BLOCK 3
Human Population Structure and Disease Pattern

BLOCK 3 : HUMAN POPULATION STRUCTURE AND DISEASE PATTERN

UNIT 7 MATING PATTERNS

Contents

Learning Objectives

After reading this unit, you will be able to:

- Understand the importance of mating pattern in the human population;
- Learn about different types of mating patterns (random and non-random mating);
- Describe the assortative mating; and
- Know about positive and negative assortative mating.

7.0 INTRODUCTION

Mating refers to selecting a mate by the opposite sex, who participates in sexual reproduction, thereby transmitting hereditary characters from one generation to another. The transmission of hereditary characters depends on who mates whom, and the pattern of male-female pairings is known as 'mating system' or 'mating pattern'. The term 'mating system' or 'mating pattern' occupies an essential component in reproductive biology. In a human population, the selection of mates is determined by different societal norms unique to each culture.

A mating system concerns the distribution of genes in the population, and ultimately reproduction; within a population, defining which males' mate with which females and the conditions under which those mating occur. Mating systems are evolved outcomes; they emerge from the costs and benefits of males and females, affecting mate choice and competition for mating opportunities, which in turn leads to adaptations. However, mating systems also affect subsequently evolved outcomes. They impose selective pressures on males and

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females in that they affect the reproductive strategies and tactics most beneficial, under various circumstances, to male and female individuals. In brief, the mating systems exhibit all the possible dimensions of sexual reproduction.

In general, mating is restricted within the species; thus, the genetic characters are transmitted through generations and confined to a given species. Therefore, mating within a species is compatible, and mating between species is unlikely and is supposed to be incompatible. However, there is an increasing interest in epidemiological questions in the population structure, emphasizing the distribution of genetic and physiological attributes, how they are related to the distribution of specific diseases, and how population structure influences these distributions. Thus, predicting mating behavior would be highly beneficial to studying population structure in both its traditional and more recent forms. One technique that can help to evaluate the importance of cultural preferences and biological, demographic, and social constraints on mating patterns is potential mates' analysis.

The best model of the mating pattern is presented by Thomas Malthus (1872) in his "Essay on the principle of population". Mating patterns are essential in shaping the structure of the human population, including demography, preserving the genetic structure, impacting the constitution and pace of change, and providing a mechanism for social and cultural cohesiveness that keep the population in check and then allow it to grow. The mating patterns and preferences have been the core research areas for anthropologists and population geneticists for more than a century. Charles Darwin (1871), in his book 'Descent of Man and Selection in Relation to Sex', recognized sexual or mate selection as an important causative factor as that of natural selection in evolution. Anthropologists focus on the genetic structure of a population determined by mating pattern and evolutionary changes produced by it. It may not be worth mentioning here that genotype frequencies are determined in part by mating patterns.

Check Your Progress

1) What do you mean by mating pattern?

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7.1 STRATEGIES OF HUMAN MATING

Mating is the core of the evolutionary process that produces adaptations. It is known as the differential reproductive success of individuals because of the heritable differences in corresponding genetic traits. Homo sapiens sapiens have descended from a common ancestor who had started practicing mating in different forms. The descendants have inherited the mating strategies and

propelled them to success. According to some theories, people look for mates who resemble the opposite sex, or mates who have similar characteristics or opposite to one's own, or mates to make an equitable exchange of valuable resources. According to the sexual strategies theory, the patterns in mating behavior have survived as they are evolutionarily adaptable. Sexual behaviors evolved to aid our forefathers in reproducing successful progeny. Mating is a universal urge practiced by people of all cultures and has an undeniable evolutionary underpinning. Successful mating requires solutions to several complex adaptive problems. These include selecting fertile mate, engaging in all of the necessary sexual and social behaviors required for successful conception, childbirth, and child-rearing to occur. One of the interesting features of human mating is that a single strategy cannot characterize human mating patterns. Long-term committed mating is one of the strategies, often, but not always, characterized by a formal public commitment such as marriage. In such long-term mating, both the partners equally participated in the resultant offspring. Consequently, according to the theory of parental investment, sexual selection favors both sexes' high levels of selectivity. Not all human mating, however, lasts a long time. The mating relationship can last a few months, or a few weeks, or a few days, or even a few minutes. This end of the temporal continuum is known as short-term mating (Buss & Schmitt, 1993). Furthermore, we humans display remarkable creativity and have the ability to mix and match mating strategies.

Humans are neither solely monogamous nor solely promiscuous; neither polygynous nor polyandrous, which strategies a particular person chooses deeply dependent on circumstances. The circumstances may include the sex ratio in the mating pool (i.e., the ratio of males to females that are looking for a mate), a person's mate value (i.e., how desirable they are to the members of the opposite sex), and the prevailing cultural norms of the ethnic groups (Buss, 2004).

7.1.1 Patterns of Spouse Selection

In human society, choosing of mate mostly happens non-randomly concerning certain favourable characteristics. Some of the patterns for the selection of mates practices in India are as follows:

Endogamy

Endogamy is a regulation of selecting mates that states that a person must marry inside a particular or defined group to which they belong. An endogamous group may be within a caste, clan, racial, ethnic, or religious group. In India, religious and caste endogamy are two of the most common types of endogamy. Inter religious weddings are lawful, but they are not often practiced or familiar. The functioning of endogamy varies depending on the place and the type of religion. For example, marriage within relatives is favoured among several caste populations in South India. Marathi, Telugu, Tamil, and Kannada speaking communities prefer marriages with cross-cousins (children of father's sisters or mother's brothers).

Hypergamy

The act of a person marrying a higher caste person or having higher social status than themselves is known as hypergamy. The husband's position is always higher than the wife's, according to hypergamy's rule. Those who observe this guideline always seek males with a better social status than their daughters. This practice has happened chiefly inside castes or subcastes rather than between castes. All castes incline hypergamous stratification. For example, hypergamy has been observed among groups such as the Rajput and Jat of North India, the Anavil Brahmin and Patidar of Gujarat, the Maithil Brahmin of Bihar, the Rarhi Brahmin of Bengal, and also to a lesser extent among the Kanyakubja and Saryupari Brahmin of Uttar Pradesh.

Exogamy

Marriage between the members of a specific community is prohibited. Therefore, there is a custom of marrying outside the clan, caste, or community. Restriction of marriage may include all people with whom genealogical kinship can be traced, or it may be broad enough to cover all those with whom genealogical kinship can be traced. For example, in North India, a girl born within a village is considered the village's daughter and cannot marry a male from her community. In South India, one's sisters or brothers and classificatory parallel cousins constitute the exogamous unit of the considered generation. Therefore, they could not get married among themselves.

Arranged Marriages

In India, most marriages are set or arranged on behalf of the boy or girl engaged in the marriage. An arranged marriage is one in which their parents or elders choose the bride and groom. The practice of arranged marriages in India increases because of the certain restrictions in the rules of selection of mates among different societies, namely (i) the existence of endogamy rules that limit marriage alliance within certain groups, (ii) exogamy rules that prohibit marriage within a gotra. As a result of these considerations, arranged marriages are the ideal method of selecting a mate. In most societies in India, restrictions are placed on avoiding the free interactions between a boy and a girl for choosing their mate. Though the measure of participation in choosing one's life partner has shown variations between different groups, by and large, a marriage arranged by parents or elders is the most prevalent form of selection of a mate in India.

In brief, the mating patterns practiced by the human populations could be broadly categorised into two groups. These are generally classified as random mating and non-random mating. These mating patterns are described below.

7.2 RANDOM MATING

The term random mating is defined as the mating of individuals regardless of any form of preferences like physical, genetic, or social. In simple words, the random mating between two individuals is not influenced by any external

factors, including environmental, hereditary, or social interaction. There is an equal chance of being selected for mates, i.e., an individual of one sex has an equal probability of mating with any individual of the opposite sex in the population. It is the simplest type of mating system in the human population. No factors are present to cause a genetic change in a random mating population from generation to generation. A population undergoing random mating is often referred to as a panmictic population, or it is said to be in a state of panmixia. In random mating, mate preferences and social factors prevent marriages in a random manner. Whether a mating is random or not depends on the particular character or locus. For example, people do not usually ask about the blood groups of the partner before marriage. Therefore, in practical, we can assume that the distribution of blood groups in the population is the outcome of random mating. In simple words, the choice of a partner, in random mating, is not influenced by the genotypes. In random mating, the union of individuals with the particular trait of interest occurs according to the product rule of probability.

Random mating is the simplest form of mating in which individuals choose mates randomly and independently of the genotypes of interest. It implies that the probability of genotypes being mates is the product of the frequencies of the two genotypes. The implications of this mating system were modeled by Wilhelm Weinberg (1908), a German physician, and Geoffrey Hardy (1908), an English mathematician working on the inheritance of Mendelian traits in the human population. They independently developed and remained a cornerstone of population genetic theory. In a population such as we are considering, random mating of zygotes is equivalent to the random union of the total gametic output of the population and the effect of one generation of mating on the frequencies of the gametic genotypes (Geiringer, 1944).

Box 7.1 : Random Mating

Random mating does not mean promiscuity; it simply means . . . that in the choice of mates . . . there is neither preference for nor aversion to the union of persons similar or dissimilar concerning a given trait or gene. Thus, a population may be randomly mating with regard to most loci but be mating non-randomly concerning to others that are influencing mate choice (Wallace and Dobzhansky, 1959).

Primary importance in population genetics determines Genetic frequencies in a large population of sexually reproducing organisms mating at random in non-overlapping generations with negligible mutation and selection. Random mating is one of the essential factors assumed in the Hardy-Weinberg principle (Edwards, 2008). The gene frequency remains constant, and the Genetic distribution for any Mendelian factor reaches a stationary state in the first generation of random mating (Hardy, 1908). According to him, random mating is equivalent to randomly drawing a gamete out of the gene pool. In other words, the probability of drawing a gamete bearing a particular allele is the same as the frequency of that allele in the gene pool. Therefore, drawing a second gamete from the gene pool is at random and independent from the

first. During the early 20th century, Hardy and Weinberg demonstrated that in a randomly mating population, the population's gene pool would not change if the population is not subject to any other evolutionary process. The population will remain in equilibrium. For example, let us consider a particular trait 'height' in a population controlled by the dominant 'A' allele and recessive 'a' allele. If the population follows random mating, there are nine possible mating patterns of the trait controlled by alleles 'A' and 'a' (Table 7.1). When the population has equal frequencies (50%) of each of the two alleles, the expected offspring genotype frequencies with random mating are shown below. The genotype frequencies for AA homozygous dominant and aa homozygous recessive will be 25% each; Aa heterozygous will be 50%. The population will remain with this ratio for every generation if random mating continues and no other evolutionary forces are operating.

Table 7.1. : Mating pattern of a trait in a random mating population

Possible parent mating		**Expected offspring genotype frequencies**		
Male	Female	AA	Aa	aa
AA	AA	4		
AA	Aa	2	2	
AA	aa		4	
Aa	AA	2	2	
Aa	Aa	1	2	1
Aa	aa		2	2
aa	AA		4	
aa	Aa		2	2
aa	aa			4
Total		**9 (25 %)**	**18 (50%)**	**9 (25%)**

The effect of random mating on the distribution of allele frequencies is the foundation of 'Population Genetics'. The consequences of random mating are fully demonstrated in Hardy-Weinberg Law. The law states that the allele frequencies will remain the same from generation to generation, if the population follows random mating provided no external forces are acting on it, no differential viability or fertility of genotypes, no mutation, and that the population is sufficiently large to obviate unrepresentative sampling of gametes.

7.3 NON-RANDOM MATING

In human populations, the mating pattern is random in nature. However, people usually choose mates non-randomly, particularly on easily observable traits. In most societies, cultural values and social rules primarily control the selection of mates. In a community, matings are commonly made between similar people with similar traits, such as in between white skin colours, tall statures, and rich families. In general, individuals in populations do not always follow random mating patterns. Non-random mating occurs when a person prefers mates. The preference could be of any characteristics of one's choice. It is based on the

phenotypes rather than between relatives. For example, it could be particular superior characteristics, similar traits to themselves, arranged marriage, or aesthetics. In non-random mating, the selection of mates does not occur by chance, and there is always human interference in selecting mates. It occurs when the probability that two individuals in a population will mate is not the same for all possible pairs of individuals. In simple words, non-random mating means the selection of a mate is influenced by phenotypic differences. Generally, it is based on underlying Genetic differences.

Practicing non-random mating in a population can increase the frequency of homozygotes by two possible mechanisms that are either assortative mating or consanguinity. A population substructure can also be formed from geographic stratification and or non-random mating. Assortative mating is another form of mating type that deviates from random mating. The term 'assortative' refers to classifying and selecting traits. Assortative mating is non-random mating because of more or less frequent mating among specific individuals than expected in a random mating population. It is the tendency for human beings to choose partners who share similar characteristics such as height, intelligence, and racial origin for marriage.

