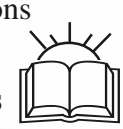
UNIT 3 POPULATION GENETICS

Contents

Learning Objectives



Once you have studied this unit, you should be able to:

- know the importance of population genetics;
- understand what Mendelian population is and clearly differentiate breeding population and effective population;
- understand the concepts of genetic polymorphism;
- understand the significance of Hardy-Weinberg Law;
- explain how mutation, migration, selection, inbreeding, genetic drift, consanguineous and non-consanguineous mating disturb the genetic equilibrium and also clearly understand the process of evolution of populations; and
- know what genetic load is, and understand the effect of consanguineous marriages.

3.1 INTRODUCTION

Genetics, a discipline of biology, is the study of fundamental units of inheritance called genes, heredity, and variation in living organisms. This hereditary material (gene), whether as a unit of segregation, recombination, mutation, or function, is the unifying idea basic to the field of genetics. In 1866, Gregor Mendel put forward the mechanisms for heredity and variation. The Mendelian laws: the independent segregation and recombination of dominant and recessive characters constitute the cornerstone of the modern science of genetics. Mendel's monumental work (1866) on the principles of inheritance, that is, Mendel's Laws of inheritance, remained long ignored, and only received attention in 1900, sixteen years after his death (1884). It was not until 1900, when three botanists, de Vries, Correns, and Von Tschermak independently rediscovered the Mendelian Principles. Later Mendel's experiments were extended to many species of plants and animals including man.

Human genetics is a subject of special interest to us as students of anthropology. Human genetics itself is further subdivided into the areas of medical genetics, biochemical genetics, cytogenetics, somatic cell genetics, immunogenetics, formal or mathematical genetics, population genetics, and anthropological genetics. These subdivisions of human genetics are closely interrelated and interdependent. For example, the study of the distribution and evolution of the abnormal haemoglobins in human populations witnessed the union of medical genetics, biochemical genetics, formal genetics, and population genetics.

Population Genetics

Study of a whole population is, in fact, often superior to the collection of large pedigree, because pedigree has unusual characteristics and is of specific interest, and thus is not representative of a population. Within many populations an equilibrium of genotypes prevails. This was first pointed out in 1908 independently by the mathematician G. H. Hardy and the physician W. Weinberg whose several contributions laid the foundations of the genetic study of natural populations of man and wild animals. The foundations by Sewall Wright, R. A. Fisher and J. B. S. Haldane helped the formation of modern population genetics. The mathematical theory of population genetics was developed in the early twentieth century due largely to the work of Sewall Wright, Ronald Fisher, and J. B. S. Haldane. So the population genetics deals with the consequences of Mendelian laws on the composition of the population with special reference to the effects of mutation, selection, migration, and chance fluctuation of gene frequencies.

The population considered under Hardy-Weinberg law is a unique population. It does not change genetically and cannot and does not evolve. It is an *ideal population*, because it necessarily fulfilled certain "*ideal*" conditions and is a mathematical abstraction, because no real population fulfills the ideal conditions such as large size of the population with equal sexes, random mating and equal fertility among all couples and another stipulation that the population must be free from evolutionary forces.

3.2 MENDELIAN POPULATION

A population *isolate* is that group of persons within which individuals choose their partners. Such an isolate is also called a *Mendelian population*. Ideally the population isolate inhabits an island, a mountain valley, a peninsular region, a forested area, or even a large area covering several villages, where the marriage alliance is restricted within that endogamous group.

The general and simple definition of *population* is the number of people in an area at a given time. It may be used in reference to the number of people possessing a particular character or group of characteristics in an area at a given time. It is difficult to define a particular population strictly, for the actual boundaries around a specific human population are not always easy to find. A human population is usually found in a particular place, and it is a coherent entity largely because of geographical boundaries. Regardless of how they are circumscribed, the significance that populations have for evolutionary genetics lies in the web of genetic relationships within and between them—allele frequencies, consanguinity,

mating patterns, gene flow, natural selection, etc. The genetic approach uses the concept of the *Mendelian population*, which Dobzhonsky has defined as "a reproductive community of sexual and cross fertilizing individuals which share in a common gene pool". If the isolate or the Mendelian population is not changed by natural selection, nor by mutation, nor by migration, and if the population size is large and if individuals are not mating assortatively (that is, random choice of partners), then the isolate is said to be in equilibrium. These assumptions are fundamental to population analysis and for maintaining an equilibrium of genotypes from generations to generations.

