

# UNIT 20 BEHAVIOUR OF GENES IN POPULATIONS

Structure	Page No.
20.1 Introduction	23
Objectives	
20.2 Hardy-Weinberg Law	24
20.2.1 Determination of Allelic and Genotypic Frequencies	
20.2.2 Verification of Hardy-Weinberg Law	
20.3 Influence of other Evolutionary Forces on Gene Frequencies	27
203.1 Mutation and Change in Gene Frequencies	
203.2 Selection and Change in Gene Frequencies	
203.3 Selection against a Recessive Trait	
203.4 Genetic Drift	
203.5 Migration	
20.4 Summary	37
20.5 Terminal Questions	37
20.6 Answers	38

## 20.1 INTRODUCTION

In the previous units of this course, you have studied about two major subdisciplines of science of Genetics—the transmission and **molecular** genetics. You are now aware that transmission genetics deals with the genetic processes that occur among individuals and the unit of study in transmission genetics is the individual. Essentially the study is concerned with the transmission of genetically controlled characters from individual; of one generation to another. Molecular genetics, the other subdiscipline deals with the chemical nature of hereditary material. Also molecular genetics explains the mechanisms of encoding of genetic information in DNA molecule as well as the transformation of genetic information into relevant phenotype within the cell. Naturally the unit of study of molecular genetics is the cell.

In this unit, we are to discuss about the third subdiscipline of genetics namely population genetics. The study deals with the genetics of groups of **individuals** or populations and how the genetically controlled variations evolve into adaptations. The unit of study is, therefore, a Mendelian population which **means** a group of interbreeding individuals who share a common gene pool.

The central **theorem** of population genetics was independently conceived by Godfrey H. Hardy, a British Mathematician and **Wilhelm** Weinberg, a German Physician. In this unit, we shall state and discuss Hardy-Weinberg law and that the behaviour of genes in a population does not generally **conform** to the specification of the law. In other words, behaviour of genes in populations is concerned with changes in the frequencies of genes in the population. Such changes in **the** frequencies of genes form the genetic basis of evolution. For this reason, population genetics is also referred to as evolutionary genetics.

### Objectives

After studying this unit, you should be able to:

- state the Hardy-Weinberg law and justify the relationship between **allelic** and genotypic frequencies by applying the binomial equation.

**Gene pool :** The genes shared by a group of interbreeding individuals or a Mendelian population.

- show that both allelic and genotypic frequencies are maintained constant in the **absence** of outside evolutionary forces such as mutation, selection, genetic drift and gene flow  
demonstrate that only if mutations were alone to **occur** in a population, allelic frequencies would change over a period of time and conclude that with both forward and reverse mutations occurring, an equilibrium in gene frequencies is reached over a period of time.
- define the terms adaptive value and selection coefficient and explain that natural selection can either reduce or maintain genetic variations depending on the extent of adaptation of a trait to the **environment**
- describe that genetic drift results in random changes in the frequency of genes in small populations and that it may also result in loss of genetic variation.
- discuss that the gene flow can either introduce new alleles into a population or modify the frequency of existing alleles.

---

## 20.2 HARDY WEINBERG LAW

---

Hardy-Weinberg law states that in a population, the frequencies of a pair of alleles remain constant generation after generation provided that

- i) the population is infinitely large
- ii) the population is randomly mating one
- iii) there is no mutation, genetic recombination, gene migration and selection occurring in the population (in other words, the population is free from external evolutionary forces acting on it).

The conditions (i), (ii) and (iii) listed above have to be satisfied to a large extent if the gene frequencies are to be maintained in an equilibrium in a population for several generations.

The first condition is that populations must be infinitely large. If populations were to be small then the gene frequencies tend to drift every generation due to chance deviation or sampling error (refer to Unit 13 of LSE-07 course). The word 'drift' would refer to large and sudden changes in gene frequencies in a population. You may say that genetic drift varies directly with the size of the **population**. That is smaller the population, greater will be the genetic drift. We also mention here although in small populations drift is quite often common, the populations need not be "infinitely large" for Hardy-Weinberg law to be true.

The second condition requires that matings among the individuals of the population must be random. By random mating it is meant that any genotype should be able to mate with another genotype and there should not be any restriction in mating between genotypes. Also it means that matings occur between genotypes in proportion to the frequencies of **genotypes** in population. Stated differently, random mating means that the probability of mating between two genotypes is equal to the product of the two genotype frequencies. For instance, let us say that there are three genotypes AA, Aa and aa in a population occurring in a frequency of **0.36, 0.48** and **0.16** respectively. This means that assuming there are 1000 individuals in a population, 360 of them are AA, 480 of them are Aa and 160 of them are aa. The probability of mating occurring between genotypes AA and Aa should then be  $0.36 \times 0.48 = 0.1728$  and that of AA and aa should be equal to  $0.36 \times 0.16 = 0.0576$ . You could observe from these examples that the probabilities of other possible matings are equal to the products of the **genotypic** frequencies when mating is random.