Assortative mating occurs when some observable phenotype or social characteristic influences the selection of a spouse. Preference for choosing mates based on phenotypic characters is well known in the human population. Phenotypic choices are made on several physical characteristics, including age, height, weight, skin pigmentation, eye, and hair color. Other behavioral and social factors commonly practiced in mate selection are educational level, occupation, socioeconomic status, religion, smoking, alcohol consumption, language, and culture. It is also affected by several other biological and social factors. Assortative mating can impact on genetic structures of a population if associated with genetic variation. It may also affect heritability estimates, create correlations among different traits that were initially unrelated, and affect trait variance within and between the populations.

Assortative mating increases the homozygosity of variants associated with the traits influenced by selecting mates, leading to an increase in genetic variance. The variance will not cause any change in allele frequencies unless there is any differential selection acting in the population. The genetic contribution to variance in polygenic traits increases due to assortative mating (Wright, 1921). Assortative mating could be classified into positive assortative mating and negative assortative mating. Preferred mating partners can have more similar (positive) or dissimilar (negative) phenotypes than expected in random mating. In a population, positive assortative mating or negative assortative mating occurs if the mated pairs are composed of identical phenotypes more often or less often than expected by random mating, respectively (Hedrick, 2016). Phenotypically similar individuals might have identical genotypes, and it is commonly observed in consanguineous, non-random mating, resulting in inbreeding.

Box 7.2 : Sexual Selection

Sexual selection is defined as the members of one biological sex select partners of the other opposite sex to mate with competing with the other members of the same sex. In humans, the selection of mates is the primary mechanism of sexual selection. It is the selection of most preferred females by males possessing specific characteristics. It favors the traits that help in competition over mates. The theory of sexual selection deals with the evolution of characteristics due to mating rather than survival (Darwin, 1859; 1871).

Check Your Progress

2) Differentiate between random and non-random mating?

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7.3.1 Positive Assortative Mating

Positive assortative mating is the most common non-random mating pattern in humans. It occurs when individuals mate with people who are phenotypically or genetically like themselves. Positive assortative mating is also known as homogamy. In positive assortative mating, there is a greater chance of likes mating each other than unlikes. In such mating, individuals tend to choose mates who resemble themselves more frequently than would be expected by chance. The selection of phenotypically alike mates may be based on the biological, social, or both of the characteristics. It may include intelligence, stature, or skin pigmentation. For example, a mating of an individual having a tall and slender phenotype with the individual having a similar phenotype that is tall and slender. When we consider mating in terms of genotypes in extreme conditions, it is positive assortative mating between individuals with identical genotypes for traits controlled by two autosomal alleles. In such case, the possible mating would be homozygous dominant with homozygous dominant genotypes (AA X AA), heterozygous with heterozygous genotypes (Aa X Aa), and homozygous recessive with homozygous recessive genotypes (aa X aa) (Table 7.2). The effect of such positive assortative mating will lead to the progressive increase in homozygous genotype frequency (42%, AA and aa) and a corresponding decrease in heterozygous genotype frequency (17%, Aa) in the population. If positive assortative mating is practiced in each generation, then this polarizing trend will continue in the population.

Table 7.2. : Mating pattern of a trait in a positive assortative mating population

Possible parent mating		Expected offspring genotype frequencies		
Male	Female	AA	Aa	aa
AA	AA	4		
Aa	Aa	1	2	1
aa	aa			4
Total		**5 (42%)**	**2 (17%)**	**5 (42%)**

7.3.2 Negative Assortative Mating

Negative assortative mating is the least common non-random mating pattern in the human's population. It occurs when people select mates who are phenotypically different from themselves on certain traits. It is also referred to as heterogamy. In negative assortative mating, the mating pairs are dissimilar in phenotype than would be expected by chance. For example, it would occur if tall and slender people were mated only with short and stout individuals. Negative assortative mating is just the opposite of positive assortative mating.

When we look into the negative assortative mating pattern of a particular trait controlled by two autosomal alleles, there are six possible mating patterns by the combination of three different genotypes. The genotypes are homozygous AA genotypes, heterozygous Aa genotypes, and recessive homozygous aa genotypes controlled by the dominant 'A' and recessive 'a' alleles (Table 7.3). In a population, the effect of negative assortative mating is a progressive increase in the frequency of heterozygous genotypes (66.6%, Aa), and a corresponding decrease in homozygous genotypes (16.7%, AA and aa). It may be noted that negative assortative mating has the reverse effect, increasing in heterozygous genotype frequencies as opposed to that of positive assortative mating, which increases homozygosity in the population.

Table 7.3. : Mating pattern of a trait in a negative assortative mating population

Possible parent mating		Expected offspring genotype frequency		
Male	Female	AA	Aa	aa
AA	Aa	2	2	
AA	aa		4	
Aa	AA	2	2	
Aa	aa		2	2
aa	AA		4	
aa	Aa		2	2
Total		**4 (16.7%)**	**16 (66.6 %)**	**4 (16.7%)**

Check Your Progress

1) What are the different types of assortative mating? Explain.

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7.4 CONSEQUENCES OF ASSORTATIVE MATING

Positive assortative mating, which happen to be between close relatives, may lead to inbreeding as we know that inbreeding is the genetic outcome of consanguineous marriage. The progeny of consanguineous parents is, by definition, inbreed. Consanguineous marriage is a marriage between genetically related individuals who have one or more biological ancestors in common. Therefore, positive assortative mating between close relatives increases the chance of inbreeding as there is a greater chance that the partners will have genes in common. Relatedness is a state of infinite degree. We all humans are related at some stage in the history of evolution as we have evolved from common ancestors.

In most societies, marriage regulates mating patterns, and consanguineous marriage leads to inbreeding. The most common form of consanguineous marriage with appreciable frequency is first-cousin marriage. Though it is not common among all the human populations but it does occur. Consanguineous marriages are practiced in different forms to a greater or lesser extent among most religious and ethnic groups. In India, the highest frequencies of inbreeding population resulting from consanguineous marriage are reported from South India.

Inbreeding is considered as the first general class of departures from random mating. It increases the degree of homozygosity to the offspring and also the expression of recessive traits. In an inbreeding population, the percentage of heterozygotes is reduced by a half in each generation and increases both the type of homozygotes i.e., dominant and recessive homozygous genotypes. This effect holds true in any form of inbreeding if practiced in the population. If the parents have a more distant relationship, there will be a lesser rate of decrease of heterozygosity. In humans, the closest type of mating practiced is between first-cousins.

The relationship between two individuals is measured genetically as the 'coefficient of relationship'. It is the probability by which at a particular locus, both the individuals possess a gene identical by descent. For example, the relationship coefficient between parents and children and in between siblings is half (1/2). It means 50% of their genes are in common. Similarly, the coefficient of relationship between grandparent and grandchildren, uncle and aunt, or in

between nephew and niece is 1/4 (i.e., 25% of their genes are shared). The coefficient relationship between the first cousins is 1/8, which means 12.5% of their genes are common.

The consanguineous marriages are important from the genetic point of view, as these may lead to a greater manifestation of recessively inherited conditions owing to increased homozygosis. For example, an increased likelihood of having an affected offspring (genetically recessive homozygous disorder) if a carrier marries a cousin rather than an unrelated person. Through inbreeding, deleterious recessive genes achieve the stage of homozygosity. And it may be further associated with 'inbreeding depression' due to decreased heterozygosity in the population. The heterozygotes have greater biochemical versatility and then homozygotes increasing the degree of population biological fitness.

On the other hand, the unlike individuals (i.e., those with different genetic or phenotypic characteristics) have better chances of mating with each other in negative assortative mating. Sometimes, it is also observed that the immigrant groups are marrying outside of their community. Such situations lead to hybridization and increases in heterozygosity in the population as genetic consequences. Generally, it is believed in genetics that individuals with many heterozygous traits show greater vigour, viability, and fertility when compared to individuals with homozygous conditions. This increase in viability in heterozygous individuals is collectively known as 'Heterosis' or 'Hybrid Vigour'. Heterosis is a phenomenon complementary to 'inbreeding depression'. In other words, the phenomenon of heterosis is simply inbreeding depression in the reverse direction. It can be better understood by crossing the inbred lines. The progeny of such crossing will show an increase in the characters that previously reduces fitness from inbreeding. The fitness lost due to inbreeding tends to be restored among the progeny of crosses between the inbred strains.

Check Your Progress

4) What do you know about the consequences of assortative mating?

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7.5 SUMMARY

Mating determines the transmission of hereditary characters from one generation to another. In a population, mate choice is defined by various rules and regulations of marriage depending on the culture one follows, forming specific patterns of male-female pairs called the mating system. In population genetics, the mating patterns are classified into random mating and non-random mating.

Random mating is defined as the mating of individuals regardless of any physical, genetic, or social preference. A random mating population is the simplest model of population genetics in which no factors are present to cause a hereditary change from generation to generation. Therefore, the population will be in Hardy Weinberg Equilibrium. On the other hand, non-random mating occurs when mate selection was made based on a particular observable phenotype or social characteristics preference. The non-random mating is represented by assortative mating, which is deviated from random mating. Assortative mating could be further divided into positive and negative assortative mating. In positive assortative mating, individuals tend to choose mates who resemble themselves, whereas the mating pairs are dissimilar in phenotype than expected by chance in negative assortative mating.

Both types of assortative mating have significant effects on the genetic structure of the human population. Positive assortative mating may increase the recessively inherited traits owing to increased homozygosis. In contrast, the negative assortative mating increases heterozygosity as genetic consequences in the population. Positive assortative mating increases the chance of inbreeding, and the deleterious recessive genes achieve the stage of homozygosity leading to 'inbreeding depression'. However, negative assortative mating increases the viability in heterozygous individuals, collectively known as 'heterosis'. The phenomenon of heterosis is inbreeding depression in the reverse direction. The fitness lost because of inbreeding effects tends to be restored among the progeny of crosses between the inbred strains. Therefore, non-random mating operates selection causing evolutionary processes of a population.

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7.7 ANSWERS TO CHECK YOUR PROGRESS

1) Mating pattern refers to selecting a mate by the opposite sex, who participates in sexual reproduction, transmitting hereditary characters from one generation to another. The transmission of genetic characters depends on who mates whom, and the pattern of male-female pairings is known as 'mating system' or 'mating pattern'. The term 'mating system' or 'mating pattern' occupies an essential component in reproductive biology. Mating patterns are essential in shaping the structure of the human population, including demography, preserving the genetic structure, impacting the constitution and pace of change, and providing a mechanism for social and cultural cohesiveness that keep the population in check and then allow it to grow.

2) Random mating is the simplest form of mating in which individuals choose mates randomly and independently of the genotypes of interest. In random mating, mate preferences and social factors prevent marriages in a random manner. No elements are present to cause a genetic change in a random mating population from generation to generation. Random mating is one of the essential factors assumed in the Hardy-Weinberg principle. In a randomly mating population, the population's gene pool would not change if the population is not subject to any other evolutionary process. The population will remain in equilibrium.

   Non-random mating occurs when a person prefers mates. The preference could be of any characteristics of one's choice. It is based on the phenotypes rather than between relatives. In non-random mating, the selection of mates does not occur by chance, and there is always human interference in selecting mates. It occurs when the probability that two individuals in a population will mate is not the same for all possible pairs of individuals. In simple words, non-random mating means the selection of a mate is influenced by phenotypic differences.

3) Assortative mating is non-random mating because of more or less frequent mating among specific individuals than expected in a random mating population. Assortative mating could be classified into positive and negative assortative mating. Preferred mating partners can have more

similar (positive) or dissimilar (negative) phenotypes than expected in random mating. In a population, positive-assortative mating or negative-assortative mating occurs if the mated pairs are composed of identical phenotypes more often or less often than expected by random mating, respectively. Positive assortative mating is the most common non-random mating pattern in humans. It occurs when individuals mate with people who are phenotypically or genetically like themselves. It is also known as homogamy. In positive assortative mating, there is a greater chance of likes mating each other than unlikes. In such mating, individuals tend to choose mates who resemble themselves more frequently than would be expected by chance. Negative assortative mating is the least common non-random mating pattern in the human population. It occurs when people select mates who are phenotypically different from themselves on certain traits. It is also referred to as heterogamy. In negative assortative mating, the mating pairs are dissimilar in phenotype than would be expected by chance.