Although, all human gene pools are open to varying degree, it is evident that panmixis does not take place within the total species. The more important mechanisms maintaining genetic isolation of populations today are cultural rather than geographical.

Breeding Population

In addition to the problem introduced by the biological openness of human-population systems, accurate definition of a human Mendelian population is complicated by the fact that man clusters in social groupings which may or may not serve as biological breeding units. So the first problem of the population geneticist, therefore, is to identify and describe, as accurately as possible, the biological population before he can undertake an analysis of the gene pool and forces acting on it. Because direct analysis of a population's gene pool is impossible, all conclusions regarding its composition are necessarily inferential, and must be made on the basis of direct examination of the phenotypes of the reproducing individuals. To infer the composition of a gene pool at a single point in time the population geneticist must first enumerate and describe the actual progenitors, that is, the parents in a population. These progenitors constitute the breeding population.

3.3 GENETIC POLYMORPHISM

Genetic polymorphism is defined as the occurrence together in the same habitat at the same time of two or more distinct forms of a species in such proportions that the rarest of them cannot be maintained merely by recurrent mutation (Ford, 1940). Genetic polymorphism can also be defined as the occurrence in the same population of two or more alleles at one locus, each with appreciable frequency (Cavalli-Sforza and Bodmer, 1971). A formal definition, such as the above, based on the frequency of genes that are found in a population is likely to be the most satisfactory. At one time, polymorphism was defined in terms of the selective mechanisms responsible for maintaining relatively high gene frequencies of two or more alleles at a locus. Because it is difficult to determine these mechanisms or the nature of these forces and therefore can hardly be useful in a definition. This is the reason why it is difficult to accept unequivocally Ford's definition. However, it is likely that Ford's definition applies to many, if not most, instances of polymorphism (Ford, 1964).

Some knowledge of the theory of polymorphism is essential for a clear understanding of the blood groups and the kindred phenomena. It should be noted that the definition of polymorphism excludes the following forms of variation.

- ❖ Geographical races, White, Mongoloid and Negroid types of man. These are normally maintained by isolation from one another. It should be stressed here, that the occurrence of polymorphism in one district and its absence or different nature in another, may be an important attribute of distinct communities.
- ❖ 'Continuous variation' under multifactorial (or environmental) control, such as height, is brought about by cumulative effect of segregation taking place at many loci. This cannot be considered as polymorphic condition, as it is not maintained in the population by selection.
- ❖ The segregation of rare recessive, albinism for example or rare heterozygous conditions, such as Huntington's chorea, are eliminated by selection and maintained only by mutation. Hence they cannot give rise to polymorphism.

It must be noted that polymorphism cannot normally be maintained environmentally.

Genetic polymorphisms are very common phenomena in all human populations. Most of the polymorphisms encountered in human populations so far fall into two main categories: blood-cell antigens (blood groups) and blood proteins (serum proteins). The first category of polymorphisms, the kind detected by immunological techniques, is that of blood groups or blood cell antigens, of which the ABO blood groups are an outstanding example. The second category of polymorphisms, most of them detected by electrophoretic techniques, for which complications due to incompatibility are not known or are not likely to occur, comprises proteins found in the blood either in the free, liquid portion (serum or plasma) or in its cells (red or white). The modern techniques of biochemistry revealed how widely individual men and populations differ in the various enzymes and proteins systems of the body.