Hardy-Weinberg law also requires that populations must be free from outside evolutionary forces. Hardy-Weinberg law aims at defining the role of heredity alone

The term frequency used in this unit refers to the number of times an allele or a genotype occurs in a population.

in changing the gene frequencies and the influence of reproduction on genotypic frequencies. Therefore, while applying Hardy-Weinberg law, we assume that there is no influence of other evolutionary forces such as mutation, natural selection, genetic recombination and gene migration. Later in this unit we shall discuss the role of other evolutionary forces in bringing about changes in gene frequencies. Before we proceed further attempt the following SAQ.

### SAQ 1

Fill in the blanks with suitable words.

- i) For Hardy-Weinberg equilibrium to be true, the population has to be .....
- ii) .....and ..... are certain factors which influence gene frequencies in a population.
- iii) A group of interbreeding individuals who share a common gene pool is called .....
- iv) Hardy-Weinberg law defines role of ..... in changing gene frequencies and the influence of ..... on genotypic frequencies.
- v) Assuming the genotype BB has a frequency of 0.35 in a population, that bb is 0.25 and Bb is 0.4, the probability of the possible mating occurring between , genotypes Bb and bb is equal to .....

### 20.2.1 Determination of Allelic and Genotypic Frequencies

In the previous section we stated the Hardy-Weinberg law. We shall verify the law based on certain arithmetic calculations. Prior to that we shall learn to determine the frequencies of alleles and genotypes in a population. One can represent Hardy-Weinberg formulation by the simple algebraic expression  $p^2 + 2pq + q^2$ , which is essentially the expansion of the binomial expression  $(p + q)^2$ . The two expressions can be effectively used to determine the frequencies of alleles of an ideal population. By ideal population we mean that it satisfies all the conditions required to maintain the frequencies of alleles at equilibrium generation **after** generation. To illustrate this idea let us consider a pair of alleles A and a, A being dominant to a. When the genotype AA mates with genotype aa, a heterozygous population of Aa is initially formed. Assuming the population is randomly mating, it will have three genotype classes AA, Aa and aa. Let us assume that the population consists of 1000 individuals 360 belonging to genotype AA, 480 to Aa and the remaining 160 to aa. Let us assume that frequency of A is p and a is q. Therefore, the frequency of genotypes AA, Aa and aa in the population is

$$\text{frequency of AA} = p^2 = 360/1000 = 36\% = 0.36$$

$$\text{frequency of Aa} = 2pq = 480/1000 = 48\% = 0.48$$

$$\text{frequency of aa} = q^2 = 160/1000 = 16\% = 0.16$$

Based on the above data it is possible to obtain the frequency of alleles A and a in the population.

$$\text{frequency of a} = q = \sqrt{\text{frequency of aa}} = \sqrt{0.16} = 0.4$$

$$\text{and the frequency of A} = p = (1 - q) = 1 - 0.4 = 0.6$$

$$\text{frequency of A} = 0.6$$

$$\text{frequency of a} = 0.4$$

$$\begin{aligned}
 \text{frequency of A} &= p = 0.6 \\
 \text{frequency of a} &= q = 0.4 \\
 (p + q) &= 1 \\
 (p + q)^2 &= (0.6 + 0.4)^2 = 1 \\
 p^2 + 2pq + q^2 &= (0.6)^2 + 2(0.6)(0.4) + (0.4)^2 \text{ where} \\
 p^2 &= \text{frequency of AA,} \\
 2pq &= \text{frequency of Aa and} \\
 q^2 &= \text{frequency of aa} \\
 &= 0.36 + 0.48 + 0.16 = 1 \\
 &\quad \text{AA} \quad \text{Aa} \quad \text{aa}
 \end{aligned}$$

You may observe here that the total allelic frequency ( $p + q$ ) = 1 and the total genotypic frequency,  $(p + q)^2$ , is also equal to 1.

### 20.2.2 Verification of Hardy-Weinberg Law

In the last subsection, you have learnt to calculate the frequencies of alleles and genotypes, when the number of individuals under each category of genotypes is known. Let us use these frequencies to verify the Hardy-Weinberg law. The law requires that the population must be randomly mating and that there are no outside evolutionary forces acting on the population. The three genotypes (AA, Aa and aa) when they are randomly mating can form nine mating types.

AA × AA  
 aa × aa

We earlier said that the probability of each mating occurring is the product of the frequencies of mating genotypes.

Thus

$$\begin{aligned}
 \text{AA} \times \text{AA} &= 0.36 \times 0.36 = 0.1296 \\
 \text{AA} \times \text{Aa} &= 0.36 \times 0.48 = 0.1728 \\
 \text{AA} \times \text{aa} &= 0.36 \times 0.16 = 0.0576 \\
 \text{Aa} \times \text{AA} &= 0.48 \times 0.36 = 0.1728 \\
 \text{Aa} \times \text{Aa} &= 0.48 \times 0.48 = 0.2304 \\
 \text{Aa} \times \text{aa} &= 0.48 \times 0.16 = 0.0768 \\
 \text{aa} \times \text{AA} &= 0.16 \times 0.36 = 0.0576 \\
 \text{aa} \times \text{Aa} &= 0.16 \times 0.48 = 0.0768 \\
 \text{aa} \times \text{aa} &= 0.16 \times 0.16 = 0.0256 \\
 &\quad \underline{\underline{1.0000}}
 \end{aligned}$$

Let us calculate the frequencies of the genotypes of the offspring of the above matings.