4) Positive assortative mating between close relatives increases the chance of inbreeding as there is a greater chance that the partners will have genes in common. Inbreeding is considered as the first general class of departures from random mating. It increases the degree of homozygosity to the offspring and also the expression of recessive traits. The unlike individuals (i.e., those with different genetic or phenotypic characteristics) have better chances of mating with each other in negative assortative mating. Such situations lead to hybridization and increase in heterozygosity in the population as genetic consequences. Generally, it is believed in genetics that individuals with many heterozygous traits show greater vigour, viability, and fertility when compared to individuals with homozygous conditions. The negative assortative mating increases viability in heterozygous individuals.

UNIT 8 BIOLOGICAL CONSEQUENCES OF MATING SYSTEMS

Contents

Learning Objectives

After reading this unit, you will be able to:

- Understand consanguineous marriages and its consequences;
- Identify the prevalence of consanguinity in Indian population;
- Define inbreeding and outbreeding: their differences and significance; and
- Explain the degree of biological relationship and Genetic load.

8.0 INTRODUCTION

Mating is the pairing of male and female individuals, usually for sexual reproduction. The human mating system is a set of individuals behaviors to select, attract, and retain mates. It is a product of a natural selection called 'sexual selection'. The mating system refers to the general pattern by which males and females mate within a population or species. Thus, the system indicates how individuals are grouped and how mating achieves through time in a population. The diversity of human mating systems has a significant role in testing evolutionary hypotheses and shaping a population's genetic structure. The variation in the mating patterns and demographic history of mating, plays a substantial role in shaping nucleotide variation within and among the people.

The uniqueness of human mating strategies asserts in their relationship with certain cultural variables, that is, the institution of marriage, a union between a man and a woman. Generally, marriage is considered one of the most complex

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relationships in human society. Marriage forms specific rules and norms that regulate reproduction in all human societies. Marriage shows a considerable variation with great ethnic diversity. Consanguineous marriage, marriage between closely related individuals, is one of the common types of marriage practiced in different parts of the globe since ancient times. By closely related individuals, we mean not beyond great great grand parents (not beyond third cousin). The consanguineous marriage is deeply rooted in the beliefs and socio-cultural norms of society. Consanguineous marriage can be categorized based on the level of relationships between spouses. For example, a marriage occurred between progeny of sibling such as first-cousins marriage, second cousins' marriage and, uncle-niece marriage. Among the different types of consanguineous marriage, the first cousin marriage is the most common form found with appreciable frequencies in other societies.

Inbreeding is the outcome of consanguineous marriage. The term 'inbreeding' is defined as the mating of individuals with one or more biological ancestors in common. It means mating between genetically related individuals. The progeny of consanguineous mating is described as 'inbreed'. The term consanguineous is more qualitative and often used in anthropology and social sciences to describe marriage patterns. It describes the relationship of individuals through marriages. However, it is also used in studying human population genetics for its significant contribution in shaping the genetic structure. On the other hand, inbreeding is more quantitative and often use in clinical genetics, describing the degree of biological relationship and consanguinity between the parents and their offspring. Inbreeding is departure from random mating and the genetic consequences of consanguineous marriages.

Consanguinity increases the degree of inbreeding. Concisely, inbreeding is considered as the genetic consequence of consanguineous marriage. Moreover, different countries show distinct factors associated with the prevalence of consanguinity, such as socio-economic status, religion, rural and urban settings. Parental consanguinity has been associated with adverse outcomes on reproductive health, morbidity, infant and child mortality. Moreover, it is also associated with a significantly increased risk for congenital malformations and autosomal recessive diseases.

In the present unit, we will try to understand the concept of consanguinity and its consequences; inbreeding and outbreeding and its significance; how to calculate the inbreeding coefficient and the estimation of the magnitude of genetic load.

8.1 PARENTAL CONSANGUINITY

8.1.1 Consanguineous Marriages

The term consanguinity has originated from two Latin words, 'con' meaning common, and 'sanguineus' meaning blood, referring to a relationship between individuals related biologically by sharing common ancestors. Therefore, two individuals are said to be consanguineous if they have at least one common ancestor. In simple words, consanguineous marriage may be defined as those

types of marriages that take place between closely related individuals. We all human beings are, to some extent, consanguineous as have descended from a common ancestor. The offspring of consanguineous parents may be at increased risk of genetic disorders because of chances of inheritance of recessive allele at the same locus due to the common ancestor of the parents. The probability of inheritance of detrimental recessive genes in offspring is higher if the biological relationship between parents is significantly closer.

Based on the biological relationship, consanguineous marriages are categorised into three types which can be expressed in terms of degrees of relationship such as first-degree relationships, second-degree relationships, and third-degree relationships. Marriage of an individual with parents (mother or father), siblings, or children by sharing half of the genes is called first-degree relationship. The first degree of biological relationships such as father and daughter, or mother with son, or in between brother and sister is generally not practiced. Such relationships are considered as 'incest taboo' by most societies. In second-degree relationships, the one-quarter proportion of genes is shared between consanguineous relations such as uncle niece marriage. The third degree of consanguineous marriage is with the relations sharing a one-eighth proportion of genes such as first-cousin marriage. The first-cousin marriage could be cross-cousin marriages (FSD, MBD) or parallel cousin marriages (FBD, MSD). The third degree of relationship is followed by different degrees of relationships such as fourth degree (e.g., second-cousin marriage), fifth-degree, and so on (Figure 8.1).

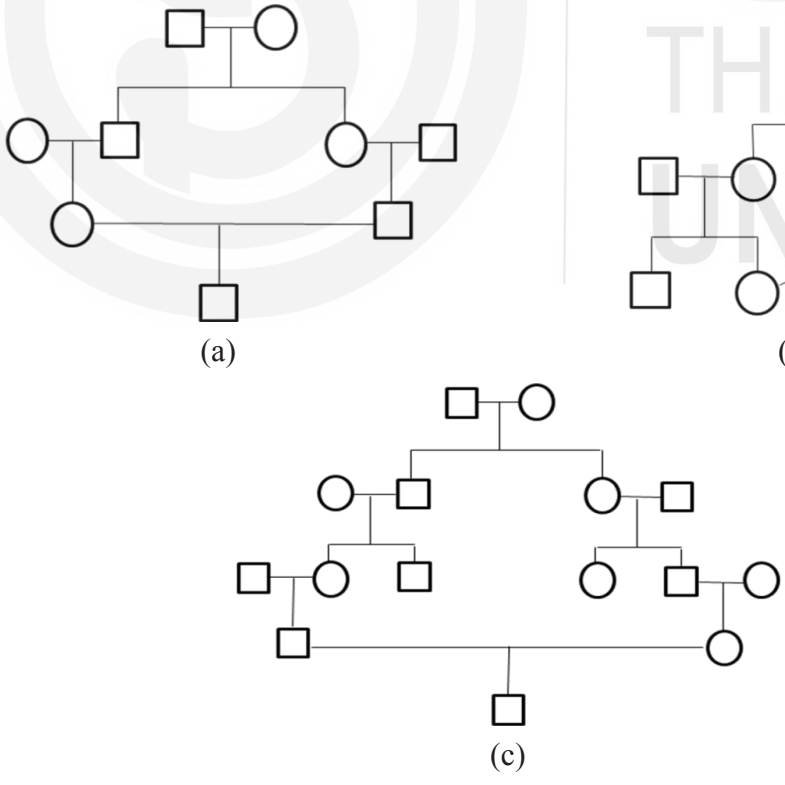

Fig. 8.1: Types of Consanguineous Marriages (a) Uncle-niece marriage; (b) First cousin marriage; (c) Second cousin marriage

Consanguineous marriage is practiced in different societies to different degrees. This type of marriage is traditionally practiced in most communities around the world. Marriage between close biological relatives is generally considered a taboo and illegal in some other communities. However, consanguineous unions are believed to be a stable form of relationship because it strengthens family relations with in-laws, and favours cultural traditions' practice and continuity. It is reported that consanguinity is associated with the levels of education, socio-economic conditions, and is commonly practiced in rural communities.

Box 8.1 : First-Cousin Marriage

In consanguineous societies, the third degree of relationship that is the first-cousin marriage is commonly practiced and widely accepted form of marriage. There are four possible types of first-cousin marriages. It could be between a person with father's brother's daughter (FBD), or with father's sister's daughter (FSD), or with mother's brother's daughter (MBD), or with mother's sister's daughter (MSD). If the marriage of a person is with FSD or MBD then that type of consanguineous marriage is known as cross-cousin marriage; whereas the marriage is with FBD or MSD are known as parallel cousin marriages (Bittles 2002).

Check Your Progress

1) What do you mean by consanguineous marriage?

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8.1.2. Prevalence of Consanguineous Marriages in Indian Population

Consanguineous Marriages by State/Union territory

The prevalence of consanguineous marriages varied across different countries; around 20% of the world population prefers this type of marriage. Consanguineous marriages are usually associated with various demographic features, such as religion, geography, ethnicity, and culture. In India, consanguineous marriage is practiced since ancient times and among most religious and ethnic groups to a greater or lesser extent (Shangvi, 1966). Various studies reported that socio-economic and cultural factors played an essential role in practicing this type of marriage in India. It is more prevalent among societies with close economies, higher rates of illiteracy, culturally isolated, geographical variation, population size, and lack of awareness (Rao, 1975; Bhinder, 2017). Approximately fourteen percent of marriages in India are reported to be consanguineous marriages, and it is more concentrated in the southern states except for Kerala (NFHS-4, 2015-16). The people of Kerala practiced strict avoidance of consanguineous marriage. The highest records of

consanguineous marriages are reported from South India. About one-third of Tamil Nadu, Lakshadweep, Andhra Pradesh, and Telangana reported being in consanguineous marriages. Of all the union territory, Pondicherry records the highest level of consanguineous marriage (uncle-niece) in a single generation with 54.9% (Puri et al., 1978). The population in Pondicherry follows a long tradition customary of the first-cousin union (mating between a man and his mother's brother's daughter) (Sastri, 1976). Marriage between relatives is believed to ensure the maintenance of their property in South India. They believe that marriage between the known and close relatives will help in strengthening the family ties socially and economically. Moreover, consanguineous marriage is also practiced to minimize the dowry in some societies. The distribution of consanguineous marriages shows wide variations across the country. The practice of consanguineous marriage seems to be preferential among the south Indian populations. However, such marriages are strictly prohibited in the northern and eastern parts of the country, with few exceptional cases. According to the National Family Health Survey (NFHS), the overall prevalence of consanguineous marriage is 16.3%. All the Southern states, namely, Andhra Pradesh, Tamil Nadu, and Karnataka (except Kerala) show the highest percentage of consanguineous marriages reaching up to 48.2%. The state of Tamil Nadu reports the highest frequency of consanguineous marriage in India, particularly from the Arcot district.

Of all the states in the southern region, Kerala had the least frequency of consanguineous marriage (11%) due to the strict avoidance of the practice of consanguineous marriage in the state. Meanwhile, in the western region, the highest frequency of consanguineous union is reported from Maharashtra with 28.3%. The consanguineous unions are infrequent in the northern, eastern, and north-eastern states since there is a restriction on practicing such marriages by the majority of the population. Most of the northern states of India record consanguineous marriages up to 10% except the state of Jammu & Kashmir, which reports 16.5%. The most minor frequency of consanguineous marriage in India is recorded from Dadra & Nagar Haveli in the western whereas Mizoram in the eastern side with only 1.4% and 1.9%, respectively. The uncle-niece marriage is the most common marriage among the different forms of consanguineous marriages practiced in India. It is found with high proportions, particularly in the coastal areas of southern India. When we look into the levels of ethnic groups, the Dravidians have the highest frequency of consanguinity while compared with other ethnic groups that are the Indo-Aryan, Indo Scythian, and Mongoloid ethnic groups.

The overall rate of consanguineous marriages showed a declining trend in India compared to NFHS-1 (16.3%) and NFHS-4 (14%). There is a significant decrease in the practice of consanguineous marriage among the eastern, north-eastern, and western regions of India according to the NFHS-1 and NFHS-4. It is also evidenced from other studies (Allendorf and Pandian, 2016; Banerjee and Roy 2002; Sharma et al. 2020). There is a declining trend in the southern region as per the NFHS-4 report; however, the area still shows a higher incidence of consanguineous marriage as compared with the rest of the states in the country. There is slight increase in frequency of consanguineous marriage from NFHS-

1 to NFHS-4 in some of the states such as Gujarat (6.6 to 9.8%), Tripura (7.1 to 12%), Nagaland (4.4 to 9.2%), Assam (3.3 to 7.5%), Odisha (9 to 12.9%), and Jammu & Kashmir (16.5 to 20.4%). The state of Mizoram reports the least frequency of consanguinity in India (Figure 8.2).

