

## Biology 3A Laboratory Mendelian, Human and Population Genetics

### OBJECTIVE

- Calculate Hardy-Weinberg equation.

### INTRODUCTION – Population Genetics

During 1908, G. H. Hardy, an English mathematician, and G. Weinberg, a German physician, independently solved why genetic variation exist in populations. They discovered that sexual reproduction alone will do nothing to change the frequencies of alleles in population. Meaning that dominant alleles will not replace recessive alleles as long as populations met the following five conditions:

1. Population size is very **large** so that random chance events will not affect allelic frequencies.
2. Mating between members of the population occur at **random**.
3. There must be no **net mutation** of one allele to the other allele.
4. There is **no gene flow** between populations due to immigration or emigration.
5. There must be **no natural selection** against either allele.

If all five conditions are perfectly met, the frequency of each allele will remain exactly the same from generation to generation. Because these proportions are not changing, the population is considered to be in **Hardy-Weinberg equilibrium**.

Suppose that the frequency of *A* allele is *X*, and there are two alleles for this gene. The frequency of the second allele, *a*, has to be equal to  $1 - X$  because the sum of the two alleles must be 100% or a frequency of 1. The chance of getting the genotype *AA* is the chance of getting one *A* allele times the chance of getting a second,  $X^2$ . To avoid confusion, the frequency of the dominant allele is called *p*, so the frequency of the recessive allele is called *q*. For any gene that has two alleles, *p* plus *q* must always equal 1 ( $p + q = 1$ ). The chance of getting the *AA* genotype is  $p^2$ . The of getting the *aa* genotype is  $q^2$ . The heterozygous genotype can be formed by getting a recessive and a dominant allele ( $q \times p$ ) or the dominant and recessive allele ( $p \times q$ ). Therefore, the chance of the heterozygous genotype, *Aa*, is  $qp + pq$  or  $2pq$ . Knowing this, one can make predictions regarding genotypic frequencies for successive generations of a population in Hardy-Weinberg equilibrium once *p* and *q* are known. The following equation is the binomial expansion of  $(p + q)^2$ .

<i>p</i> = frequency of the dominant allele	Frequency:	$p^2$	+	$2pq$	+	$q^2$
<i>q</i> = frequency of the recessive allele	Genotype:	AA		Aa + aA		aa

If more than two alleles are involved, more unknowns can be added and each term will represent the frequency of a corresponding genotype.

The frequency of phenotypes can then be stated once the frequencies of the genotypes are known:

Dominant phenotype (A-):  $AA + Aa \rightarrow p^2 + 2pq$

Recessive phenotype (aa):  $q^2$

Because the frequencies of the dominant and recessive phenotypes must also add up to 1, the second frequency is known as soon as one or the other is calculated.

The Hardy-Weinberg Law is very useful because deviations from the Hardy-Weinberg frequencies indicate the effects of selection, non-random mating or other factors that influence the genetic make up of the population.

For this experiment, your lab section will constitute a **population** of organisms of the same species that occur in the same area that interbreed or share a common **gene pool**, all the alleles at all gene loci of all the individuals in the population. We will calculate Hardy-Weinberg from our data regarding PTC and sodium benzoate tasting.

In actual populations, it is usually not possible to tell genotypes of all the individuals, especially dominant phenotypes. We will not be able to differentiate homozygous dominant (*AA*) individuals from heterozygous dominant individuals (*Aa*). The phenotype that is easily observed or measured is the recessive phenotype ( $aa = q^2$ ). Once the recessive frequencies are determined, we can calculate the dominant frequencies since all frequencies must add up to 1 ( $p^2 + 2pq + q^2 = 1$  and  $p + q = 1$ ).