Parents	Ratio of offspring	Frequencies of genotypes of offspring			Total frequency	
		AA	Aa	aa		
AA × AA	1:4:4	0.1296			0.1296	
AA × Aa	1AA : 1Aa	0.0864	0.0864		0.1728	
AA × aa	1Aa		0.0576		0.0576	
Aa × AA	1AA : 1Aa	0.0864	0.0864		0.1728	
Aa × Aa	1Aa	2Aa : 1aa	0.0576	0.1152	0.0576	0.2304
Aa × aa	1Aa : 1aa		0.0384	0.0384	0.0768	
aa × AA	1Aa		0.0576		0.0576	
aa × Aa	1Aa : 1aa		0.0384	0.0384	0.0768	
aa × aa	1aa			0.0256	0.0256	
		0.3600	0.4800	0.1600	1.0000	

Therefore, at the end of the first generation the frequencies of genotypes have once again remained constant when all the conditions of Hardy-Weinberg law are met. It is needless to point out here that the frequencies of alleles A and a will also be 0.6 and 0.4 respectively, since there is no change in genotypic frequencies.

In the next section we shall start analysing one by one the role of outside evolutionary forces such as mutation and selection in influencing the Hardy-Weinberg law. Prior to that attempt the following SAQ.

### SAQ 2

- a) The frequency of the allele D in a population is 0.55. What is the frequency of the recessive allele in the same population?
- .....
- b) Two allele C and c have their frequencies as 0.7 and 0.3. What are the frequencies of the possible genotypes that these alleles could form?
- .....

## 20.3 INFLUENCE OF OTHER EVOLUTIONARY FORCES ON GENE FREQUENCIES

In the previous section we stated the Hardy-Weinberg law and showed that the allelic and genotypic frequencies remained the same after a generation when there was random mating in the population and the other evolutionary forces are **absent**. You have also learnt that these evolutionary forces include mutation and selection. In this section we shall look into the influence of such evolutionary forces on allelic and genotypic frequencies. Besides the influence of evolutionary forces, it is true that populations are usually small and the matings among the individuals are non-random, all of which once again contribute to change in gene frequencies. The net effect of the changes in gene frequencies over a number of generations in a population is the evolution of gene pool. The evolution is essentially due to the interplay of different evolutionary factors. We mentioned mutation and selection earlier. Genetic drift and gene migration are two other factors.

### 20.3.1 Mutation and Change in Gene Frequencies

Mutations are heritable changes in the genetic material occurring in a locus (refer to Unit 16 of LSE-03). Mutations are the raw materials for evolution. Mutations often convert one allelic form into another leading to a change in the phenotypic expression. The rate at which mutations occur is usually small. It varies between

$10^{-4}$  to  $10^{-6}$  in eukaryotes and  $10^{-7}$  or  $10^{-9}$  in prokaryotes. **This** means that in eukaryotes, it is only one out of 10,000 to 1,000,000 loci **undergoes** mutation. Mutation rate in effect **determines** the rate at which the gene frequencies change. If the mutations are lethal the rate of change in the **frequencies** of alleles is high as persons carrying lethal mutations do not survive.

We earlier said that mutations are the sources of all genetic variability. First mutations cause genetic variations and **then in response** to various evolutionary forces, the different alleles increase or decrease in frequency.

Many mutations are detrimental and therefore eliminated from the population. Some mutations confer adaptations on individuals who possess them and therefore are likely to spread in the **population**. Whether a mutation confers any advantage or disadvantage on individuals who possess them depends on the environment in which these individuals live. A mutation which is advantageous in one particular environment may prove to be otherwise in a changed **environment** (refer to units 11 of LSE-07 for a detailed discussion on mutations and evolution).

The mutation of a wild type allele to its mutant **form** is referred to as forward mutation. For instance mutation of A to a. If mutant locus undergoes mutation to its wild type, then it is referred to as reverse mutation. Reverse mutation rates are smaller than forward mutation rates. Now, let us look into the mechanism by which mutations alter the gene frequencies.

Let us consider a hypothetical population in which the allele A has the frequency of  $p$  and allele a has the frequency of  $q$ . Let us also say that A mutates to a (forward mutation) at a rate of  $u$  and a mutates to A (reverse mutation) at a rate of  $v$ . We assume that the population is large and there is no selection operating on the population. The change in frequency of A depends on the mutation rate  $u$  and on the initial frequency of A,  $p$ . For instance, assume that there are 100,000 alleles in a population and the mutation rate is  $10^{-4}$ . If all the alleles in the population are A and  $p = 1.0$ , the number of alleles that would undergo mutation is  $100,000 \times 10^{-4} = 10$ . Contrarily if there are only 10,000 alleles then the number of mutant alleles in a generation would be  $10,000 \times 10^{-4} = 1$ . The decrease in the frequency of A as a result of mutation to a is  $up$ . Similarly the increase in frequency of a as a result of mutation from A is  $vq$ . The change in **frequency** =  $vq - up$ . As we **pointed** out earlier, **when** the frequency of A is high, the **number** of alleles undergoing mutation is also **high**. As the frequency of A decreases, the number of alleles undergoing mutation also decreases. Initially  $q$  will be small, but as more and more A alleles mutate to a,  $q$  increases and the number of alleles undergoing reverse mutation also increases. Essentially this would result in an equilibrium, a point at which the number of alleles undergoing forward mutation becomes equal to the number of alleles undergoing reverse mutation. Also, at this point there will be no further change in gene frequency although mutations continue to occur in both the directions. **As** the equilibrium is reached, the change in the frequency of A, will be equal to zero.

$$\Delta p = vq - up = 0$$

$$\text{i.e., } vq = up.$$

$$\text{Since } p = (1 - q)$$

$$vq = u(1 - q)$$

$$vq = u - uq$$

$$vq + uq = u$$

$$q(v + u) = u$$

$$\hat{q} = \frac{u}{(v + u)}$$

$q$  is known as equilibrium value of  $q$ . Similarly the equilibrium value of  $p$  i.e.,  $p$  can be obtained and this will be  $\hat{p} = \frac{v}{v+u}$ .