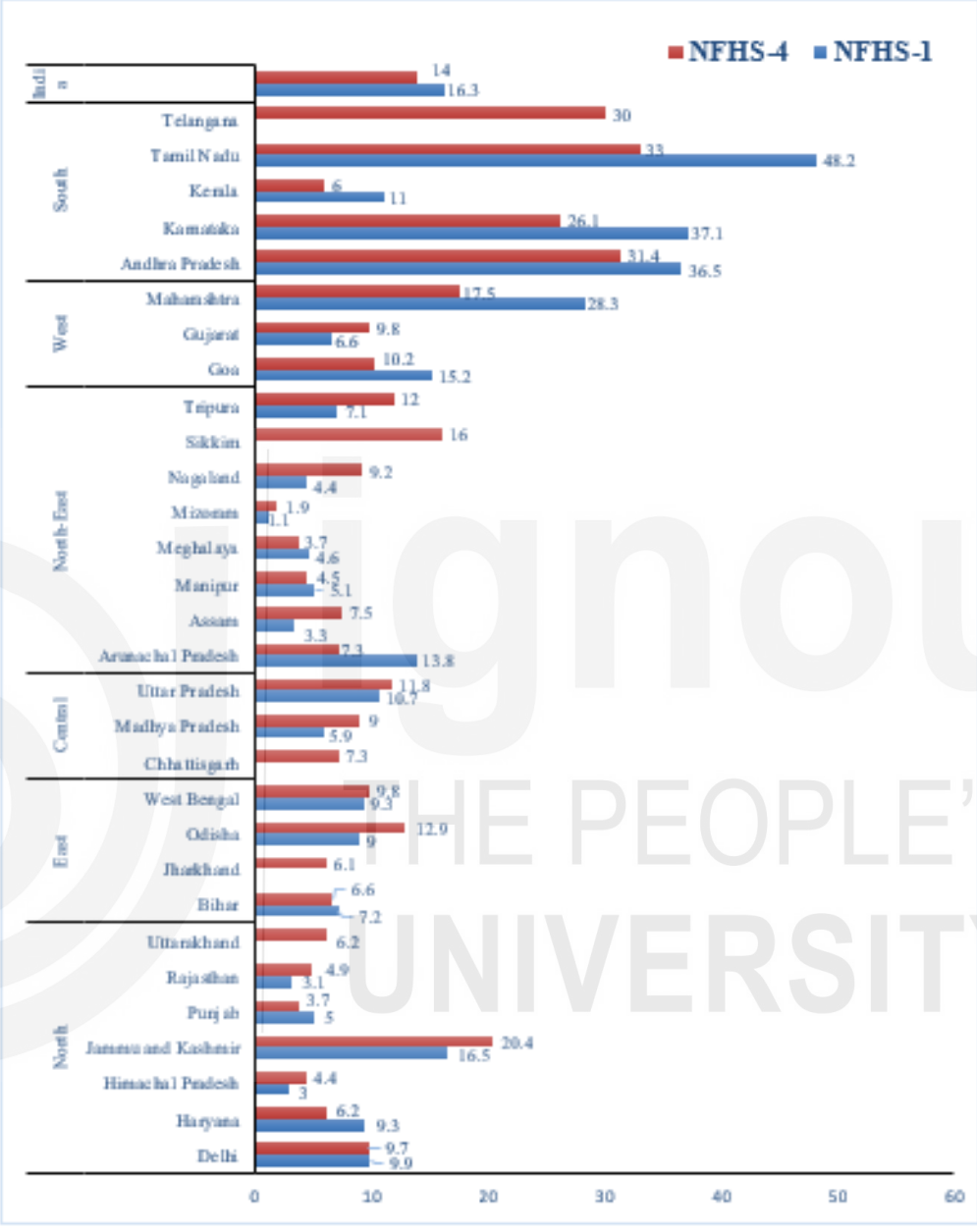

Fig. 8.2: Prevalence of Consanguineous Marriages across the Indian states
(***Source:*** adapted from NFHS-1 and NFHS-4)

Among the Muslim population of India, there is no comparable division of the north-south region as consanguineous marriage is common all over the country. The consanguineous levels are reported to be different among Muslims between specific communities. The Shia community of Lucknow in North India has a high consanguineous marriage rate of 43.4%, while the Mappillas Muslim community from Kerala reports a rate of 10% (Basu, 1975; Bittles and Hussain, 2000). A high frequency of consanguinity is reported among the Asna Ashariya, Shiekh Sunni, Dawoodi Bohra, and Muslims of Delhi and West Bengal (Basu and Roy, 1972; Basu, 1975; Rizvi and Buzarbarua, 1993). In Madhya Pradesh,

the Muslim community practices the highest rate of consanguineous marriage and the rate is comparatively low among the Hindus (Goswami, 1970).

As per the NFHS report, most religious groups practice consanguineous marriages to different degrees. Practice of consanguineous marriage by most of the religious groups showed an increase in trend from NFHS1 to NFHS-4 except the Muslims and the Buddhists. However, the Muslim community shows comparatively the highest rate of consanguineous marriage than the others. Compared to the NFHS-1 report, the higher rate of consanguineous marriage among the Hindu (13.5%), Christian (16.6%), and Jain (19.3%) are reported in the NFHS-4. However, the rate of Consanguineous marriages among Muslims (19.8%) and Buddhists (9%) shows decreased levels in the NFHS-4 report (Figure 8.3).

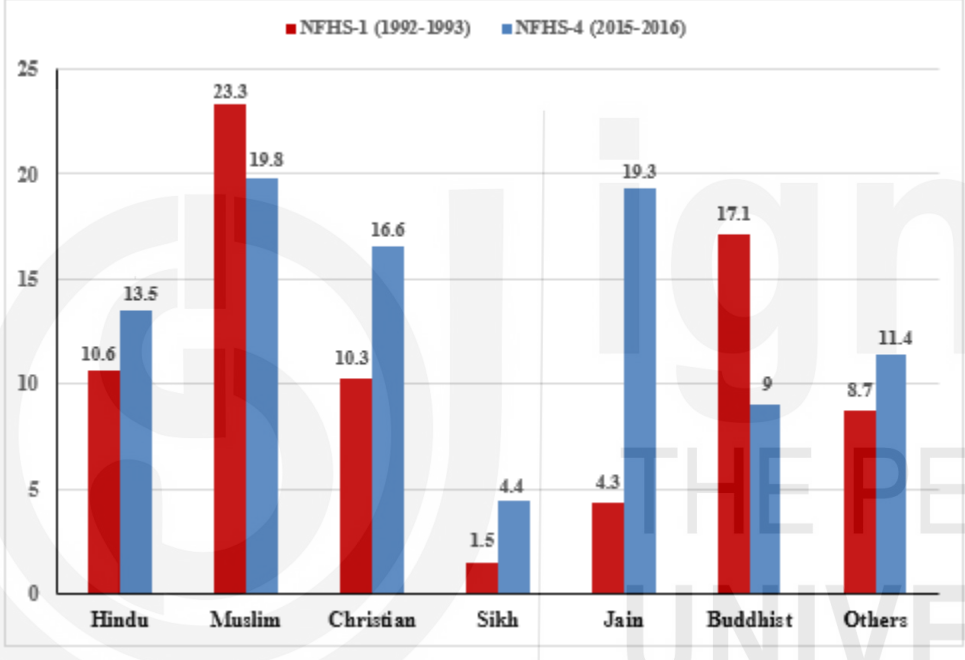

Fig. 8.3: Consanguineous marriage practices by the different religious groups in India
(***Source:*** adapted from NFHS-1 and NFHS-4)

Check Your Progress

2) Briefly explain the practice of consanguineous marriages in India.

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8.1.3 Biological Consequences of Parental Consanguinity

Consanguinity means blood relationship by descent from a common ancestor, and a consanguineous marriage is the union between biologically related individuals. It is reported that the closer the relatives, the more genetic components are shared among the individuals. The offspring of such marriages

carry double doses of recessive traits inherited from the common ancestors than less closely related individuals. Consanguineous children are more frequently homozygous for various alleles than the children of non-consanguineous parents. Thus, there is an increased chance of carrying the parent's recessive traits by the children. Children of consanguineously married parents experience several genetic disorders. Several studies have reported that parental consanguinity has been associated with adverse reproductive outcomes such as stillbirths, low birth weight, preterm delivery, abortion, infant and child mortality, congenital disabilities, cognitive impairments, increased risk for cardiovascular diseases, malformations, and other complex disorders (Badruddoza, 1994; Fareed, 2014; Bittles, 2010). Moreover, consanguineous unions lead to an increase in the expression of autosomal recessive disorders. The offspring of consanguineous parents are at increased risk for expression of recessive traits, for example, phenylketonuria (PKU), hydrocephalus, albinism, beta-thalassemia, six-fingered dwarfism, tay-sachs, alkaptonuria, cleft lip, and cystic fibrosis. There is an increase in genetic disorders, particularly among those societies where consanguineous marriages are culturally accepted (Fareed et al., 2016). These mainly result from the inheritance of recessive genes from a common ancestor. History of parental consanguinity shows a significant relationship with the illiterate, early age at marriage, and thus, offspring with such history have been reported to have congenital anomalies and low birth weight (Joseph, 2015).

8.1.4 Effects of Parental Consanguinity in India

The effect of consanguinity on the offspring has been widely studied across different populations of India. Studies have reported a significant decrease in anthropometric variables such as birth weight, length, head circumference, and gestational period of the newborn babies of consanguineously married couple than non-consanguineous union (Badaruddoza, 1998). Moreover, the lower mean birth weight is more frequent in males compared to females. Moreover, it also results in gynaecological immaturity affecting both mother and children. Such marriages also lead to a decline in reproductive and survival rates due to the expression of rare homozygous recessive traits. It is also evident that there is an increase in the number of miscarriages or stillbirths among consanguineous mothers (Bittles 2001, 2002). The stillbirth rate and malformation rates were significantly higher in uncle niece marriages than first cousin marriages and later cousin marriages reported in the South Indian population (Kulkarni, 1989). A wide range of studies in India have shown that postnatal mortality is higher among consanguineous unions' progeny due to the expression of deleterious recessive genes. Moreover, postnatal mortality and infant mortality are significantly high among children born out of consanguineous marriages than non-consanguineous union mortality (Hussain et al. 2001).

Consanguineous marriage leads their progeny to diagnose various genetic disorders. Some of the most common effects are congenital disorders including neural tube defects and congenital heart defects. There is an increased risk of cognitive impairment in consanguineous progeny (Fareed, 2014). Moreover, decreased intelligence quotient (IQ) levels and increased levels of intellectual and developmental disability (IDDs) have been reported among consanguineous

children compared to non-consanguineous children (Durkin et al., 1998). Children born to consanguineous parents also tend to have biochemical anomalies like amino acid disorders, glucose-6 phosphate deficiency, mucopolysaccharidoses, and cerebral lipidoses.

The influence of consanguinity on fertility and mortality is hard to understand. They are also dependent on several non-genetic variables, such as maternal age, birth interval, nutritional status, and so forth. There is a lack of information regarding the actual affected incidence rates for genetic disorders reported from the progeny of parental consanguinity. As most of the affected cases are reported from the country's rural populations, insufficient data failed to explain the possible effect of consanguinity on adult-onset diseases in India. In brief, consanguineous marriage has become one of the strong predictors of the onset of different genetic disorders in both infant and adult populations.

Check Your Progress

3) Briefly explain the biological consequences of inbreeding

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8.2 INBREEDING AND OUTBREEDING

Inbreeding

The term inbreeding is defined as the mode of mating or breeding involving two closely related individuals. Therefore, the process of producing offspring from consanguineous parents is called inbreeding. It is the genetic outcome of consanguineous marriage. In most societies, marriage regulates mating patterns, and consanguineous marriage leads to inbreeding. In population genetics, inbreeding refers to individuals departed from non-random mating to random mating in the population. Inbreeding increases the degree of homozygosity to the offspring, thus enabling their deleterious or recessive traits. Therefore, it results in a decline in reproduction and survival (reproductive fitness), known as inbreeding depression.

Outbreeding

outbreeding refers to the mating or breeding between unrelated individuals. As oppose to inbreeding, outbreeding increases genetic diversity, and hence the chances of having a disease or congenital abnormalities decrease in the population. In contrast to inbreeding, outbreeding increases the higher level of heterozygosity (Figure 8.4).