Polymorphism in Man

The human polymorphism may conveniently be introduced by two examples which reveal their essential qualities. Let us discuss about sickle-cell anaemia, which is genetically controlled by a gene which produces the disease when homozygous and is responsible merely for the sickling trait when heterozygous. This gene affects the formation of haemoglobin, but not for all, it only affects the structure of erythrocytes that assumes a sickle-like shape, that leads to haemolysis severe enough to cause an extreme and often fatal anaemia. Those who merely manifest the sickling trait appear on the other hand, to be perfectly healthy. Though their blood also contains the exceptional haemoglobin, but in smaller proportion, so the shape of the erythrocytes is normal when in circulation. It must be noted that the anaemia is recessive while the formation of the abnormal haemoglobin is not. In spite of the fact that the homozygotes suffer from this heavily lethal disease which usually eliminates them, the heterozygotes are quite common in certain regions of European population, as in some parts of Greece and Italy, and in African tribes. Evidently the heterozygotes must have an advantage which strongly counter-balances the destruction of the homozygotes in these areas. Allison (1954) discovered that the sickle-cell trait confers marked immunity against malaria especially, due to *Plasmodium falciparum*. The polymorphism is established only in those places where malaria is common.

Allison (1954) further observed polymorphism involving another genetically controlled disease, thalassaemia. Many homozygotes, and perhaps a few of the heterozygotes, die, yet the gene is present in 10 per cent of the population in

some of the districts of Greece and Italy where malaria is endemic. In India, particularly in parts of central India, such polymorphism exists where sickle-cell anaemia has been found to be prevalent in malaria endemic regions.

One of the oldest known such polymorphism is the ability to taste phenyl-thio-carbamide (PTC), or phenyl-thio-urea (PTU). For some people PTC has only a faint taste or no taste at all; for others it has a very bitter taste. More specifically there is a single dominant gene *T* (with incomplete penetrance) that determines a high sensitivity for the taste of PTC. Non-tasters are homozygous for the recessive allele *t*. When both parents are non-tasters, all their children are non-tasters. When one parent is taster and the other is not, either all or half of their children will be tasters, depending on whether or not the parent of the dominant (tasting) type is homozygote or heterozygote.

Genetic polymorphisms may be '*transient'* or '*balanced*'. Genetic polymorphisms are called balanced, if selection favors the heterozygotes. When selection favors the heterozygotes, a stable equilibrium may be achieved and substantial frequencies of both alleles may be maintained in one environment. The balanced or stable polymorphism is the result of natural selection operating as a *stabilizing* agent.

It is difficult to establish whether a polymorphism is stable or transient. However, direct evidence for at least one balanced polymorphism is available; the polymorphism for the group of haemoglobins, including haemoglobin S, in the presence of malaria.

3.4 HARDY-WEINBERG LAW

Definition

Mathematician Godfrey Hardy and physician Wilhelm Weinberg independently showed in 1908 that population gene frequencies remain constant from generation to generation under a system of random union of gametes in fertilization when the frequencies of the heterozygotes are equal to twice the product of the square roots of the two homozygotes: p^2 AA + 2pq Aa +- q^2 aa = 1, where p and q are the frequencies of genes A and a ($p + q$ = l) in the population, which is ideally large, with non overlapping generations, sexes equally distributed and all parents are equally fertile, and where there are no changes in gene frequency due to mutation, gene flow, selection or genetic drift, or where mutation and selection rates are balanced so that there is no net change in gene frequencies.

This theory is considered as the cornerstone of population genetics because it mathematically describes the behaviour of genetic traits through time within a specific unit — the population. Actually, the population assumed under Hardy-Weinberg Law is a unique population. It does not change genetically, i.e., it cannot and does not evolve. It is a so-called ideal population, i.e., a hypothetical one, which means that within it certain 'ideal' conditions must necessarily be fulfilled. The ideal population is a mathematical abstraction, because no real population ever fulfills all of the necessary conditions, that is, the population must be large, the sexes must be equally distributed, mating must be random (panmictic[1]), all parents must be equally fertile, and must be free from the four forces of evolution; that is, mutation, natural selection, genetic drift, and gene flow.

[1] A population undergoing random mating is often referred to as a panmictic population, or it is said to be in a state of panmixia.

The Hardy-Weinberg Law deals with the simplest genetic case, that of a single locus carrying only two alleles, *p* and *q*. The manner in which genetic stability is maintained under a two-allele system is best understood if the gene pool is visualized as divided into two component sexual units: one unit containing all the male gametes (spermatozoa), carrying the alleles *p* and *q*, the other unit containing all the female gametes (ova) in equal numbers. The relative proportions of *p* and *q* are identical between the two sex units. If all the male and female gametes mate randomly, the offspring will be distributed as shown in the box. Whether or not the gene pool is initially in equilibrium, after one generation of random mating, genetic equilibrium at a single locus is established and then perpetuated at the same gene frequencies through subsequent generations.

