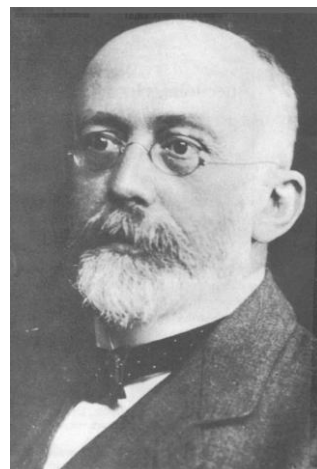


Introductory population genetics and Hardy-Weinberg law

Contents: Definition; Hardy-Weinberg law; Limiting factors of the law; Derivation of the Hardy-Weinberg equation; Proof of the Hardy-Weinberg law; Calculation of gene and genotype frequencies in a population; Suggested reading.

Definition

Population genetics is a branch of Genetics that deals with the inheritance of allelic frequencies (*i.e.* gene and genotype frequencies) in a population. It is to be noted that the Mendelian or classical genetics does not give an idea of the frequencies of the alleles in a sample or population.



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Fig 13.1 G. H. Hardy (left) and W. Weinberg (right)

Hardy-Weinberg law

In a large random mating population (such population is called panmictic population), the relative frequencies of alleles of a trait tend to remain constant from generation to generation only if there is no selection, mutation, migration, genetic drift, meiotic drive etc. in operation.

British mathematician Geoffrey H. Hardy and German physician Wilhelm Weinberg demonstrated the above properties of a population independently in 1908.

Limiting factors of Hardy-Weinberg law

1. Selection,
2. Mutation,
3. Migration,
4. Genetic drift,
5. Meiotic drive,
6. Small population and assortative mating etc.

Background information

- Peter Fox (1932) discovered that in human population, many persons taste a weak solution of PTC (phenyl-thio-carbamide, $C_7H_8N_2S$) bitter, while others find it tasteless.
- Later on, it was discovered that this human trait is controlled by a pair of autosomal alleles, T and t, where T is completely dominant over t ($T > t$).
- So, the tasters belong to genotypes TT and Tt, whereas the non-tasters are always of the genotype tt.

Derivation of the Hardy-Weinberg equation

- Let's consider that a human population is initially composed of 1000 individuals, with equal numbers of homozygous tasters TT (say, 500) and non-tasters tt (say, 500), where $p = 0.5$ and $q = 0.5$;
- Since marriages take place at random, the population is panmictic with respect to this trait;
- In parental generation (P), the possible marriages are: TT×TT, TT×tt, and tt×tt;
- Types of gametes are: T and t;
- Let p = frequency of the dominant allele (T) and q = frequency of the recessive allele (t);
- The gametes and their combinations can be shown in the following checker board or Punnett square:

| | | |
|-------|--------------|--------------|
| ♀/♂ | T (p) | t (q) |
| T (p) | TT (p^2) | Tt (pq) |
| t (q) | Tt (pq) | tt (q^2) |

The expected genotypic frequencies of the F_1 generation are-

$$\begin{aligned} &= p^2 + 2pq + q^2 = 1.00 \text{ (since frequency of all alleles must add to unity)} \\ \text{or, } &(p + q)^2 = 1.00 \\ \text{or, } &p = 0.5 \text{ and } q = 0.5 \end{aligned}$$

Proof of the Hardy-Weinberg law

According to the Hardy-Weinberg equation, $p^2 + 2pq + q^2 = 1.00$, which is also known as the equation of genetic equilibrium.

In F_1 generation, the population will consist of 25% homozygous tasters (TT), 50% heterozygous tasters (Tt) and 25% non-tasters (tt); that means, there are 75% tasters and 25% non-tasters in the population, which supports Mendelian monohybrid ratio of 3: 1.

Now, according to Hardy-Weinberg law, the frequencies of T and t alleles in the F₂, F₃ and subsequent generations will remain the same as in the P generation.

Let us prove the above proposition.