Now, we shall try to explain the above expressions in numerical terms. Let us say that in a population frequency of  $A = p = 0.6$  and that of  $a = q = 0.4$ . Allele  $A$  mutates to  $a$  at a rate of  $u = 4 \times 10^{-5}$  and  $a$  undergoes reverse mutation at a rate of  $v = 2 \times 10^{-5}$ . The change in gene frequency in the first generation is

$$\begin{aligned} \Delta p &= vq - up \\ &= (2 \times 10^{-5} \times 0.4) - (4 \times 10^{-5} \times 0.6) \\ &= -0.000016. \end{aligned}$$

The frequency of  $A$  in the 211d generation is  $0.6 - 0.00016 = 0.59984$  and that of  $a = (1 - p) = 0.40016$ . At equilibrium, the frequency of allele  $a$ ,  $q$  is equal to

$$q = \frac{u}{u+v} = \frac{(4 \times 10^{-5})}{(4 \times 10^{-5}) + (2 \times 10^{-5})} = 0.667.$$

You should remember that mutation rates are ordinarily very low and for the change in gene frequency to occur only due to the mutation pressure, it must take several generations. For instance if frequency of  $A$  is 1.00 then it might require 1000 generations to change the frequency to 0.99 at a mutation rate of  $1 \times 10^{-5}$ . It might take 2000 generations to change it from 0.5 to 0.49 and roughly 10,000 generations to change it from 0.1 to 0.09. Such calculations hold good only if there are no reverse mutations. But with reverse mutations occurring, the rate may be even slower.

To summarise it could be said that gene frequencies change over a period of time if mutations occur in the population and other evolutionary forces are not active. Further, with both forward and reverse mutations occurring, the gene frequencies tend to reach an equilibrium over a period of time.

### SAQ 3

Match the following:

- |                                  |                                   |
|----------------------------------|-----------------------------------|
| a) $p =$                         | i) $\left( \frac{u}{u+v} \right)$ |
| b) $\hat{q} =$                   | ii) zero                          |
| c) $\hat{p} =$                   | iii) $(1 - q)$                    |
| d) At equilibrium<br>$vq - up =$ | iv) one                           |
| e) $(p + q) =$                   | v) $\left( \frac{v}{v+u} \right)$ |

### 20.3.2 Selection and Change in Gene Frequencies

The theory of natural selection propounded by Charles Darwin explains the process of evolutionary change. (For a detailed discussion on natural selection refer to units 11 and 12 of LSE-07.) Natural selection is a process which has a direction and order. It aims to promote adaptations in populations. Adaptations refer to the phenotypic traits or expressions that make the organisms more suited to their immediate environment. Adaptations besides contributing to the increased survival of the organisms, also increase their reproductive efficiency. Adaptations arise from among the variations in the traits of the organisms. Variations are universal in nature. No single trait is fixed. All traits exhibit a measure of variability. Such of those variations which are useful to the organism in terms of its survival and

reproduction in a given environment are deemed as adaptations. The green colour of the insect living on a tree that helps it to escape predation by enemies is a clear case of adaptation. Other insects may possess slightly different colours but may **not** be able to **camouflage** themselves among green leaves of the tree and therefore easily picked up predators such as birds. Insects which are well adapted to live in such an environment have better chances of survival and leave behind more number of offspring. In fact, natural selection is related to the notion of differential reproduction. Whereas survival *per se* is important, it is the reproduction that ensures that one's genes are passed on to the succeeding generation. What is more important is not the survival of individuals but the genes which they leave behind, and for this reproduction must occur. Therefore, natural selection is quantified by assessing reproduction. **The** relative reproductive **efficiency** of genotypes is **termed** Darwinian fitness or adaptive value.

Fitness is usually symbolised as  $w$ . Since it is a relative value, it is assigned the value of 1 to that genotype that produces maximum number of offspring. For instance, let us say that **two** alleles A and a form genotypes AA, Aa and aa. AA produces four offspring, Aa produces six offspring and aa two offspring. Here Aa produces maximum number of offspring and therefore, is assigned the fitness value of 1. The fitness value of other genotypes is calculated relative to the genotype with the fitness value of 1, in this instance, Aa. Therefore, AA has a fitness value ( $w$ ) of  $4/6 = 0.67$  and that of aa is  $2/6 = 0.33$ .