Male gametes → ↓ Female gametes	P	q
P	P^2	pq
Q	pq	q^2

Genotype frequencies: $p^2+2pq+q^2$

Let us examine what would be expected under random mating in a simple and general case of an autosomal locus with two alleles A and a with frequencies, *p* and *q* and the corresponding genotypes AA, Aa and aa with the corresponding frequencies, $p^2:2pq:q^2$. The various mating types and the expected progeny are given in the following table.

Mating Type	Frequency of Mating	Expected Frequency		
		AA	Aa	aa
AA x AA	P^4	P^4		
AA x Aa	$2p^3q$	p^3q	p^3q	
AA x aa	p^2q^2		p^2q^2	
Aa x AA	$2p^3q$	p^3q	p^3q	
Aa x Aa	$4p^2q^2$	p^2q^2	$2p^2q^2$	p^2q^2
Aa x aa	$2pq^3$		pq^3	pq^3
aa x AA	p^2q^2		p^2q^2	
aa x Aa	$2pq^3$		pq^3	pq^3
aa x aa	q^4			q^4
Total	*	$P^4+2p^3q+p^2q^2$ = $p^2(p^2+2pq+q^2)$	$2p^3q+4p^2q^2+2pq^3$ = $2pq(p^2+2pq+q^2)$	$p^2q^2+2pq^3+q^4$ = $q^2(p^2+2pq+q^2)$

*$p4+4p^3q+6p^2q^2+4pq^3+q^4 = p^2(p^2+2pq+q^2)+ 2pq(p^2+2pq+q^2)+ q^2(p^2+2pq+q^2) = p^2+2pq+q^2$

The above table presents a formal demonstration or derivation that $p^2+2pq+q^2$ is an equilibrium.

More precisely, Hardy-Weinberg equilibrium postulates a set of conditions where no evolution occurs. If all the conditions are satisfied, allele frequencies will not change (that is, no evolution will take place) and a permanent equilibrium will be maintained as long as these conditions prevail. However, it is obvious that the Hardy-Weinberg ideal population can never be found in the real sense in human populations. First, the formula provides a standard against which genetic change in a population may be measured and predicted. The formula serves as a basic theorem which can be expanded and elaborated by other mathematical models that deal with changes in populations (Jurmain et al 1998).

Secondly, the Hardy-Weinberg formula may be applied to large populations to provide an estimate of gene frequencies at a single point in time.

Population genetics is the study of allele frequencies in groups of organisms of the same species in the same geographic area.

The genes in a population comprise its gene pool.

Microevolution reflects changes in allele frequencies in populations. It is not occurring if allele frequencies stay constant over generations (Hardy-Weinberg equilibrium).

Five factors can change genotype frequencies - nonrandom mating, gene flow, genetic drift, mutation, and natural selection.

3.5 DEVIATIONS FROM HARDY-WEINBERG LAW OR FACTORS AFFECTING GENE FREQUENCIES

The discussion above relates to an 'ideal' population. By definition such a population is large and shows random mating with no new mutations, and no selection for or against any particular genotype. For some human characteristics, such as neutral genes for blood groups or enzyme variants, these criteria can be fulfilled. However, in genetic disorders, several factors can disturb the Hardy-Weinberg equilibrium by influencing either the distribution of genes in the population or by altering the gene frequencies. These factors include:

- Non-random mating
- Mutation
- Selection
- Small population size
- Gene flow (migration).

- **Non-random mating**

Random mating, or *panmixis,* refers to the selection of a partner regardless of that partner's genotype. *Non-random mating* can lead to an increase in the frequency of affected homozygotes by two mechanisms, either assortative mating or consanguinity.

Assortative mating

Assortative mating is the tendency for human beings to choose partners who share characteristics such as height, intelligence and racial origin for marriage.