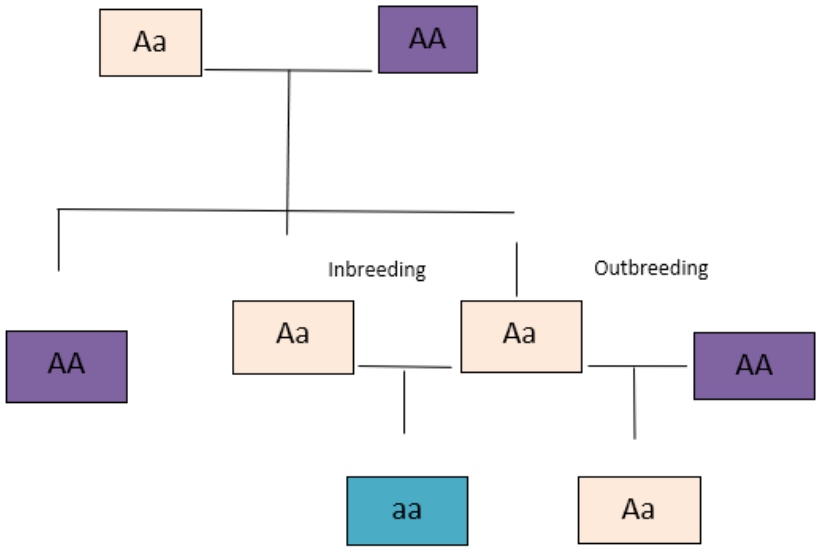

Fig. 8.4: Distribution of allele in Inbreeding and Outbreeding

(Note: A = Dominant allele; a = Recessive deleterious allele)

In a small population, closely related individuals tend to mate with one another by carrying the same deleterious allele. The offspring of such union inherit both the copies of the recessive deleterious allele; thus, the offspring suffer from the consequences of expressing the harmful allele.

With the help of inbreeding, recessive genes are expressed in offspring and thus stabilized by bringing homozygosity in the population. Those deleterious recessive genes that express as inefficient or defective individuals are weeded out from the population. Outbreeding tends to keep the recessive genes in heterozygosity. However, outbreeding decreases constancy by increasing heterozygosity for different individuals in contrast to inbreeding. Outbreeding tends to keep the recessive genes in heterozygous conditions, and they rarely get a chance to come together and express themselves. Inbreeding tends to establish pure lines in a population if continued over a long period, whereas significant variations for natural selection are provided by outbreeding due to heterozygosity.

8.3 INBREEDING COEFFICIENT (F RATIO)

In general, inbreeding is defined when both parents have ancestors in common. The level of inbreeding depends on how closely related these parents are across their ancestral lines. It is measured or quantified by using standardized mathematical formulae. Sewall Wright (1922) introduced the two primary measures used in genetic relationships, such as 'inbreeding coefficient (F)' and 'coefficient of relationship (r)' for measuring the degree of inbreeding. The coefficient of inbreeding (F) is defined as the proportion of genes at which an individual of consanguineous matting is homozygous by descent. The relationship coefficient (r) measures the degree of consanguinity (or biological affinity) between two individuals. As each parent passed on 50% of its genes to their offspring, there is 0.5 probability that a particular allele is passed on. Thus, the coefficient of relationship 'r' is calculated by using the following formula:

$$r = \{(0.5)^n\}$$

Where n is the number of steps apart between two individuals via any common ancestor. For example, the coefficient of relationship (r) of a parent and child with one step apart is calculated as follows:

$$r = \{(0.5)^1\} = 0.5;$$

If the coefficient of relationship (r) is calculated between grandparent and grandchild two steps apart is calculated as follows:

$$r = \{(0.5)^2\} = 0.25.$$

The coefficient of inbreeding (F) is defined as the probability of two homologous alleles of an individual being identical by descent and the genetic correlation between uniting gametes. Generally, Fi for an individual is calculated by using the formula:

$$F_i = \sum(1/2)^n (1+F_A)$$

Where $\sum$ is the sum of all paths in the relationships that goes through common ancestor; n is the number of individuals along the path connecting the two parental gametes through a common ancestor; FA is the inbreeding coefficient of the common ancestors.

In general, the inbreeding coefficient (F) is half of the coefficient of relationship (r). For example, in the incestuous form of relationships such as between father-daughter, mother-son, or brother-sister, the coefficient of relationship (r) shares half of their genetic materials (r=0.5). Therefore, any siblings of such incestuous parents would be homozygous at 1/4 of gene loci (F=0.25). In the case of uncle-niece marriage, both the parents share 1/4 of their genetic components, and hence, the coefficient of relationship (r) is 0.25. On the other hand, inbreeding in their progeny will be half of the coefficient of relationship (r), i.e., F=0.125. Relationship of individuals measured by the coefficient of relationship and inbreeding coefficient in different levels of degrees are shown in Table 8.1.

Table 8.1: Relationship of individuals in terms of Consanguinity

Biological Relationship	Genetic Relationship	Coefficient of relationship (r)	Coefficient of inbreeding (F)
Incest	1st Degree	0.5	0.25
Uncle-niece	2nd Degree	0.25	0.125
First cousin	3rd Degree	0.125	0.0625
First cousin once removed	4th Degree	0.0625	0.0313
Second cousin	5th Degree	0.0313	0.0156
Second cousin once removed	6th Degree	0.0156	0.0078
Third cousin	7th Degree	0.0078	0.0039

(***Source:*** Adapted from Vogel and Motulsky's Human Genetics, 2010)

Check Your Progress

4) How are degrees of genetic relationships measured?

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8.4 GENETIC LOAD

Genetic load is defined as the measure of extent to which average fitness of a population is decreased. In other words, genetic load measures the rate of damage caused by the presence of certain deleterious genes in the population (Muller, 1950). According to Crow (1958), the genetic load of a population refers to the relative decrease in population fitness if all individuals in that population had the optimum genotype. The definition of genetic load may be redefined as the decreased average fitness proportion compared to what it would be if the mutations were absent. It also measures the total number of disadvantageous genes present in a particular population. It may be caused by different factors based on the types of load that decrease the population's average fitness.

There are different forms of genetic load such as mutational load, segregational load, recombination load, drift load, and migration load. The mutational load is caused by the decrease of the mean fitness of the population in the presence of a deleterious mutation in the population. The segregational load is caused by decrease in fitness in a population due to segregation of homozygotes over the heterozygote and recombination load is caused by favourable gene through recombination. The drift load is the fixation of deleterious allele frequency due to small population size and migration load is caused by adaptation to the new environment by immigrants.

The deleterious or disadvantaged gene reduces the fitness level of the population by causing morbidity, mortality, and sterility. It could happen in two ways, such as in heterozygote condition, when the deleterious gene is dominant; and in the homozygous condition in which the deleterious gene is recessive. To sum up, genetic load refers to genetic variation that decreases the average fitness of a population compared with a theoretical model that a slow inbreeding rate allows selection to purge the deleterious alleles from the populations.

8.5 SUMMARY

Marriage is considered one of the most complex relationships in human society. In India, marriage shows a considerable variation with great ethnic diversity and religious variation. Marriage between closely related individuals 'consanguineous marriage' is a traditional and most common marriage performed in most societies since ancient times. Such marriage is deeply rooted in the beliefs and customs of society. The prevalence of consanguineous marriage is

observed very frequently in some communities, particularly in South India. The least prevalence of consanguineous marriages are reported from the country's north-eastern region. Children of consanguineous union parents are at a greater risk of being homozygous for a harmful gene and subsequently from the expression of recessive traits, and autosomal recessive genetic disorders tend to be diagnosed. The adverse consequences of consanguineous marriages recorded among woman with early age at marriage and children often show congenital anomalies and low birth weight. Assessing the biological implications of consanguinity in terms of morbidity and mortality is still facing many problems in our country.

The control and management of the consequences of consanguineous marriage is a challenging mission. People are not fully aware of health consequences of consanguineous marriage, and appropriate diagnostic facilities are not available in most of the states. Moreover, people could not afford the expenses for diagnosis even if they have the facilities. Therefore, premarital counselling is needed for couples with a family history of anomalies to avoid consanguinity. Moreover, preconception genetic counselling should also be done for consanguineous parents to avoid genetic disorders, facilitating informed family planning. The rate of consanguineous marriage in India could be decreased to some extent by improving educational status as well as improving socio-economic status. It could also be achieved through making awareness at community levels and also developing community education programmes.

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8.7 ANSWERS TO CHECK YOUR PROGRESS

1) Consanguineous marriage is defined as that marriage which takes place between individuals related biologically, i.e., sharing of common ancestors. Thus, the chances of inheritance of a mutant allele present at the same locus are significantly high as both parents have a common ancestor. The progeny of such marriage is homozygous for a deleterious gene and later tend to affected by autosomal recessive genetic disorders. Thus, parental consanguinity is reported to be associated with various unfavourable biological consequences such as stillbirths, low birth weight, preterm delivery, abortion, infant and child mortality, and congenital disabilities.

2) The highest rates of practicing consanguineous marriages are mainly concentrated in South India. There has been only a modest reduction in the frequency of consanguineous marriages over the years when comparing the NFHS-1 and NFHS-4 reports in southern states. This further shows that such marriages form an integral part of social and cultural life across the country. According to the NFHS-4, Tamil Nadu and Mizoram record the highest and lowest level of consanguineous marriage, 33% and 1.9% in India.

3) In Inbreeding, the deleterious mutants or recessive traits carried by the population are fully expressed due to an increase in homozygosity, while in Outbreeding, the deleterious mutants carried by the population are only partly expressed in each generation, is concealed mainly by heterozygosis with more favourable alleles.

4) Mathematically the degree of genetic relationship is quantified by using standardized formulae. In general, the degree of genetic relationships can be measured by two basic measures, such as the coefficient of relationship (r) and coefficient of inbreeding (F). The coefficient of inbreeding is the probability that two alleles at a randomly chosen locus are identical by descent. However, the coefficient of relationship (r) measures the degree of biological relationship between two individuals who share a common ancestor.

UNIT 9 POPULATION AND DISEASE ASSOCIATION STUDIES

Contents

Learning Objectives

After reading this unit, you will be able to:

- Elucidate the definitions of blood group, blood group system, definition of antigens and their expression in other tissues;
- Explain the reasons for inherited variation and distribution in RBC antigens and present status;
- Understand about antibodies, classification and nomenclature of antigens and their applications; and
- Expose to the knowledge on the association of blood groups and diseases.

9.0 INTRODUCTION

A group of red cells (RBC) or erythrocyte surface antigens in an individual is called 'blood group', whereas blood group systems are defined as the systems of one or more antigens controlled by a single gene or closely related genes. The specific sites located on proteins, glycolipids or glycoprotein components of RBC membrane and interacting with immune system are termed as antigens (ISBTweb.org). Antigens are considered as secondary gene products whereas the glycosyltransferase enzymes to which antigens are attached as primary gene products. It has been observed that evolution of antigens occurred earlier in ecto and endodermal tissues rather than RBC and hematopoietic cells and therefore also referred as histo-blood group antigens. Decreased level of A and B antigens were reported in patients with leukaemia and a plastic anaemia (Ewald and Sumner, 2016). Besides RBC, blood group antigens are also found on leukocytes, platelets, plasma proteins, and body fluids such as amniotic fluid,

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urine, saliva, gastric secretions, sweat, breast milk and seminal fluid, neurons, epithelial and endothelial cells (Ewald and Sumner, 2016). Each blood group system is genetically unique (ISBT web.org). Those who secrete blood group antigens in their body fluids are called secretors and those unable to secrete are termed as non-secretors.

Blood Group Systems

The blood group systems have been studied by Anthropologists to understand population variation and in racial classification. A number of blood group systems were discovered. American biologist, Karl Landsteiner discovered the A, B, O of ABO blood group system in 1901. A year later, in 1902, de Castello and Steini discovered the AB blood group. Rh blood group was discovered by Karl Landsteiner and Weiner in 1940. Before that in 1927, Landsteiner and Levine discovered the MNS blood groups. M+ and N+ RBCs are found among 75% of the population and M+N+ RBCs are the most common genotype found in 50% of population. In the same year they also discovered another blood group P. Later, other blood groups like the ABH (1930), Lutheran (1946), Kell (1946), Duffy (1950), Kidd (1951), and Diego (1954) were discovered by Lehrs, Callender & Race, Coombs et al, Cutbush et al, Allen et al, Levine et al, respectively.

9.1 INHERITED VARIATION AND DISTRIBUTION IN RBC ANTIGENS AND PRESENT STATUS

Blood group of a person is determined by the presence or absence of inherited variation of RBC surface antigens. To identify any blood group antigen the governing genetic variation has to be identified, sequenced and established that it is influencing a particular phenotype. As per the International Society of Blood Transfusion (ISBT) till June, 2021, 43 blood group systems containing 345 RBC antigens and governed by 48 genes are identified (ISBT web.org). The frequency of O blood group is higher in world population followed by A, B and AB and the incidence of B is higher in India (Dean, 2005). Founder effect and in areas endemic to malaria, selection is responsible for the difference in the frequency of blood groups in world populations (Anstee, 2010). The blood group system, symbol, gene names, number of antigens and chromosome location are presented in Table 9.1. The details of antigens of blood groups are given table 9.2. The number of antigens range from one to 56. The genes of these blood groups located on chromosomes 1, 2, 3, 4, 6, 7, 9,11, 12,13,15,16, 17,18,19,20, 22 autosomes and X chromosome. The Rh blood group system is more complex due to the presence of 56 antigens and ABO system is polymorphic due to the presence of >20 sub-groups (Ewald and Sumner, 2016).