Fitness value is a measure of reproductive efficiency of an organism in terms of natural selection. The converse, namely the measure of relative intensity of selection against a genotype is called selection **coefficient**. Selection coefficient symbolised as  $s = 1 - w$ . In the example cited above, the genotype AA will have a selection coefficient of  $1 - 0.67 = 0.33$ ; Aa has a  $s$  value of  $1 - 1 = 0$  and that of a is  $1 - 0.33 = 0.67$ .

Does natural selection act to change the allelic frequencies in a population? The answer appears to be yes. Depending on **the** type of environment in which it acts, selection may prefer to maintain a constant frequency of an allele by eliminating variations in a population. Or it may endeavour to change the frequency by maintaining variations. The relative fitness of the individuals in a population and the initial gene frequencies influence the action of natural selection.

In Table 20.1 we have computed fitness value and selection coefficient for three genotypes we have chosen as example. Table 20.2 explains the influence of selection on gene frequencies.

**Table 20.1 Calculation of fitness value ( $w$ ) and selection coefficient ( $s$ ) of three genotypes**

Genotypes	AA	Aa	aa
No. of breeding adults in generation 1	12	20	10
No. of offspring produced by all adults in generation 2	48	120	20
Average number of offspring produced per adult	$48/12 = 4$	$120/20 = 6$	$20/10 = 2$
Fitness value ( $w$ ) (relative number of offspring produced)	$4/6 = 0.67$	$6/6 = 1$	$2/6 = 0.33$
Selection coefficient ( $s$ )	$1 - 0.67 = 0.33$	$1 - 1 = 0$	$1 - 0.33 = 0.67$

Table 20.1 shows the method of **arriving** at the fitness value and selection coefficient, when the number of individuals belonging to various genotypes and number of offspring they produce are **known**. From Table 20.2 you could learn how the gene frequencies are altered under the influence of natural selection.

**Table 20.2 Influence of Natural Selection on the Changes in Gene Frequencies In a Population**

Genotypes	AA	Aa	aa
Initial genotype frequency	$p^2$	$2pq$	$q^2$
Fitness value (w)	$w_{11}$	$w_{12}$	$w_{22}$
Frequency of genotypes after selection	$p^2 w_{11}$	$2pq w_{12}$	$q^2 w_{22}$
Relative genotype frequency after selection	$P' = \frac{p^2 w_{11}}{\bar{w}}$	$H' = \frac{2pq w_{12}}{\bar{w}}$	$Q' = \frac{q^2 w_{22}}{\bar{w}}$
(where $\bar{w}$ = mean fitness value of population = $p^2 w_{11} + 2pq w_{12} + q^2 w_{22}$ )			

Gene frequency after selection:

frequency of A =  $p' = P' + \frac{1}{2}(H')$

frequency of a =  $q' = 1 - p'$

Change in gene frequency of A due to selection =

$\Delta p = p' - p$

Changes in gene frequency of a due to selection =

$\Delta q = q' - q$

In Table 20.2, the frequency of different genotypes after selection is calculated by multiplying the initial frequency (frequency before selection) with the fitness value of each genotype ( $w_{11}$  for AA,  $w_{12}$  for Aa and  $w_{22}$  for aa). The sum of the genotypic frequencies after selection =  $\bar{w}$  and is known as mean fitness value of the population. The frequency of each genotype after selection is divided by the mean fitness value to obtain the relative frequency of each genotype. From these values the allelic frequencies can be obtained by using the following formula.

Frequency of allele A after selection =  $p' =$  frequency of AA after selection +  $\frac{1}{2}$  ( frequency of Aa after selection ) and  $q' = (1 - p')$ .

Let us substitute the numerical values for the above expressions as shown in Table 20.3. Let us consider two alleles A, and a with initial gene frequencies of 0.6 and 0.4 respectively.

**Table 20.3**

Genotypes	AA	Aa	aa
Initial genotype frequencies	$p^2$ $0.6 \times 0.6$ $= 0.36$	$2pq$ $2 \times 0.6 \times 0.4$ $= 0.48$	$q^2$ $0.4 \times 0.4$ $= 0.16$
Fitness value (w)	$w_{11}$ $= 0.67$	$w_{12}$ $= 1$	$w_{22}$ $= 0.33$
Frequency after selection	$p^2 w_{11} =$ $0.36 \times 0.67$ $= 0.240$	$2pq w_{12} =$ $0.48 \times 1$ $= 0.48$	$q^2 w_{22} =$ $0.16 \times 0.33$ $= 0.053$
Mean fitness value of the population $\bar{w} =$	$p^2 w_{11} + 2pq w_{12} + q^2 w_{22}$ $0.24 + 0.48 + 0.053$ $= 0.773$		
Relative genotype frequency after selection	$P' = \frac{p^2 w_{11}}{\bar{w}}$ $H' = \frac{2pq w_{12}}{\bar{w}}$ $Q' = \frac{q^2 w_{22}}{\bar{w}}$  $= \frac{0.24}{0.773}$ $\frac{0.48}{0.773}$ $\frac{0.053}{0.773}$ $0.31$ $0.62$ $0.07$		

Frequency of A after selection =  $p' = P' + \frac{1}{2}(H')$

$= 0.31 + \frac{1}{2} \times 0.62 = 0.62$

And the frequency of a after selection =  $1 - p' = 1 - 0.62 = 0.38$

Change in frequency due to selection for allele A

$$\Delta p = p' - p = 0.62 - 0.6 = 0.02$$

$$\text{and for allele a} = \Delta q = q' - q = 0.38 - 0.4 = -0.02$$

In other words, after selection process, the frequency of A has increased from 0.6 to **0.62** and that of a has decreased from 0.4 to **0.38**.

