

EXPERIMENT 4

AIM: Calculate genotypic, phenotypic and allele frequencies from the data provided.

BACKGROUND/THEORY: Transmission of Mendelian traits from parents to progeny is essential for analyses of variation and variability in large groups of organisms. Heritable phenotypic variation is subjected to the five major evolutionary forces leading to changes in the phenotype of the individuals in different groups. During the 1930s, several researchers including R A Fisher, worked to reconcile Mendelism with Darwinism, it led to the rise of the field of **population genetics**, in which speciation was seen as resulting from genetic changes within a lineage as reflected in changes in gene frequency. Thus, evolution operates at level of genes, organisms and populations. Thus, a comprehensive definition of evolution would be “**descent with modification, encompassing evolutionary change at genetic, organismal and/or population levels**”.

Population genetics is a field of study that deals with understanding the causes of the observed patterns of genetic variation within populations (or gene pools) and divergence among populations. It uses both empirical tools, measuring variation in natural populations, and theoretical tools, which attempt to explain the observed variation with quantitative modeling.

Two approaches/tools are used to study genetic structure of populations:

- **Empirical tools (experimental)** - these are used for measuring variation in natural populations.
- **Theoretical tools (hypothetical)** - these are used to explain the observed variation with quantitative modeling.

The relationship between genotypes and phenotypes in populations, was proposed independently by Godfrey H. Hardy (1877–1947), a British mathematician and Wilhelm Weinberg (1862–1937), a German physician.

The **HARDY-WEINBERG LAW** serves as a foundation for population genetics. It offers a simple explanation for how the Mendelian principles of segregation influence allele and genotype frequencies in a population. It explains what happens to the allele and genotype frequencies of a population as the alleles are passed from generation to generation in the **absence of evolutionary forces**.

The Hardy–Weinberg law is divided into three parts- a set of assumptions and two major results.

Part 1 (Assumptions): In an infinitely large, randomly mating population, free from mutation, migration, and natural selection (note that there are five assumptions);

Part 2 (Result 1): the frequencies of the alleles do not change over time, where **p** is the allele frequency of **dominant allele A** and **q** is the allele frequency of **recessive allele a**; and

Part 3 (Result 2): the genotypic frequencies remain in the proportions (frequency of AA), (frequency of Aa), and (frequency of aa). **The sum of the genotype frequencies equals 1** (frequency is a proportion and it always ranges between 0 and 1).

MATHEMATICAL NOTATIONS and CALCULATIONS

- **Allele frequency** is calculated using the formula $p + q = 1$
- **Genotype frequency** is calculated using $(p+q)^2 = 1$
- where **p** is the *allele frequency of dominant allele A* and **q** is the *allele frequency of recessive allele a*

Calculation of genotype frequencies at a specific locus (f):

f = number of individuals with a particular genotype/ total number of individuals in the population

Calculation of allele frequency of recessive /dominant allele:

f(A) or f(a) = Number of copies of a given allele / Sum of counts of all alleles in the population

For a single locus with two alleles A & a, alleles are expected to combine into genotypes following the simple laws of probability—AA , Aa , aa.

Table 1. Possible combinations of A and a gametes from gametic pools for a population.

Table 1. Genotype numbers and genotype frequencies in a hypothetical population.

Genotypes	Number with these Genotypes	Genotype Frequencies
AA	180	0.36
Aa	240	0.48
aa	80	0.16
Total	500	1.0

(Reference iGenetics, Russell)

Allele frequencies can be calculated using the genotypic frequencies using the following equations:

1. The Hardy- Weinberg equations are modified when applied to X-linked traits and to multiple allelic series.
2. **Hardy Weinberg Law in Plant Evolution:** Plant breeding deals with the alteration of the traits of plants to generate desired characteristics. In population genetics, the most frequently used mathematical model is the **Hardy–Weinberg Equilibrium** (HWE). The genotype and allele frequencies of future generations can be computed with the help of this principle. The equilibrium of earlier and current populations can also be interpreted from this principle.
3. Hence, the HWE has ecological significance. The facultative clonal plants are inherently problematic subjects for the application of the Hardy Weinberg principle. The problematic areas or assumptions that are not followed according to HWE are the generations overlapping in the case of clonal plants. Life spans of these plants are extreme i.e; some live for a short span and some for a long span. Hence, the study of generations cannot be done in such cases.
4. In the case of dioecious plants, the criteria of equal sex ratio are not maintained. In spite of these limitations, HWE can be used to obtain values such as expected heterozygosity or fixation index. In plants where clonality is not maintained, the **Hardy-Wienberg principle** can be used to calculate the genotype frequencies, it is extensively used in plant breeding to select for appropriate varieties.
5. The random-mating assumption is often violated in breeding populations because breeding populations are smaller than natural plant populations. Thus, a mating design that minimizes gamete (allele) sampling errors is an important consideration. The breeder must be aware of several factors:
6. Self-pollinated population — allele frequency will remain in equilibrium (assuming a sufficiently large population, no selection, or other factors that disturb equilibrium).

However, with each successive generation of self-pollination, the genotype frequency of homozygous loci will increase and the frequency of heterozygous loci will decrease.

7. Ultimately, the heterozygous genotype will be eliminated from the population with continued selfing.
8. Cross-pollinated population — sampling errors occur if plants in the population differ in their vigor, time of flowering, or mate more frequently with plants in close proximity.
9. Selection for or against a particular allele will alter the allele and genotype frequencies of the population. Selection against a dominant allele (i.e., selection for homozygous recessive) will remove the dominant allele from the population in a single generation.
10. Selection against a recessive allele will require more than a few generations to remove the recessive allele from the population because the homozygous dominant and heterozygous genotypes have indistinguishable phenotypes.
11. In addition to being able to estimate allele and genotype frequencies, the breeder also needs to understand the gene action affecting the character of interest. The breeding of cross-pollinated crops differs from self-pollinated species because of differences in the structures of their gene pools and opportunity for genetic recombination.

Reference:

1. <https://iastate.pressbooks.pub/cropgenetics/chapter/population-genetics-2/>
2. <https://www.nature.com/scitable/topicpage/the-variety-of-genes-in-the-gene-6526291/>

PROBLEMS & SOLUTIONS

CASE 1: Petal coloration of pea plants has a complete dominance relationship where purple petals are dominant over white petals. There are 276 plants, 273 have purple petals. Find:

- a. the frequency of the dominant and recessive alleles
- b. the frequency of individuals with the dominant, heterozygous, and recessive genotype.
- c. In the next generation, of total 552 plants, 546 have purple petals. Is the population in Hardy-Weinberg Equilibrium? Solve for p and q.

CASE 2: In a wild prairie grass, the genotypes with at least one dominant allele T produce plants that are 100 cm tall. The homozygous recessive (tt) is 50 cm tall. If the frequency of allele t is 0.6. Calculate the frequency of individuals that are homozygous dominant and heterozygous.

CASE 3: In Petunia, multiple alleles at a single locus determine flower color. The allele for Red (C^B) is dominant to the allele for pink (C^P) and to the allele for yellow (C^Y). The dominance hierarchy among these alleles is $C^B > C^P > C^Y$. In one population sample, the following color phenotypes were recorded:

Red - 236 Pink - 231 Yellow - 33.

Assuming that this population is in Hardy–Weinberg equilibrium (large, randomly mating, and free from evolutionary processes), Calculate the frequencies of the alleles. How can the equilibrium be altered in the next generation?

CASE 4: In a population of maize plants, a transgene for herbicide resistance has been introduced by chance contamination of 500 seeds from a homozygous transgenic plant. A farmer plants 10,000 seeds and makes selections for the best performing vigorous plants after herbicide spray. Calculate the frequency of the transgene (**note that this locus does not exist in the natural / non transgenic plants**). In the next generation if cross is made between a transgenic and wild type plant will the Hardy Weinberg equilibrium be maintained?