Check Your Progress

1) Till now how many blood groups are identified and inform the number of antigens and genes involved in them.

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9.1.1 Antibodies

The presence of the antigens is identified by antibodies or commercially available antisera. Antibodies are proteins produced by B lymphocytes when exposed to antigens. On exposure to antigens, B lymphocytes are transformed into plasma cells and secrete antibodies. Antibodies are of five types i.e. IgG, IgM, IgD, IgE and IgA. Blood Antibodies are mostly IgG, IgM and IgD types. They are formed naturally due to their exposure to the antigens present in the environment (natural antibodies) (ABO blood group system) or RBC from other individuals (immune antibodies). Antibodies are reported to be inherited. Antibodies when exposed to the foreign antigens have been shown to reject transplanted organs in recipients and induce abortions (Ewald and Sumner, 2016).

9.1.2 Nomenclature of RBC Antigens

Blood group antigen names are given alphabetically and credit of discovery is given to the researchers who produce antibody against the blood group antigen. Each RBC antigen is given a number (6 digit number, first three numbers indicate blood group (Example ABO-001 and Rh-004) and last three digits indicate the order of the discovery of antigen. For instance, in ABO blood group, number 001 is given to A antigen because it is first discovered and 002 is given to antigen B) and assigned to blood group system or collection (antigen not meeting the criteria of blood group but linked genetically or biochemically to a particular blood group) or series (antigen not meeting either criteria of blood group or collections) (Dean, 2005). Since 1980 onwards the task of devising standards for blood group technology is shouldered by International Society of Blood Transfusion working party on Terminology for Red Cell surface antigens.

9.1.3 Classification of RBC Antigens

Blood group antigens are classified into different groups based on functions: ABO, H, Lewis, I, PIPK, Globoside, Forssamine (Transferases); Kell, Yt, Dombrock (Enzyme); Chido/Rodgers, Cromer, Knops (Complement regulations), Gerbich, MNS (Structural), Rh, RHAG, Diego, Colton, Gill, Kidd, Kx, Lan (Transport/ Channel), Indian, Lutheran, Landsteiner-Weiner, John Milton Hagen, Xg, Ok (Adhesion) and Duffy, Sciana and Raph (Receptor) (Ewald and Sumner, 2016). ISBT also classified RBC surface antigens based on functions in to different categories such as component of glycocalyx (MNS); structural component

(Diego and Gerbich); enzymes (Yt, Kell and Dombrock); receptor and adhesion molecules (Duffy and Luthern); membrane transporters (Diego and Kidd) and complement regulatory glycoproteins (Cromer and Knops) (ISBTweb.org).

9.1.4 Applications of RBC Antigens

Among blood groups identified in human beings, the ABO blood was first discovered in 1900 by Karl Land Steiner. First genetic polymorphism studies conducted using ABO blood group system and first human evolutionary tree was constructed using allele frequency data of ABO system. For management of clinical conditions, typing of blood group is done using commercial antisera. The typing involves antigen and antibody interaction. The antibody presents in the antisera bind with antigen present on the surface of RBC and cause agglutination of RBC. The blood group typing is done using various methods such as slide test, tube test, microplate, column/gel centrifugation and molecular imprinting. In clinical applications particular component of blood is used in transfusion such as plasma, platelets, packed RBC and immunoglobulins after cross matching particularly in case of ABO system. Apart from clinical applications, blood group data is also used for solving paternity disputes, identification of suspects at the crime scenes and finding evolutionary relationships between populations.

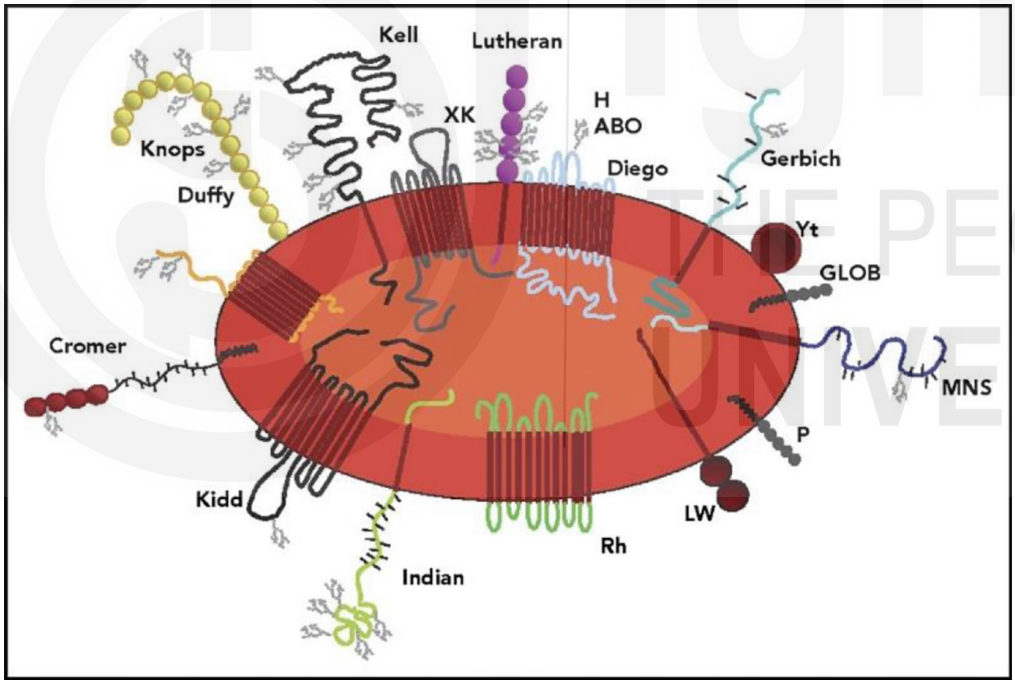

Fig. 9.1 : RBC membrane with representative blood group antigens
(***Source:*** https://onlinelibrary.wiley.com/doi/full/10.1111/voxs.12593

9.2 BLOOD GROUPS AND DISEASES

Blood group antigens play an important role in the causation or prevention of diseases. Presence and absence of antigens cause changes in the RBC membrane and functions associated with these membranes make the carriers of blood groups susceptible to diseases. Non-secretors are at increased risk of infection due to the binding of organisms to the polysaccharide on RBC surface

whereas in case of secretors the presence of antigens in body fluids prevent their binding (Abegaz, 2021). Difference in the expression of RBC antigens has been shown to either increase or decrease the susceptibility of individual to the infections (Fan et al.2020). These diseases are mostly found to be associated with phenotypes of ABO blood group system (Ewald and Sumner, 2016). The blood groups which are reported to be associated with different disease are presented in Table 9.3. From this table till now the involvement of ABO, Rh, Lewis, Duffy, MN, Knops, RHAG and Raph blood groups in causation of communicable and non-communicable diseases were reported. The cause and effect relationship between blood groups and diseases needs to be proved by validated mechanisms.

Check Your Progress

2) Name the blood groups which are associated with malaria?

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Table 9.1: Blood Group Systems in Humans

S. No.	Blood Group System	Symbol	Genes (s)	Antigens (n)	Chromosome location
1	ABO	ABO	*ABO*	4	9q34.2
2	Rh	RH	*RHD, RHCE*	56	1p36.11
3	MNS	MNS	*GYPA, GYPB*	50	4q31.21
4	Duffy	FY	*ACKR1*	5	1q21-q22
5	Kell	KEL	*KEL*	36	7q33
6	Lutheran	LU	*BCAM*	*27*	*19q13.2*
7	P1PK	P1PK	A4GALT	3	22q13.2
8	Lewis	LE	*FUT3*	6	19p13.3
9	Kidd	JK	*SLC14A1*	3	18q11-q12
10	Diego	DI	*SLC4A1*	23	17q21.31
11	Yt	YT	*ACHE*	5	7q22
12	Xg	XG	*XG,CD99*	2	Xp22.32
13	Scianna	SC	*ERMAP*	9	1p34.2
14	Dombrock	DO	*ART4*	10	12p13-p12
15	Colton	CO	*AQP1*	4	7p14
16	Landsteiner-Wiener	LW	*ICAM4*	3	19p13.2
17	Chido/Rodgers	CH/RG	*C4A, C4B*	9	6p21.3
18	H	H	*FUT1; FUT2*	1	19q13.33
19	Kx	XK	*XK*	1	Xp21.1
20	Gerbich	GE	*GYPC*	13	2q14-q21
21	Cromer	CROM	*CD55*	20	1q32

22	Knops	KN	*CR1*	12	1q32.2
23	Indian	IN	*CD44*	6	11p13
24	Ok	OK	*BSG*	3	19p13.3
25	Raph	RAPH	*CD151*	1	11p15.5
26	John Milton Hagen	JMH	*SEMA7A*	8	15q22.3-q23
27	I	I	*GCNT2*	1	6p24.2
28	Globoside	GLOB	*B3GALNT1*	2	3q25
29	Gill	GIL	*AQP3*	1	9p13
30	Rh-associated glycoprotein	RHAG	*RHAG*	4	6p12.3
31	FORS	FORS	*GBGT1*	1	9q34.13-q34.3
32	JR	JR	*ABCG2*	1	4q22.1
33	LAN	LAN	*ABCB6*	1	2q36
34	Vel	VEL	*SM1M1*	1	1p36.32
35	CD59	CD59	*CD59*	1	11p13
36	Augustihe	AUG	*SLC29A1*	4	6p221.1
37	Kanno	KANNO	*PRNP*	1	20p13
38	SID	SID	*B4GALNT2*	1	17q21.32
39	CTL2	CTL2	*SLC44A2*	2	19p13.2
40	PEL	PEL	*ABCC4*	1	13q32.1
41	MAM	MAM	*EMP3*	1	19q13.33
42	EMM	EMM	*PIGG*	1	4p16.3
43	ABCC1	ABCC1	*ABCC1*	1	16p13.11

Source: **www. isbtweb.org**

Table 9.2: Antigens of Blood Groups in Humans

S.No	Blood group system	Antigens
1	ABO	A, B, A B, A1
2	Rh	D, C,E,c,e,f,Ce,Cw,Cx,V,Ew,G,Hro,Hr,hrs,VS,CG,CE,Dw, c-like, cE, hrH, Rh29, Goa, hrB, Rh32,Rh33,HrB,Rh35,Bea, Evans, Rh39, Tar, Rh41, Rh42, Crawford, Nou, Riv, Sec, Dav, JAL, STEM, FPTT, MAR, BARC, JAHK, DAK, LOCR, CENR, CEST, CELO, CEAG, PARG,CEVF,CEWA,CETW
3	MNS	M, N, S,s, U, He, Mia, Mc, Vw, Mu, Mg, Vr, Me, Mta, Sta, Ria, CIa, Nya, Hut, Hil, Mv, Far, SD, Mit, Dantu, Hop, Nob, Ena, ENKT, 'N' Or,DANE, TSEN,MINY, MUT,SAT, ERIK,OSa, ENEP,ENEH, HAG, ENAV, MARS, ENDA, ENEV,MNTD,SARA,KIPP, JENU,SUMI,
4	Duffy	FYa, FYb,FY3,FY5,FY6
5	Kell	K,k,Kpa,Kpb,Ku,Jsa,Jsb,UIa,K11,K12,K13,K14,K16 ,K17,K18,K19, Km,Kpc,K22,K23,K24,VLNA, TOU, RAZ,VONG,KALT,LTIM, KYO, KUCI, KANT, KASH, KELP,KETI,KHUL,KYOR,KEAL
6	Lutheran	Lua,Lub,Lu3,Lu4,Lu5,Lu6,Lu7,Lu8,Lu9,Lu11,Lu12,Lu13, Lu14,Lu16,Lu17,Aua,Aub,Lu20, Lu21,LURC,LUIT,LUGA, LUAC, LUBI, LUYA,LUNU, LURA