### 20.3.3 Selection against a Recessive Trait

Many new traits arise as recessive mutations and are subject to severe selection pressure. Obviously such recessive mutants have a reduced fitness value. Under such circumstances both the dominant homozygote and the heterozygote have a fitness value of 1. This means that there is no selection pressure on these genotypes. The fitness value of recessive genotype would be  $(1 - s)$  where  $s$  is the **selection coefficient**. Let us tabulate the frequency and the fitness values of these genotypes.

Genotype	Frequency	Fitness Value
AA	$p^2$	1
Aa	$2pq$	1
aa	$q^2$	$1 - s$

Assuming the frequency of genotypes are in Hardy-Weinberg proportions, the contribution of each of genotype to the next generation will be the product of their frequency and fitness value. Thus, the frequency of genotypes in the next generation will be

$$\text{AA} \quad p^2 \times 1 = p^2$$

$$\text{Aa} \quad 2pq \times 1 = 2pq$$

$$\text{aa} \quad q^2 \times (1 - s) = (q^2 - sq^2)$$

The mean fitness value of the population would then be

$$p^2 + 2pq + q^2 - sq^2$$

$$\text{Since } p^2 + 2pq + q^2 = 1,$$

$$\text{the mean fitness value} = (1 - sq^2).$$

The relative genotypic frequencies after selection will be

$$\text{AA} = \frac{p^2}{(1 - sq^2)}$$

$$\text{Aa} = \frac{2pq}{(1 - sq^2)}$$

$$\text{aa} = \frac{q^2 - sq^2}{1 - sq^2}$$

In order to obtain the frequency of allele a after selection, that is  $q'$  value, we have to add the frequency of homozygote (aa) and half of the frequency of heterozygote (Aa)

$$q' = \frac{q^2 - sq^2}{1 - sq^2} + \frac{1}{2} \times \frac{2pq}{1 - sq^2}$$

$$\begin{aligned}
 &= \frac{q' - sq'}{(1 - sq^2)} + \frac{pq}{(1 - sq^2)} \\
 &= \frac{q' - sq' + pq}{1 - sq^2} \\
 q' &= \frac{q' + pq - sq^2}{1 - sq^2} \\
 &= \frac{q(q + p) - sq^2}{1 - sq^2}
 \end{aligned}$$

$$\text{Since } (q + p) = 1$$

$$q' = \frac{q - sq^2}{1 - sq^2}$$

and

$$p' = (1 - q')$$

The above expression helps you to determine the frequencies of a pair of alleles when there is a selection pressure on the recessive genotype and the **homozygous** dominant and heterozygous genotypes have a fitness value of 1. At this point we end our discussion on the role of selection in changing the gene frequencies. Before we proceed to discuss the role of genetic drift **and** gene migration in changing the gene frequencies in populations, attempt the following SAQ.

#### SAQ 4

- i) Define the terms adaptive value and selection coefficient.

.....  
 .....  
 .....

- ii) If allele has an adaptive value of 0.55, what is its selection coefficient?

.....

- iii) Genotype AA has a value of  $w = 0.5$  and its frequency in the population is 0.6, genotype Aa has a  $w = 1.0$  and has an initial frequency of 0.3. Genotype aa has  $w = 0.2$  and has an initial frequency of 0.2. What is the **mean** adaptive value of the population ( $\bar{w}$ )?

.....

#### 20.3.4 Genetic Drift

For gene frequencies to remain **constant** generation after generation, Hardy-Weinberg law requires that population to **be** infinitely large. But usually natural populations are not infinitely large. At the same time, they are large enough that the gene frequencies do not change drastically. But there are several small populations in which there are random changes in gene frequencies. Such random changes in gene frequencies due to chance event are called **genetic drift**. The phenomenon was first brought to light by an American population geneticist **Sewall** Wright, hence come to be known as **Sewall** Wright effect.

Random changes in gene frequencies or genetic drift is an important evolutionary force in small populations. To illustrate genetic drift in small populations, let us

look into the following example. Imagine a small population of mice living in a barn of a farmer as four or five extended families. The farmer tries a variety of ways to get rid of the mice. He may try to set traps, use a shot gun, make surprise visits, employ cats and other means as well. Such acts of the farmer exert a severe selection pressure. Under such **circumstances** the traits that would be selected are the swiftness, short tails, hearing acuity and cautiousness. In other words, the frequencies of genes for such traits would be very high. Contrarily the gene for thick fur on the body will have a low frequency as thick fur is not the trait that will be chosen in the present environment. Assuming hearing acuity has a frequency of 0.9 in the population, thick fur may have a frequency of 0.2. Now, let us imagine a sudden environmental change. A severe winter sets in. The farmer confines himself to the fire place. Such of those traits which had a very **high** frequency when the farmer was hunting the mice will lose their significance in the altered environment. So, the next generation of mice with thick fur alone survive and produce offspring with thick fur. The frequency of the gene for thick fur increases in population and that of hearing acuity decreases. In the next generation the respective frequencies of genes for hearing acuity and fur thickness could be respectively 0.3 and 0.8. Since the population is small, severe winter eliminates from the environment mice with less fur in the body. Assuming the population consisted of **1000** individuals, it is unlikely that most individuals with less thick fur would be eliminated from the population by chance alone and the frequency for hearing acuity would not decrease randomly from 0.9 to 0.3.