Consanguinity

Consanguinity is the term used to describe marriages between blood relatives who have at least one common ancestor no more remote than a great-great grandparent. Widespread consanguinity in a community will lead to a relative increase in the frequency of affected homozygotes with a relative decrease in the frequency of heterozygotes.

- **Mutation**

The validity of the Hardy-Weinberg principle is based on the assumption that no new mutations occur. If a particular locus shows a high mutation rate then there will be a steady increase in the proportion of mutant alleles in a population. In that case the law will not be applicable.

- **Selection**

In the 'ideal' population there is no selection for or against any particular genotype. In reality for deleterious characteristics there is likely to be negative selection with affected individuals having reduced reproductive fitness in genetical sense, as the genes would not be transmitted in the next generation. In the absence of new mutations this reduction in fitness will lead to a gradual reduction in the frequency of the mutant gene and will cause disturbance of Hardy-Weinberg equilibrium.

Selection can act in the opposite direction by increasing fitness. For some autosomal recessive disorders there is evidence that heterozygotes show a slight increase in biological fitness as compared with unaffected homozygotes. This is referred to as heterozygote advantage. The best understood example is sickle-cell disease in which affected homozygotes have severe anemia and often show persistent ill-health. However, heterozygotes are relatively immune to infection with *Plasmodium falciparum* malaria because if their red blood cells are invaded by the parasite they undergo sickling and are rapidly destroyed. In areas in which this form of malaria is endemic, carriers of sickle-cell anemia, who are described as having sickle-cell trait, are at a biological advantage as compared with unaffected homozygotes. Therefore, in these communities, there will be a tendency for the proportion of heterozygotes to increase relative to the proportions of normal and affected homozygotes. Once again this will result in a disturbance of Hardy-Weinberg equilibrium.

We have earlier discussed about selection favouring heterozygotes as in sickle-cell anaemia, and thalassaemia. There is also the opposite situation, that is selection against heterozygotes, as we find in maternal-foetal incompatibility (Erythroblastosis fetalis) as is observed for the allele *R* (Rh blood group), and also for other blood group genes (Rh-ABO incompatibility).

Mutation alters genotype frequencies by introducing new alleles.

Heterozygotes and new mutations maintain the frequencies of deleterious alleles in populations.

Different alleles are more likely to confer a survival advantage in different environments. Cycles of infectious disease prevalence and virulence often reflect natural selection.

In balanced polymorphism, a disease-causing allele persists because heterozygotes resist a certain infectious illness or environmental condition.

Gene flow alters genotype frequencies by adding and removing alleles from populations.

Clines are gradual changes in allele frequencies between neighboring populations.

Geographical barriers and language differences often create great differences in allele frequencies.

Genetic drift occurs when a subset of a population has different allele frequencies than the larger population.

The founder effect occurs when a few individuals leave a community to start a new settlement. The resulting population may, by chance, either lack some alleles from the original population or have high frequencies of others.

- **Genetic drift**

In a large population the numbers of children produced by individuals with different genotypes, assuming no alteration in fitness for any particular genotype will tend to balance out, so that gene frequencies will remain stable. However, in a small population it is possible that by random statistical fluctuation one allele could be transmitted to a high proportion of offspring by chance, resulting in marked changes in allele frequency from one generation to the next, so that Hardy-Weinberg equilibrium is disturbed. This phenomenon is referred to as random genetic drift. If one allele is lost altogether then it is said to be extinguished and the other allele is described as having become fixed (www. faculty.ksu.edu).

- **Gene flow (migration)**

If new alleles are introduced into a population as a consequence of migration with subsequent intermarriage, this will lead to a change in the relevant allele frequencies. This slow diffusion of alleles across a racial or geographical boundary is known as gene flow. The most widely quoted example is the gradient shown by the incidence of the B blood group allele throughout the world. This allele is thought to have originated in Asia and spread slowly westward as a result of admixture through invasion.

