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ZO 354: Genetics

Semester V

By

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Population genetics

Population genetics

Study of the frequency of genes and genotypes in a mendelian population is known as population genetics. In other words, it is a branch of genetics which deals with the frequency of genes and genotypes in mendelian populations. Before dealing with population genetics, it is essential to define mendelian population, gene frequency and genotype frequency.

There are two important features of Mendelian population, viz:

- 1) Random mating, and
- 2) Equal survival of all genotypes.

In case of random mating, each individual of one sex has equal chance of mating with every individual of opposite sex. In other words, there is no restriction on mating of one individual with other individuals. Such inter-mating populations are also known as panmictic populations.

Random mating populations maintain high level of variability and adaptability. Random mating individuals belong to the same species and same gene pool. The gene pool refers to the sum total of genes in a mendelian population.

Gene Frequency:

Gene frequency refers to the proportion of different alleles of a gene in a random mating population. It is also known as genetic frequency. In other words, the proportion of each type of allele at a particular locus in a random mating population is referred to as gene frequency. The composition of a population is described in terms of gene frequencies.

Estimation of gene frequencies in a population consists of three important steps as given below:

1. Sampling: First a random sample of individual is drawn from the random mating population under study. The size of sample should be adequate to represent all the individuals of a population.

2. Classification:

After sampling, the individuals are grouped into different classes for a gene and their number is counted.

3. Calculation of gene frequency:

Suppose a random sample of 100 individuals was drawn from a random mating population of four 'O' clock plant (*Mirabilis jalapa*). Out of 100 plants, 30 were with red flower, 40 with pink flower and 30 with white flower.

Now, the allele frequency will be worked out as follows:

(a) In four o'clock plant, a cross between red and white flowered strains produces pink flower in F_1 and red, pink and white flowered plants in 1 : 2 : 1 ratio in F_2 generation. Thus, plants with red colour are homozygous for dominant allele (RR) and individuals with white flower colour are homozygous for recessive allele (rr).

(b) Each heterozygous individual with pink colour will have dominant (R) and recessive (r) alleles in equal number.

1. Number of R alleles in the Sample (30 individuals)

= 2 (No. of red individuals) + No. of pink individuals

$$= (2 \times 30) + 40 = 100$$

2. Proportion of R alleles in the sample

= Number of RR Alleles/2 (Total plants in a sample)

$$= 100/(2 \times 100) = 100/200$$

$$= 0.50$$

Similarly, the number of r alleles

$$= (2 \times 30) + 40 = 100$$

Proportion of r alleles

$$= 100/(2 \times 100) = 100/200$$

$$= 0.50$$

Therefore, the frequency of RR and rr alleles is 0.50 each.

Genotype Frequency:

It refers to the ratio of different genotypes in a mendelian population. Genotypic frequency is also known as zygotic frequency. The estimation of genotypic frequency for a gene in a population also consists of three important steps mentioned above.

Thus, the genotypic frequency of three types of individuals from the above sample will be calculated as ratio of each individual, class or genotypes to the total individuals in a sample. Thus,

1. Frequency of Red (RR) individuals = $30/100 = 0.30$

2. Frequency of Pink (Rr) individuals = $40/100 = 0.40$

3. Frequency of White (rr) individuals = $30/100 = 0.30$

Hardy-Weinberg Law:

Foundation of population genetics was laid by G.H. Hardy, an English mathematician and W. Weinberg, a German physician in 1908. They independently discovered a principle concerned with the frequency of genes (alleles) in a population. Their principle is commonly known as Hardy-Weinberg Law.

The Hardy-Weinberg Law states that:

1. In a random mating population, the frequency of genes and genotypes remains constant generation after generation, if there is no selection, mutation, migration and random genetic drift.
2. They also developed a mathematical relationship to describe the equilibrium between alleles. According to this relationship, the frequencies of three genotypes for a single locus with two alleles (A and a) are in the ratio of $P^2AA : 2PqAa : q^2aa$, where P and q are the frequencies of allele A and 'a' respectively. $P + q$ are always equal to 1 or $P = q = 0.50$.
 $P + q = 1$ or $P = 1 - q$ and $q = 1 - P$

Hardy-Weinberg Equilibrium Hardy-Weinberg Equilibrium, also referred to as the Hardy-Weinberg principle, is used to compare allele frequencies in a given population over a period of time. A population of alleles must meet five rules in order to be considered "in equilibrium": 1) No gene mutations may occur and therefore allele changes do not occur. 2) There must be no migration of individuals either into or out of the population. 3) Random mating must occur, meaning individuals mate by chance. 4) No genetic drift, a chance change in allele frequency, may occur. 5) No natural selection, a change in allele frequency due to environment, may occur.

Hardy-Weinberg Equilibrium never occurs in nature because there is always at least one rule being violated. Hardy-Weinberg Equilibrium is an ideal state that provides a baseline against which scientists measure gene evolution in a given population. The Hardy-Weinberg equations can be used for any population; the population does not need to be in equilibrium.

There are two equations necessary to solve a Hardy-Weinberg Equilibrium question:

$$p + q = 1$$

$$p^2 + 2pq + q^2 = 1$$

p is the frequency of the dominant allele.

q is the frequency of the recessive allele.

p^2 is the frequency of individuals with the homozygous dominant genotype.

$2pq$ is the frequency of individuals with the heterozygous genotype.

q^2 is the frequency of individuals with the homozygous recessive genotype.