The change in environment causing a heavy mortality in natural populations is one of the several ways in which genetic drift occurs. Such a drift is essentially caused by sampling error. Sampling error would refer to chance deviations from expected **ratios**. For example, in an infinitely large population, infinitely large number of gametes are produced. The frequency of a pair of alleles in such a population, let us say, is  **$p$  and  $q$**  and the frequency of genotypes they form are  $p^2$ ,  $2pq$  and  $q^2$ . Such frequencies would remain same in the next generation as well. But, from such a large population, if you take a sample, then the number of progeny of this sample is limited. Just by chance or error, the sample may deviate from the large gene pool. The potential for deviation is much larger in **smaller** and smaller samples.

All genetic drift is due to sampling error. And the sampling error may occur in a number of **ways**. They are:

- **Genetic drift** arises **when, the** population continuously **remains** small over many **generations**.
- Genetic **drift may** arise through founder effect. By founder effect it **is meant** that population initially consists of a small number of individuals but may subsequently grow in size and later may consist of large number of individuals. But the gene pool of the population is derived from the genes of individuals who originally founded the population. The founder population may not be the representative of the parent population from which it is formed.

An often cited example for **founder** effect in natural population is that of Dunker families living in United States. **Dunkers** are a religious sect who migrated from Germany to Pennsylvania in USA between 1719 and 1729. Fifty Dunker families emigrated, settled in **USA** and remained as an **isolated** group and formed a small population. They very rarely married outside their communities. During **1950s**, geneticists studied the Dunker population consisting of 300 individuals. Studies showed that frequencies of some of **genes** of **Dunker population** were very different from the frequencies of genes of population of US where Dunkers have settled as well as from the frequencies of genes of population in Germany from where they migrated. Table 24.4 shows study of the frequencies of alleles ABO blood group system in Dunker population, American population and German **population**.

**Table 20.4** Comparison of frequencies of blood group alleles and phenotypes of American, German and Dunker populations.

Population	Allelic frequencies			Phenotypic frequencies			
	I <sup>A</sup>	I <sup>B</sup>	I <sup>O</sup>	A	B	AB	O
Dunker	0.38	0.03	0.59	0.593	0.036	0.023	0.348
USA	0.26	0.04	0.70	0.431	0.058	0.621	0.490
W.Germany	0.29	0.07	0.64	0.455	0.095	0.041	0.410

You could observe from the Table 20.4 that the frequencies of alleles controlling blood group system of Dunkers are different from those of **Americans and Germans**.

You should remember that Dunkers have largely remained isolated and did not mix with the American population. Under such circumstances only the **phenomenon** of drift could explain for producing random changes in the gene pool of Dunker population. To begin with, it was founder effect as only 50 families migrated from Germany to get settled in **US** in 1729. From then on drift had influenced gene frequencies in each generation, since the population size had always remained small.

- The third form of sampling error is due to bottleneck effect. Bottleneck effect occurs when the population size is drastically reduced in one generation. During such reduction in the size of the population, some genes may be lost from the gene pool by chance. We earlier pointed out that reduction in population size may occur due to several reasons including natural calamities. Such reduction in population sizes cause random changes in frequencies of gene and the resultant drift.

Genetic drift and the random changes in gene frequencies in small populations cause much changes in the genetic structure of the populations. Such effects can be summarised as follows:

Drift causes changes in gene frequencies of a population over a **period** of time.

- Drift may also result in **the loss** of genetic divergence in populations. In some cases an allele may be completely lost when all the individuals possessing a particular allele in a small population are killed due to chance event.
- Drift also causes loss of genetic variation in population. With a loss of a particular allele from the population, the population may increasingly become homozygous. Decrease in **heterozygosity** results in **the** loss of genetic variation in the population.

We end our discussion on genetic drift here and proceed to discuss yet another factor that could bring out changes in gene frequencies, namely migration.

### 20.3.5 Migration

While describing Hardy-Weinberg law, we mentioned that for the law to hold true, the **population** should not be influenced by other evolutionary forces. The **population** should more or less remain a **closed** one. But most populations are not very isolated and exchange of genes with **other** populations do often occur. When new individuals enter into a population, they may either introduce a new allele into the population or alter the frequencies of the existing alleles **of the** population.

**Migration** of individuals which in effect **means** migration of genes into a **population** may alter the Hardy-Weinberg equilibrium and **can** cause the evolution of gene

frequencies in a population. To be a force in evolution, migration should ensure that migrating individuals are reproductively active contributing genes to the recipient population. In such an event, the migration can also be described as **gene flow**.

The fact that gene flow causes changes in the frequencies of allele in a population can be explained by a simple model.