3.6 CONSANGUINEOUS AND NON-CONSANGUINEOUS MATINGS

There are two general patterns of mating in human populations: random and non-random mating. Deviations from random mating can occur in two general directions. People who are related can either marry more frequently or less frequently than they would by chance. In the former case the mating system is one of inbreeding and in the latter one of outbreeding. Assortative mating is another important mating type which deviates from random mating. The assortative mating is either positive or negative. Inbreeding is defined as mating between close relatives. When the frequency of marriages between close relatives who have one or more common ancestors exceed the expected frequency under random mating in a population then it is called inbreeding and when it decreases the expected proportion then it is called outbreeding. Marriage between close relatives who have one or more common ancestors is called consanguineous marriage. Non-consanguineous marriages are between individuals of opposite sex who do not have a known common ancestor. Consanguinity refers to marriage type and inbreeding refers to the mating pattern of the population. Consanguinity is the term referred to describe the marriages between blood relatives who have one or more common ancestors and consanguinity is the name given to close

relationships (as distinct from relationships by marriage). In positive assortative mating, individuals tend to choose mates who resemble themselves (e.g., in native language, intelligence, stature, skin colour, musical talent, or athletic ability) more frequently than would be expected by chance. In negative assortative mating, the mating pairs are dissimilar in phenotype than would be expected by chance.

All societies have rules which forbid marriage between close blood relatives such as parent offspring and sibs (brother and sisters) called incest taboo. Though incest taboo is a universal feature of human society, it is complemented by a preference for marriage between certain other relatives. The most common form of consanguinity in the human population is cousin marriage. Marriage between children of siblings of the same sex (parallel cousins) is prohibited except in some Islamic societies of the Middle East where marriage between a man and his father's brother's daughter is common. There are in certain areas (South India, Japan, etc.) where marriages are commonly observed between the children of the siblings of opposite sexes (cross cousins). First cousin marriages make up almost 10 per cent. In southern part of India, especially in the state of Andhra Pradesh, among certain castes, uncle-niece unions also make up about 10 per cent of marriages. Less frequent marriage types also occur in this part of India such as the marriages between first cousins once removed, second cousins, double first cousins and aunt-nephew.

The possible types of mating between different relationships are shown in the following figure.

