorphism using **heterozygosity**, which is defined as:

$$H = 1 - \sum_{i=1}^m x_i^2$$

where  $m$  = number of alleles of the monitored gene and  $x_i$  = gene frequency of the  $i$ th allele (C-H-W applies:  $x_1 + x_2 + \dots + x_m = 1$ )

or verbally as the representation of individuals in a population who are heterozygous for a particular locus.

- **Example:** In the population, the allele representation is  $p=0.5$  and  $q=0.5$ .

$$H = 1 - \sum_{i=1}^m x_i^2 = 1 - 0,5^2 - 0,5^2 = 1 - 0,25 - 0,25 = 0,5$$

This is also the maximum that can be achieved. It is true that the larger  $m$  and the more unevenly distributed frequency  $x$ , the smaller  $H$  is.

The minimum would be for  $p \rightarrow 1$  and  $q \rightarrow 0$  ( $H \approx 0$ ), where the vast majority of homozygotes would be.

- **Heterozygosity** can therefore serve us as a good measure between subpopulations of one population.

## Stable polymorphism

- gene frequencies do not change;
- e.g. population in C-H-W equilibrium, or polymorphism maintained by the frequency of heterozygotes, or mutations and back mutations.

## Transitive polymorphism

- In a population, when due to selection one allele is gradually replaced by another, as is the case, for example, with selection against homozygotes.

## Links

### Related Articles

- Castle-Hardy-Weinberg Equilibrium
- Nucleic acid polymorphisms
- Selection
- Population

### Source

- ŠTEFÁNEK, Jiří. *Medicína, nemoci, studium na 1. LF UK* [online]. [cit. 11/02/2010]. <<https://www.stefajir.cz/>>.