Let us consider two populations, **population I** and **population II**. The frequency of allele A in population I is  $p_i$  and that of population II is  $p_{ii}$ . At each generation, let us say, some individuals randomly migrate from population I to II. At the end of migration population II will have two groups of individuals--the migrants and residents, with allelic frequencies of  $p_i$  and  $p_{ii}$  respectively. Let us designate the proportion of individuals who form migrants in population II as  $m$

Frequency of allele A in population I before migration	Frequency of allele A in population II after migration
$f(A) = p_i$	(Residents = $p_{ii}$ ; migrants = $mp_i$ ) $f(A) = mp_i + (1 - m)p_{ii}$

The change in the frequency of A due to migration =

$$\Delta p = p'_{ii} - p_{ii}$$

$$\Delta p = mp_i + (1 - m)p_{ii} - p_{ii}$$

$$\Delta p = mp_i + p_{ii} - mp_{ii} - p_{ii}$$

$$\Delta p = mp_i - mp_{ii}$$

$$= m(p_i - p_{ii})$$

As you could observe from the above equation the change in frequency due to gene flow depends on two factors. (i) The proportion of migrants in the final population ( $m$ ); (ii) the difference in the frequency between migrants and residents ( $mp_i - p_{ii}$ ).

Migration causes certain significant changes in a population. One, it might increase the net effective size of the populations. Two, it might reduce the **genetic drift** in small population as well as genetic divergence.

### SAQ 5

State whether the following statements are true or false.

- i) For **Hardy-Weinberg** equilibrium to hold good population sizes should generally be small. (True/false)
- ii) Change in gene frequencies due to chance events is called genetic drift. (True/false)
- iii) Chance deviations from expected ratios is referred to as sampling error. (True/false)
- iv) Founder effect and bottleneck effect refer to phenomena that are contrary to genetic drift. (True/false)
- v) Drift increases genetic variations in small populations. (True/false)
- vi) **Migration** of individuals in effect means the flow of genes into a population. (True/false)
- vii) Gene flow causes an increase in genetic divergence in a population. (True/false)

## 20.4 SUMMARY

---

- Hardy-Weinberg equilibrium forms the central theorem of population genetics.
  - The frequencies of a pair of alleles tends to remain constant generation after generation, if the population is infinitely large and randomly mating, and the outside evolutionary forces have no influence on the population.
  - Outside evolutionary forces such as mutation and selection significantly alter the gene frequencies over a number of generations.
  - Genetic drift caused by chance events tends to cause random and unpredictable changes in the frequencies of alleles.
  - Migration of individuals and the subsequent gene flow cause alterations in gene frequencies. Gene flow may minimise the effects of genetic drift in small populations as it increases the net effective size of the population and decreases the genetic divergence.
- 

## 20.5 TERMINAL QUESTIONS.

---

1. State the Hardy-Weinberg law.
2. ABO blood groups are controlled by a system of three alleles  $I^A$ ,  $I^B$ ,  $I^O$ . Assuming a population having these three alleles consists of genotypes which are randomly mating, what are the possible mating pairs?  
.....  
.....  
.....  
.....  
.....
3. Discuss briefly that mutations are an evolutionary force,  
.....  
.....  
.....  
.....  
.....
4. What is the role of selection in altering the gene frequencies in a population?  
.....  
.....  
.....  
.....
5. Define the term genetic drift. How does genetic drift affect gene frequencies in a population?  
.....  
.....  
.....  
.....

## SAQs

- Infinitely large
  - Mutation, natural selection, migration and genetic drift
  - Mendelian population**
  - heredity, reproduction
  - 0.1
- 0.45
  - 0.49, 0.42, 0.09**
- iii
  - i
  - v
  - ii
  - iv
- Adaptive value: Refers to the relative reproductive efficiency of a genotype.  
Selection coefficient: The amount of selection pressure on a genotype. It is related to adaptive value by the equation  $s = (1 - w)$
  - 0.45
  - 0.64
- False
  - True
  - True
  - False
  - False
  - True
  - True**

## Terminal Questions

- Hardy-Weinberg laws states that frequencies of a pair of alleles moved remain **constant** in a population generation after generation, provided that the population is infinitely large, randomly mating and that there are no outside evolutionary forces acting on the population.
- |                          |                          |                          |
|--------------------------|--------------------------|--------------------------|
| $I^A I^A \times I^A I^A$ | $I^A I^B \times I^B I^B$ | $I^A I^B \times I^O I^O$ |
| $I^A I^A \times I^A I^O$ | $I^A I^B \times I^A I^O$ | $I^O I^O \times I^A I^A$ |
| $I^B I^B \times I^B I^B$ | $I^A I^B \times I^B I^O$ | $I^O I^O \times I^A I^O$ |
| $I^B I^B \times I^B I^O$ | $I^A I^B \times I^A I^B$ | $I^O I^O \times I^B I^B$ |
| $I^A I^B \times I^A I^A$ | $I^A I^A \times I^A I^O$ | $I^O I^O \times I^B I^O$ |
|                          |                          | $I^O I^O \times I^O I^O$ |
- Mutations which cause heritable changes in the genetic material occurring in a locus are the raw materials for evolution. They are responsible for generating **variations** in the population. Such of those mutations which are advantageous to the organisms are retained in the population and their frequencies increase in the population. Accumulation of adaptations is an evolutionary process. Since one of the definitions of evolution is **that it is change in gene frequencies**, mutations being responsible for change in gene frequencies, contribute to the evolutionary process.
- Refer to Section 20.3.2
- Refer to Section 20.3.4