7	P1PK	P1, Pk, NOR
8	Lewis	Lea,Leb, Leab,LebH,ALeb, BLeb
9	Kidd	Jka, Jkb,Jk3
10	Diego	Dia,Dib,Wra,Wrb,Wda,Rba,WARR,ELO,Wu,BPa,Moa,Hga, Vga,Swa,BOW,NFLD,Jna,KREP,Tra,Fra,SW1,DISK,DIST
11	Yt	Yta,Ytb,YTEG,YTLI,YTOT
12	Xg	Xga, CD99
13	Scianna	Sc1,,Sc2,Sc3,Rd,STAR,SCER,SCAN,SCAR,SCAC
14	Dombrock	Doa,Dob,Gya,Hy,Joa,DOYA,DOMR,DOLG,DOLC, DODE
15	Colton	Coa, Cob, Co3, Co4
16	Landsteiner-Wiener	Lwa,LWab,LWb
17	Chido/ Rodgers	Ch1, Ch2, Ch3, Ch4, Ch5, Ch6, WH, Rg1, Rg2
18	H	H
19	Kx	Kx
20	Gerbich	Ge2,Ge3,Ge4, Wb,Lsa,Ana,Dha,GEIS,GEPL,GEAT,GETI,G ECT,GEAR
21	Cromer	Cra,Tca,Tcb,Tcc,Dra,Esa,IFC,WESa,WESb, UMC,GUTI, SERF, ZENA, CROV, CRAM, CROZ,CRUE,CRAG,CROK,CORS
22	Knops	Kna,Knb,McCa,SI1,YKa,McCb,SI2,SI3,KCAM,KDAS,DA CY,YCAD
23	Indian	Ina,Inb,INFI,INJA,INRA,INSL
24	Ok	Oka, OKGV,OKVM
25	Raph	MER2
26	John Milton Hagen	JMH,JMHK,JMHL, JMHG, JMHM,JMHQ, JMHN,JMHA
27	I	I
28	Globoside	P, PX2
29	Gill	Gill
30	Rh-associated glycoprotein	Duclos, OIa, DSLK,Kg
31	FORS	FORS1
32	JR	Jra
33	LAN	Lan
34	Vel	Vel
35	CD59	CD59.1
36	Augustihe	AUG1, Ata, ATML, ATAM
37	Kanno	KNNO1
38	SID	Sda
39	CTL2	VER, RIF
40	PEL	PEL
41	MAM	MAM
42	EMM	Emm
43	ABCC1	WLF

Source: **www. isbtweb.org**

Table 9.3: Association of Blood Groups and Diseases

Communicable Diseases	
Blood Group	**Disease (s)**
A	Infections of Hepatitis B virus (Wang et al., 2012a), HIV, (Mohammadali and Pourfathollah, 2014), *Helicobacter pylori* (Wang et al., 2012b),COVID-19 (Alkout et al., 2020), H1N1 (Lebiush et al.,1981), *E. Coli* (Zhou and Welsby, 2014), Small Pox and *Pseudomonas Aerugionosa* (Ewald and Sumner, 2016)
B	Infections of Falciparum malaria (Panda et al., 2011), Gonorrhoea, *Streptococus pneumoniae* (Garatty , 2005), H3N2, a Type A influenza (Mackenzie and Fimmel,1978), Hepatitis-C (Erhabor et al., 2015), Tuberculosis, *E. Coli*, S. Pneumoniae , and Salmonella (Ewald and Sumner, 2016)
O	Infections of *E. coli* (Blackwell et al.2002), hepatitis-B, C (Jiang et al.2020; Aljooni et al., 2012) and Noro virus (Hutson et al.,2002; Liao et al., 2020), Plague, Cholera, Tuberculosis, Mumps (Garatty, 2005), SARS (Cheng et al., 2005) and Dengue virus (Harshan et al.,2020)
AB	Infections of HIV-2 (Abdulazeez et al., 2008) influenza A & and B (Aho et al.,1980; Naïkhin et al., 1989), Dengue hemorrhagic fever (Muruganathan et al., 2018), Smallpox, *E. Coli* and salmonella (Ewald and Sumner, 2016)
Rh	Infections of Hepatitis-B virus infection (Mohammadali and Pourfathollah, 2014)
M	Yaws (Garatty , 2005)
Knops	*Plasmodium falciparum* Malaria (Ewald and Sumner, 2016)
Duffy	*Plasmodium vivax Malaria* (Ewald and Sumner, 2016)
Lewis	Cholera (Ewald and Sumner, 2016)
Non-communicable Diseases	
A	Myocardial infarction, venous thrombo-embolism, ischemic stroke (Wiggins et al., 2009), Pancreatic cancer (Wang et al., 2012a), gastric cancer (Wang et al.2012b), nasopharyngeal cancer (Sheng et al., 2013), hyperlipidaemia, heart failure and atherosclerosis (Groot et al., 2020),diabetes, heart disease(Zhou and Welsby, 2014), Breast, Basal Cell, Non-small cell lung, Gastric, Skin, Nasopharyngeal, Hepato cellular, Bladder, Lung, Gall bladder, Ovarian, Oral, Cervical, Oesophageal, Oral and salivary gland cancers (Amjadi et al.,2015)
B	Ovarian cancer (Gates et al., 2011), Type 2 diabetes (Qureshi and Bhatti, 2003), exocrine pancreatic cancer (Ewald and Sumner, 2016) hypertension (El-Sayed and Amin, 2015); Hodgkin's lymphoma (Ewald and Sumner, 2016), myocardial infarction (Groot et al., 2020), Gastrointestinal tract, Gall bladder, central nervous system, oral and non-squamous cell, Laryngeal, Genitourinary, Liver, Ovarian, Breast, Pancreatic and Cardiac cancers (Amjadi et al.,2015), Hodgkin's lymphoma (Ewald and Sumner, 2016)
AB	Myocardial infarction, venous thrombo-embolism (Wiggins et al., 2009); Preeclampsia (Hiltunen et al.2009), nasopharyngeal cancer (Sheng et al., 2013), cognitive impairment (Ewald and Sumner,2016), Lung, Nasopharyngeal and Gastrointestinal carcinoma (Amjadi et al., 2015)

O	Meta static tumour (Ewald and Sumner, 2016) neuroendocrine tumours (Weisbroad et al., 2013), peptic ulcer (Ewald and Sumner, 2016; Garatty, 2005), Malignant Melanoma (Amjadi et al., 2015), Multiple Endocrine Neoplasia Type1, Von Hippel-Lindau and Neuroendocrine, acute lymphoblastic leukaemia, Primary and Secondary Non-Hodgkins central nervous system lymphoma (Ewald and Sumner, 2016)
Rh	Haemolytic disease of new born (Anstee, 2010), Chronic and auto immune haemolytic anaemia (Ewald and Sumner, 2016)
RHAG	Chronic and auto immune haemolytic anaemia (Ewald and Sumner, 2016)
Lewis	Peptic ulcer (Garatty, 2005)
Raph	Renal disease (Garatty, 2005)

9.3 SUMMARY

The specific sites located on proteins, glycolipids or glycoprotein components of RBC membrane and interacting with immune system are termed as antigens. Besides RBC, blood group antigens are also found on leukocytes, platelets, plasma proteins, and body fluids such as amniotic fluid, urine, saliva, gastric secretions, sweat, breast milk and seminal fluid, neurons, epithelial and endothelial cells. As per the International Society of Blood Transfusion (ISBT) till June, 2021, 43 blood group systems containing 345 RBC antigens and governed by 48 genes are identified. Founder effect and natural selection (in areas endemic to malaria and other communicable and non-communicable diseases) are responsible for the difference in the frequency of various blood groups in world populations. Antibodies are proteins produced by B lymphocytes when exposed to antigens. They are formed naturally due to their exposure to the antigens present in the environment (natural antibodies) (ABO blood group system) or RBC from other individuals (immune antibodies). Blood group antigen names are given alphabetically and credit of discovery is given to the researchers who produce antibody against the blood group antigen. Based on the functions of antigens they are classified into different categories. In clinical, forensic, medico-legal and evolutionary studies data of blood groups is useful. Till now the involvement of ABO, Rh, Lewis, Duffy, MN, Knops, RHAG and Raph blood groups in the causation of communicable and non-communicable diseases is only known.

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9.5 ANSWERS TO CHECK YOUR PROGRESS

1) As per the International Society of Blood Transfusion (ISBT) till June, 2021, 43 blood group systems containing 345 RBC antigens and governed by 48 genes are identified.

2) Blood groups B, Duffy and Knops are associated with *Plasmodium falciparum* ad Vivax Malaria.

UNIT 10 COMPARATIVE BIOLOGY

Contents

Learning Objectives

After reading this unit, you will be able to:

- Understand the position of apes and humans in the animal kingdom;
- Differentiate between hylobatidae, pongidae, and Hominidae;
- Learn the characteristic features of Chimpanzees, Bonobos, Gorilla, Orangutan, and gibbon; and
- Explore the morphological and anatomical differences between apes and humans.

10.0 INTRODUCTION

Biologically, humans belong to animal kingdom genus Homo and sapiens species. In taxonomic classification, apes and humans, both extinct and extant are grouped in the order primate of the Animal Kingdom. It was Carolus Linnaeus who introduced the term primate in the year 1758 (Linnaeus, 1758). Primates are unguiculate, claviculate placental mammals, with orbits surrounded by

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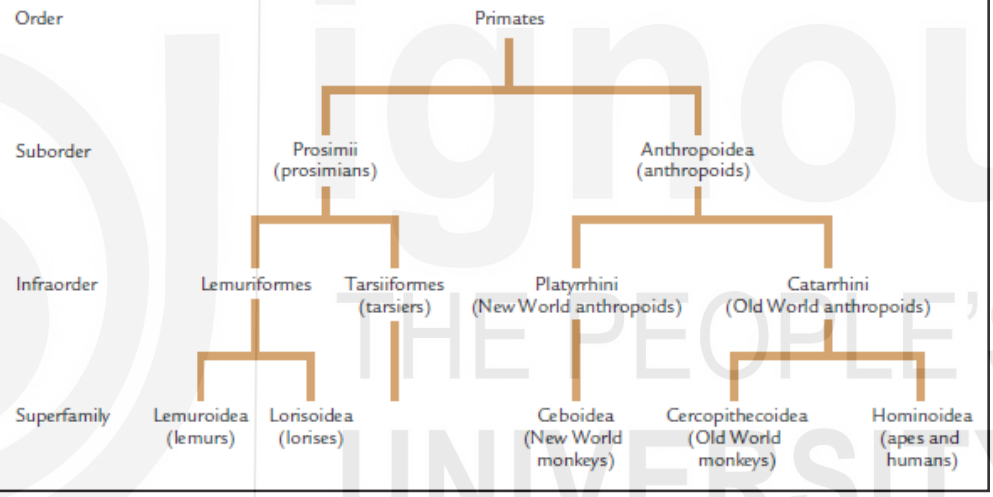
bone; three kinds of teeth, at least one time of life; brain always with a posterior lobe and calcarine fissure; the innermost digit of at least one pair of extremities opposable; hallux with a flat nail or none; a well-developed caecum; pendulous penis; testes scrotal; always two pectoral mammae (Mivart, 1873). Non-human living primates are found on almost all the world's significant landmasses except North America, North of Mexico City. Based on their habits, primates can be classified into three groups. They are nocturnal primates that carry on their activities at night, diurnal primates that are active during daytime hours, and crepuscular primates which are most active in the late afternoon, early evening and morning. Primates are more adapted to the arboreal mode of life. Their back and limb bones and muscles have been modified for different kinds of locomotion on the trees. All the primates possess prehensile hands and feet. Primates are generally known for their sociability.

The Primate Classification of Relethford (2002) is given in Box 10.1.

Box 10.1:



10.1 THE HOMINOIDEA

The apes and humans, both extinct and extant are classified in the superfamily Hominoidea.

A detailed discussion of the super family Hominoidea is given below:

The superfamily Hominoidea consists of two families, namely Pongidae and Hominidae. Pongidae is further divided into two sub families – Ponginae and Hylobatinae. Ponginae includes all the three genera, namely, Pongo with one species (P. pygmaeus), Pan with two species (P. gorilla and P troglodytes or Chimpanzee) and Gorilla with one species. Hylobatinae has only one genus called Hylobates with three species (H. lar, H. moloch and H. Syndactylus or Gibbons and Siamangs). They have the diploid chromosome of 24 pairs (symbolised 2n) or 48 chromosomes (versus 2n of 23 pairs or 46 chromosomes in Homo sapiens), the habit of nest building, and many anatomical, physiological, and biomolecular features. Furthermore, the family Hominidae consists of all the humans both extant and extinct.

10.1.1 The Pongidae

The family Pongidae is divided into two sub families: Ponginae and Hylobatinae.

The Ponginae

This group includes three giant apes – Pongo (Orangutan), Pan (Chimpanzee), and Gorilla. The term 'ape' refers to all the Ponginae, while 'great apes' refers to Chimpanzee and Gorilla. Pongids are man's closest relatives.

Pongo (Orangutan)

They are the most enigmatic of all the hominoid primates. Compared to other apes, they are primarily solitary. They are usually classified as two species, found on the Indonesian islands of Bornea (Pongo pygmaeus) and Sumatra (Pongo abelii). Bornean males have enormous flanges of flesh around the face and long, pendulous throat sacs (probably used to produce their long resonating calls). Genomic data indicate that Bornean and Sumatran Orangutans are 99.7% similar and separated about 400,000 years ago (Locke et al., 2011). It also indicates that the pongid lineage diverged from the Miocene anthropoids 18-20 million years ago (Locke et al., 2011).