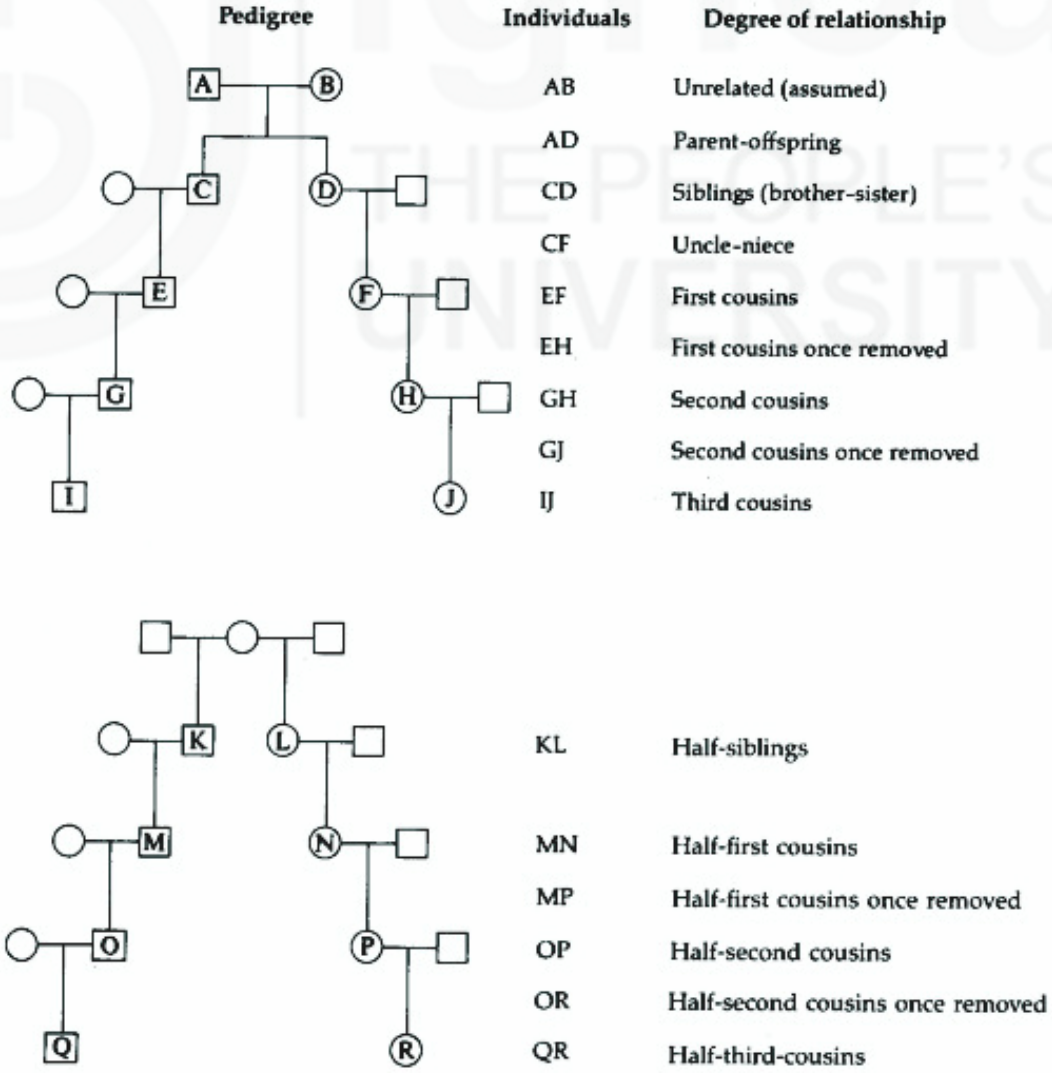


Fig. 4.1: The possible types of matings between different relationships

People choose partners for marriage, and they do not contribute the same numbers of children to the next generation. The marriage practices change allele frequencies in populations.

Traits lacking obvious phenotypes may be in Hardy-Weinberg equilibrium.

Consanguinity and endogamy increase the proportion of homozygotes in a population.

Effect of Consanguineous Marriages

The main genetic consequence of inbreeding is an increase in the proportion of homozygotes. Through inbreeding recessive genes are more easily brought to the fore.

Inbreeding Depression

Usually, inbreeding causes deterioration and outbreeding causes improvement of most of the characters. Animal breeders noticed that inbreeding particularly always lead to a deterioration in many important qualities; fertility for instance, tends to decrease and many an inbred stock, has lost because the fertility level became too low for the maintenance of the line in generations. In addition, some traits such as overall general size also decrease. This phenomenon of deterioration on inbreeding is known as *inbreeding depression*.

Heterosis

In contrast to inbreeding depression, if two independent pure lines are crossed, the hybrids between them (at least in the first generation) mostly show a considerable increase in size, fertility and many other desirable traits. This has been called *hybrid vigor* or *heterosis*, and clearly has a great potential for application in agriculture and animal husbandry. The first practical application of hybrid vigor as a technique for crop improvement was applied to corn and it led to a very significant increase in production. This practice is now being extended to other plants and animals. These inbreeding and outbreeding consequences are also seen in man. The genetic effects of inbreeding are similar to positive assortative mating. Both increase the frequency of homozygous genotypes at the expense of heterozygotes, relative to Hardy-Weinberg proportions. So it is clear that the inbreeding affects genotype frequencies and inbreeding along with selection modifies gene frequencies in a population.

It should be emphasised that the increasing homozygosity i.e., the general effect of inbreeding does not predict whether inbreeding is good or bad. It depends on the nature of the homozygotes. Many instances can be cited of talented persons whose parents were first cousins or otherwise closely related. Presumably consanguinity made it easier for 'good' genes to come together in these cases (example: Charles Darwin).

On the other hand, there is considerable evidence that homozygous recessives, albinism, alkaptonuria, etc., and the lethals are encountered with greater frequency in consanguineous marriages than in marriages of unrelated persons. Studies in Japan, where inbreeding is greater have shown increased rates of infant mortality and congenital abnormalities. Studies in France, Sweden, United States, and Japan have shown increased frequencies of certain physical diseases, and mental disorders among children of first cousin mating.

3.7 GENETIC LOAD

Among source of variability affecting Darwinian fitness (adaptive value) may lead to a genetic load. Crow (1970) proposed three definitions of genetic load of which mostly used one is that the (expressed) genetic load is the fraction by which the average population fitness is decreased in comparison with the genotype showing the highest fitness.