The possible genotypes in the F₁ generation = TT, Tt and tt;

The possible mating types will be = 3 × 3 = 9

| Mating genotypes | Frequencies of mating genotypes | Frequencies of TT | Frequencies of Tt | Frequencies of tt |
|------------------|---------------------------------|-------------------|-------------------|-------------------|
| TT × TT | p^4 | p^4 | - | - |
| TT × Tt | $2p^3q$ | p^3q | p^3q | - |
| TT × tt | p^2q^2 | - | p^2q^2 | - |
| Tt × TT | $2p^3q$ | p^3q | p^3q | - |
| Tt × Tt | $4p^2q^2$ | p^2q^2 | $2p^2q^2$ | p^2q^2 |
| Tt × tt | $2pq^3$ | - | pq^3 | pq^3 |
| tt × TT | p^2q^2 | - | p^2q^2 | - |
| tt × Tt | $2pq^3$ | - | pq^3 | pq^3 |
| tt × tt | q^4 | - | - | q^4 |

$$\text{Total frequencies} = p^4 + 4p^3q + 6p^2q^2 + 4pq^3 + q^4$$

$$\text{Total of TT} = p^4 + 2p^3q + p^2q^2$$

$$\text{Total of Tt} = 2p^3q + 4p^2q^2 + 2pq^3$$

$$\text{Total of tt} = p^2q^2 + 2pq^3 + q^4$$

$$\text{Total frequencies} = p^4 + 4p^3q + 6p^2q^2 + 4pq^3 + q^4 = (p^2 + 2pq + q^2)^2 = 1.00$$

$$\text{Total of TT} = p^4 + 2p^3q + p^2q^2 = p^2(p^2 + 2pq + q^2) = p^2$$

$$\text{Total of Tt} = 2p^3q + 4p^2q^2 + 2pq^3 = 2pq(p^2 + 2pq + q^2) = 2pq$$

$$\text{Total of tt} = p^2q^2 + 2pq^3 + q^4 = q^2(p^2 + 2pq + q^2) = q^2$$

So, in the F₂ generation, the expected frequencies of the genotypes in the population will be:

$$p^2 + 2pq + q^2 = 1.00$$

It is therefore concluded that the genotype frequencies will remain the same in the F₃, F₄ and subsequent generations.

Calculation of gene and genotype frequencies in a population

Problem 1: In a sample of 528 school students, 402 were tasters to PTC. Calculate the frequencies of the dominant and recessive genes and all three genotypes.

Solution:

According to the given problem, the number of tasters and non-tasters in the population are as follows:

Total number of students = 528
Number of tasters = 402
So, the number of non-tasters = 126
Hence, frequency of non-tasters (tt), $q^2 = 126 \div 528 = 0.24$
Therefore, frequency of the recessive gene, $q = \sqrt{0.24} = 0.49$
And frequency of the dominant gene, $p = 1 - 0.49 = 0.51$ (since $p + q = 1.00$)

| | | |
|---|-----------------------------------|--------|
| So, frequency of the homozygous tasters (TT), | $p^2 = (0.51)^2$ | = 0.26 |
| Frequency of the heterozygous tasters (Tt), | $2pq = 2 \times 0.51 \times 0.49$ | = 0.50 |
| And frequency of the non-tasters (tt) | $q^2 = (0.49)^2$ | = 0.24 |

Total genotypic frequencies, $p^2 + 2pq + q^2 = 1.00$

Problem 2: In a population of 1000 university students, 760 were tasters to PTC. Calculate the frequencies of the dominant and recessive genes along with their genotype frequencies in the population.

Answer to the Problem 2

Gene frequencies:

$$p = 0.51$$

$$q = 0.49$$

Genotype frequencies:

$$TT (p^2) = 0.26$$

$$Tt (2pq) = 0.50$$

$$tt (q^2) = 0.24$$

Suggested reading:

Falconer, DS. 1989. *Introduction to Quantitative Genetics* (3rd edn)

Gardner *et al.* 1991. *Principles of Genetics* (8th edn)

Islam, MS. 2018. *Selected Lectures on Genetics*. LAP Lambert Academic Publishing, Germany.

Li, CC. 1976. *First Course in Population Genetics*

Sinnott *et al.* 1973. *Principles of Genetics* (5th edn)

Stansfield, WD. 1991. *Theory and Problems of Genetics* (3rd edn)

Wikipedia: www.wikipedia.com

ইসলাম, ম.সা., খান, হা.সা. ও রানা, ম.হা.তা. ২০১৭। *জেনেটিক্স: মিল ও অমিলের বিজ্ঞান*। অন্যপ্রকাশ, ঢাকা।