Example 1a:

A population of cats can be either black or white; the black allele (B) has complete dominance over the white allele (b). Given a population of 1,000 cats, 840 black and 160 white, determine the allele frequency, the frequency of individuals per genotype, and number of individuals per genotype. To solve this problem, solve for all the preceding variables (p , q , p^2 , $2pq$, and q^2)

Step 1: Find the frequency of white cats, the homozygous recessive genotype, as they have only one genotype, bb. Black cats can have either the genotype Bb or the genotype BB, and therefore, the frequency cannot be directly determined.

Frequency of individuals = individuals/ Total populations

$$=160/1000$$

$$=0.16$$

Frequency of white cats = **0.16**; therefore, $qq^2 = 0.16$

Step 2: Find q by taking the square root of q^2 .

$$\sqrt{q^2} = \sqrt{0.16}$$

$$q = 0.4$$

Step 3: Use the first Hardy-Weinberg equation ($p + q = 1$) to solve for p .

$$p + q = 1$$

$$p = 1 - q$$

$$p = 1 - (0.4)$$

$$p = 0.6$$

Now that the allele frequencies in the population are known, solve for the remaining frequency of individuals by using $p^2 + 2pq + q^2 = 1$.

Step 4: Square p to find p^2 .

$$p = 0.6$$

$$p^2 = (0.6)^2$$

$$p^2 = 0.36$$

Step 5: Multiply $2 \times p \times q$ to get $2pq$.

$$2pq = 2(0.6)(0.4)$$

$$2pq = 0.48$$

Therefore:

The frequency of the dominant alleles: $p = 0.6$

The frequency of the recessive alleles: $q = 0.4$

The frequency of individuals with the dominant genotype: $pp^2 = 0.36$

The frequency of individuals with the heterozygous genotype: $2pq = 0.48$

The frequency of individuals with the recessive genotype: $qq^2 = 0.16$

Remember: Frequencies can be checked by substituting the values above back into the Hardy-Weinberg equations. $0.6 + 0.4 = 1$ $0.36 + 0.48 + 0.16 = 1$

Step 6: Multiply the frequency of individuals (p^2 , $2pq$, qq^2) by the total population to get the number of individuals with that given genotype.

$p^2 \times \text{total population} = 0.36 \times 1,000 = 360$ black cats, BB genotype.

$2pq \times \text{total population} = 0.48 \times 1,000 = 480$ black cats, Bb genotype.

Effect of Random Mating:

Random mating results in maintaining the equilibrium of gene frequency in a population. For example, if the frequency of allele A is P and that of 'a' is q. If we make a cross between AA and aa, it will produce Aa. If individuals with Aa genotype are allowed to mate randomly, the gene frequency of three genotypes will be in the ratio of $P^2AA + 2PqAa + q^2aa$ (Fig. 30.1).

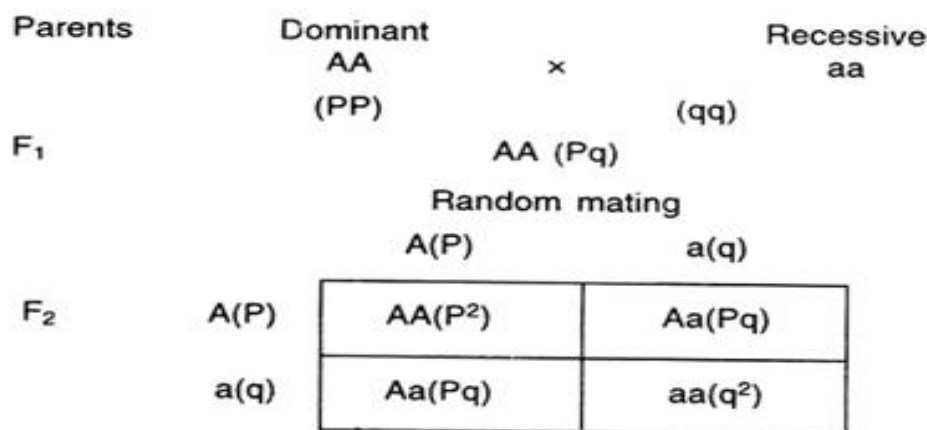


Fig. 30.1. Effect of Random Mating on gene frequency.

When gene frequencies are in equilibrium, it indicates absence of mutation, selection, migration and genetic drift in a population.

Factors Affecting Gene Frequency:

Hardy-Weinberg principle is based on three main assumptions, viz:

- (1) Random mating,
- (2) Equal survival of all genotypes, and
- (3) Absence of evolutionary forces like selection, mutation, migration and random genetic drift. Non fulfillment of these assumptions will lead to alteration in gene and genotype frequencies in a population.

However, the last assumption is seldom fulfilled. Mutation, migration and genetic drift change gene frequencies in a population. These factors are also known as forces of evolution because they play a key role in natural evolution.

These are briefly discussed below:

Selection:

Selection refers to a process which favours the survival and reproduction of some individuals in a population. The process of evolution in nature in which the fittest individuals survive and rest are wiped out is known as natural selection. Natural selection favours those characters which are advantageous for survival.

The selection by human efforts is known as artificial selection. Such selection favours those plant characters which are useful for mankind like productivity. Before discussing the effect of various types of selection, it is necessary to give brief account of fitness and selection coefficient.

Fitness:

The relative reproductive success of different genotypes of a population in the same environment under natural selection is known as fitness or selective value or adaptive value or selective advantage. It is denoted by W .