It appears that some polymorphisms exist because recurrent mutations replace genes lost to selection, whereas others exist because the heterozygote is adaptively superior and causes several alleles to persist even though many are lost due to selection against both homozygotes. The loss of individuals — often unseen individuals — under either situation because they carry certain genes, has been termed as genetic load of a species or population. So it is obvious that every human population carries a burden of deleterious mutations which impairs the fitness of the group. So the genetic load refers to the proportion by which fitness is reduced in the population due to the operations of a factor such as mutation.

So the genetic load of a species is a measure of the number of deleterious traits maintained in a population or of the damage to the population by the factors under study. It may be measured as decreased average fitness, or somewhat more specifically, as mortality, sterility, or morbidity due to specified causes, usually deleterious alleles. The genetic load of a species may be partially hidden and partially manifested. The genetic load depends on several variables — the occurrence of mutations, the number of detrimental mutations, the number of mutant recessive alleles, and the number of partially lethal mutant dominant alleles.

Genetic Radiation Hazard

In every generation numerous mutations, of every possible degree of harmfulness, will arise in human species; and in every generation, the carriers of some of these mutants — persons afflicted with hereditary diseases, malformations, or constitutional weaknesses — will die before they have children, or will remain unmarried, or will produce fewer children than they would have produced if they did not carry the mutant genes in question. The burden of genetic ill-health and abnormality in human populations is very great. And this is more so because of the genetic hazards of radiation. High-energy radiations cause two kinds of damage to living matter — physiological and genetic. Physiological damage consists of radiation burns, radiation sickness, and death, which occur soon after the irradiation (as had happened when an Atom Bomb was dropped on the twin cities of Japan by the Americans in 1945), and of various delayed effects, such as malignant growths. Genetic damage includes the mutations induced in the reproductive tissues and transmitted to the progeny. The genetic damage may inflict harm on the descendents of the exposed persons, and that too for many generations after the exposure.

3.8 SUMMARY

A population is a group of interbreeding members of the same species in a particular area. Their genes constitute the gene pool. Population genetics considers allele, genotype, and phenotype frequencies to reveal microevolution. Phenotypic

frequencies can be determined empirically. Genotype frequencies change if migration, nonrandom mating, genetic drift, mutations, or natural selection operate. In Hardy-Weinberg equilibrium, frequencies are not changing. Hardy and Weinberg proposed an algebraic equation to explain the consistency of allele frequencies. The Hardy-Weinberg equation is a binomial expansion used to represent genotypes in a population. According to Hardy-Weinberg equilibrium all individuals mate with the same frequency and choose mates without any consideration to phenotype. This seldom happens. We choose mates based on certain characteristics, and some people have many more children than others. Consanguinity increases the proportion of homozygotes in a population, which may lead to increased incidence of recessive illnesses or traits.

Clines are changes in allele frequencies from one area to another. Clines may reflect geographical barriers or linguistic differences and may be either abrupt or gradual. Genetic drift occurs when a small population separates from a larger one, or its members breed only among themselves, perpetuating allele frequencies not characteristic of the larger population due to chance sampling. A founder effect occurs when a few individuals found a settlement and their alleles form a new gene pool, amplifying their alleles and eliminating others. Mutation continually introduces new alleles into populations. Mutation does not have as great an influence on disrupting Hardy-Weinberg equilibrium as the other factors. The genetic load is the collection of deleterious alleles in a population. Environmental conditions influence allele frequencies via natural selection. Alleles that do not enable an individual to reproduce in a particular environment are selected against and diminish in the population, unless conditions change. Beneficial alleles are retained. In balanced polymorphism, the frequencies of some deleterious alleles are maintained when heterozygotes have a reproductive advantage under certain conditions.

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Sample Questions

1) What is a population? List three populations.
2) Explain the differences among an allele frequency, a phenotypic frequency, and a genotypic frequency.
3) What does Hardy-Weinberg equilibrium mean?
4) What are the conditions under which Hardy-Weinberg equilibrium cannot be met?
5) Why is knowing the incidence of a homozygous recessive condition in a population important in deriving allele frequencies?