Orangutans are large-bodied and exceptionally sexually dimorphic. Males may weigh 200 pounds (78kg), more than twice the size and weight of adult females (36 kg). They are highly arboreal. The cranium is small against the enormous facial portion, with cranial capacity ranging from 365cc to 425cc, and the supraorbital torus is absent. The nasal bones are tiny in size and have been fused. The nasal bridge is not elevated; the nasal root is very narrow. The mandible is extremely large and projected forwardly.

- The sagittal and nuchal crests are generally more developed on the skulls of males than the females (Winkler et al., 1988).
- Cranial dimorphism is identifiable in infancy and develops further during juveniles via different growth rates (Masterson and Leutenegger, 1990).
- The pelages of male and female Orangutans are polychromatic, ranging from yellowish and reddish-orange through chestnut and marron to chocolate brown.
- The skin of Orangutans varies from light to dark brown. The faces of captives that have been deprived of sunlight can be pretty pinkish (Tuttle, 1986).
- The upper premolars (P3 and P4) and the second lower premolar (P4) are bicuspid (Hooijer, 1948).
- Orangutan molars share the basic hominoid pattern of four main cusps on M1-3 and five main cusps on M1-3.

Pan (Chimpanzee and Bonobos)

The Chimpanzees and Bonobos are more or less similar to the Gorilla. However, the body of the Chimpanzee and bonobos are not so well built as Gorilla.

Chimpanzees are found in the tropical forest of Africa. They inhabit a remarkable variety of primary and secondary forests, deciduous woodlands, riverine forests, and forest-savanna ecotonal regions, extending from the Gambia and Guinea-Bissau through West and northern Central Africa to Western Uganda and north-western Tanzania. Bonobos are confined to the humid, swampy forests of the central Democratic Republic of the Congo (Gantt, 1986; Nagatoshi, 1984). They are also misleadingly called pygmy chimpanzees because of their presumed smaller overall body size.

Chimpanzees and bonobos are almost like a man in body proportion. The average height of an adult male chimpanzee is 5 feet. The weight is 60 kg, and females are a little bit smaller than the male, and their average weight is 46 kg; male bonobos weigh around 45kg, and females weigh approximately 33 kg. The cranial capacity ranges between 400 to 500 cc.

The nasal bones are small; the nasal bridge is not elevated. The jaw clearly shows a forward projection. The skull is generally oval with a small face in comparison to the head. The lips are thin. The canines are large and projecting but smaller in comparison to those of Orangutans and Gorillas. The hands are elongated and narrow, while the legs are longer and more extensive than the Orangutan. The hind limbs are poorly adapted for upright posture. The fingers are long, and the thumb is small and opposable. The extended foot possesses an opposable great toe that is not in line with other toes. The heels are rudimentary when the Chimpanzee stands erect, and the hands generally reach up to knee level. They are good climbers and brachiators. They build nests on trees and sleep there at night.

The Chimpanzees are known for their tool-using behaviour. They organise themselves into groups that are relatively smaller than those of Gorillas. They maintain a well-organised dominance hierarchy in the group, but the structure of the group is loose. The habit of grooming, particularly among adults, is more frequent in chimpanzees than in other primates.

- Genomics studies indicate that chimpanzees and bonobos diverged from other anthropoids 4.5–6.0 million years ago. Estimates of bifurcation of bonobos and chimpanzees vary from 0.9–2.5 million years ago. Chimpanzees and bonobos are moderately sexually dimorphic in body size.
- Chimpanzees have relatively large ears in comparison with other apes and humans.
- Chimpanzee pelage is polychromatically black or dark brown before greying as they mature.
- Chimpanzee skin varies from white and pinkish through bronze, coppery hues, and black.
- Bonobos have black faces, ears, palms, and soles.
- Chimpanzee skulls are much less sexually dimorphic than Gorillas and Orangutans.

- The molars of chimpanzees and bonobos are metrically and morphologically similar to human molar teeth.

Gorilla

Gorillas are found in equatorial regions of Africa, covering Cameroon at the angle of the Gulf of Guinea, southward to the mouth of the Congo River on the western side and the high lands on the volcanic mountains of Kiyu province in the eastern Congo. They are the largest and the stoutest of all the living primates, yet the shyest and gentlest primate. Schaller and Emlen (1963) divided them into mountain gorillas and low land gorillas. Mountain gorillas are found in the eastern Congo and low land gorillas in Cameroon and Gabon in Africa.

Their height ranges from five to six feet in low land gorillas and between 5.3 to 5.10 feet in mountain gorillas. The adult males weight between 350-600 pounds. They have a massive head with an enormous supraorbital torus. Their forehead is shallow. The skull is nearly oval in appearances with a sagittal crest, which is weaker in females. The cranial capacity is 550 cc in males and 450 cc in females. The bones of the cranium are big and solid, with relatively small space for the brain. The jaws are large and projected forward and downward. The upper jaw is prognathous, and the lower jaw is massive. The canines are large and interlocked; the nasal bones are long, low and narrow with a slightly elevated nasal bridge and broad forwardly directed nostrils. The manus is more human in different features than in other anthropoid apes. The upper arms are longer than the forearms, unlike in other apes, while the hands are shorter and broader than in other apes. They have a well-developed thumb and great toe, which is set apart from other digits and opposable. The legs are short, and the foot is similar to that of a man. The anatomy of the foot shows that the Gorilla is less arboreal and more terrestrial.

Their gait can be stated as obliquely quadrupedal. They live in groups ranging from 8 to 24 members, the male to female sex ratio is 1:2 or higher. They are diurnal in habit, headed by the silver-backed male leader while others follow. The grooming habit is rarely observed. Their gestation period ranges from 250 to 300 days.

- Gorillas are among the most sexually dimorphic apes. Adult males weigh between 132 and 218 kg, whereas adult females are between 68 and 98 kg.

- Infant gorillas depend entirely on their mothers and suckle for one-and-a-half years.

The female Gorilla menstrual cycle is about 30 days.

The Hylobatinae

The Hylobatinae (hylobatid apes) encompass the Gibbons or lesser apes. The term, lesser apes refers only to the fact that they are much smaller than bonobos, chimpanzees, gorillas, and orangutans. They are arboreal creatures. Slender body with extremely long arms; the hand touches the ground when standing in an erect position. They are the perfect brachiator and capable of precision

grip. Their habitat is the rain forests of Southeast Asia, including Java, Sumatra, Philippines, etc.; they are also found in Northeast India and Myanmar (Burma). The head is large, elongated and narrow. The braincase is larger than the face, and the frontal elevation is low, with about 100 cranial capacity. The face is provided with a flat and broad nose and thin lips with pronounced eye orbits composed of thick bony rims. The supraorbital torus is not found.

Another group of gibbon known as Symphalangus or Siamang lives in Sumatra. The Siamang is larger than the common gibbon. They possess laryngeal air sacs. Some scholars have concluded that the Siamang represents an intermediate form between the Gibbons and Giant apes.

Check Your Progress

1) Give a brief account on Hylobatinae?

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10.1.2 The Hominidae

In the extant, it contains a single genus called Homo and a single species called sapiens. Homo sapiens have large cranium. The foramen magnum is more anteriorly located at the base of the skull. Therefore, the head is well balanced. The orbits are located under the anterior cranial fossa. The projection of jaws is significantly reduced. Face is orthognathism. The alveolar arches are more diminutive and delicate. The chin is well developed, and the nose is elongated with a well-developed nasal bridge. The vertebral column has a forward convexity due to which it acquires an S - shape. The distance between the thorax and the pelvis is relatively great. The great toe is non-opposable and lies in line with other toes, unlike in apes. The toes, excluding the great toe, are somewhat shorter. Man's body retains scanty hair, unlike the apes and absence of tactile hairs and feelers. The rate of development is slow, and the growth period is longer and extended life span of roughly more than 50 years.

Moreover, human life histories are characterised by a lengthy period of juvenile dependency, reproductive support by older post reproductive individuals. Humans display moderate sexual dimorphism in stature, weight, and many skeletal features, including dentition. Human females are more glabrous, usually keep their head hair, and have special fat deposits in their breasts, hips, and thighs, making them more curvilinear than their male counterparts. In humans, the skin pigmentation ranges from swine-pink to very dark, purplish-brown (Buettner-Janusch, 1973; Parra, 2007). The human neurocranium is comparatively large, and the splanchnocranium is quite shortened. The cranium is high, rounded, and unadorned by superstructures. The eyebrows rise flagrantly above the orbits, and postorbital constriction is notably reduced compared to apes (Schultz, 1963). The human face is also further distinguished from apes

because our nasal bones project anteriorly from the facial plane, forming an awning over the pyriform aperture.

The human palate is parabolic compared to the U-shaped configuration of great ape plates. Hylobatid apes are more closely related to palate shaped humans. Human canines are small and bluntly pointed. They do not project much. In their low degree of canine sexual dimorphism, humans compare most closely with gibbons among the apes (Le Gros Clark, 1971). Human molars show the basic hominoid patterns of cusps and grooves. However, in some populations, the third molar reduction and even absence are noticed. Human lower molars are generally more abundant than those of great apes, and the enamel is thick. Because of the unique body proportion, the body can be easily distinguished from those of apes. Humans have longest lower limbs relative to truncal height among the anthropoid primates, and the upper limbs are also long (Swindler, 1976; Shelli et al., 1998). Man occupies the highest place in the animal kingdom because of his fully erect gait, bipedal locomotion, power of articulate speech and highly convoluted brain.

10.2 POSITION OF HUMANS IN THE ANIMAL KINGDOM

Taxonomic Category	Category to which humans belong	Biological features used to define and place humans in this category
Kingdom	Animalia	Humans are animals. We do not make our food (as plants do) but depend upon the intake of food.
Phylum	Chordata	Humans are chordates. We have a notochord (a rodlike structure of cartilage) and nerve cord running along the back of the body, as well as gill slits in the embryonic stage of our life cycle.
Subphylum	Verterbrata	Humans are vertebrates, possessing an internal backbone with a segmented spinal column.
Class	Mammalia	Humans are warm-blooded mammals covered with fur and possessing mammary glands for nourishing their young after birth.
Order	Primates	Humans are primates, an order of mammals, with generalised anatomy, a relatively large brain, and grasping hands and feet.
Suborder	Anthropoidea	Humans are anthropoids, social, daylight-active primates.
Superfamily	Hominoidea	Humans are hominoids with broad, flexible shoulders and no tail. Chimps, bonobos, gorillas, orangutans, gibbons, and siamangs are also hominoids.

Family/ Subfamily	Hominid/Hominin	Humans are hominids. We are hominoids from Africa, genetically more closely related to chimps, bonobos, and gorillas than hominoids from Asia. Some scientist uses 'hominid' to refer only to humans and their ancestors. Others include chimps and gorillas in this category, using the subfamily 'hominin' to distinguish humans and their ancestors from chimps and gorillas. The two taxonomies differ according to emphasis on genetic versus morphological similarities.
Genus Species	Homo sapiens	Humans have a large brain and rely on cultural adaptations to survive.

10.3 COMPARISION OF SOME MORPHOLOGICAL AND ANATOMICAL FEATURES AND BIOLOGICAL CHARACTERISTICS OF MAN AND APES

10.3.1 Skull

Characteristics of Modern Human Skull

- The cranial portion is more developed than the facial portion.
- Skulls are orthognathous, with smooth convexities in the facial, parietal, and occipital regions.
- The head is perfectly balanced on the spinal column.
- The average cranial capacity is 1300cc – 1450cc, three times larger than the apes.
- Nasal bones are elevated and sharply inclined to each other and presence of nasal spines.
- Diastema is absent, and the dental arch is parabolic or elliptic.
- The supraorbital ridge is moderately developed or absent.
- The zygomatic arch and mastoid process are moderately developed.
- The foramen magnum is found at or forward of the central position.
- There is a prominent symphysial region.

Characteristics of Ape skull

- The facial portion is more developed than the cranial portion.
- Apes face shows prognathism.
- Due to an underdeveloped brain, there is no convexity in the skull.
- The head is more posteriorly placed.

- The cranial capacity ranges between 128cc – 500cc.
- Nasal bones are thicker and longer, with the absence of nasal spines.
- Diastema is present, and the dental arch is U- Shaped.
- The supraorbital ridge is well developed to protect the eyes and serve the buttress to resist the tremendous upward pressure exerted by the jaw in chewing.
- The foramen magnum is just below the skull towards the rear of its centre.
- There is no prominence in the symphysial region.
- The mandible is massive.

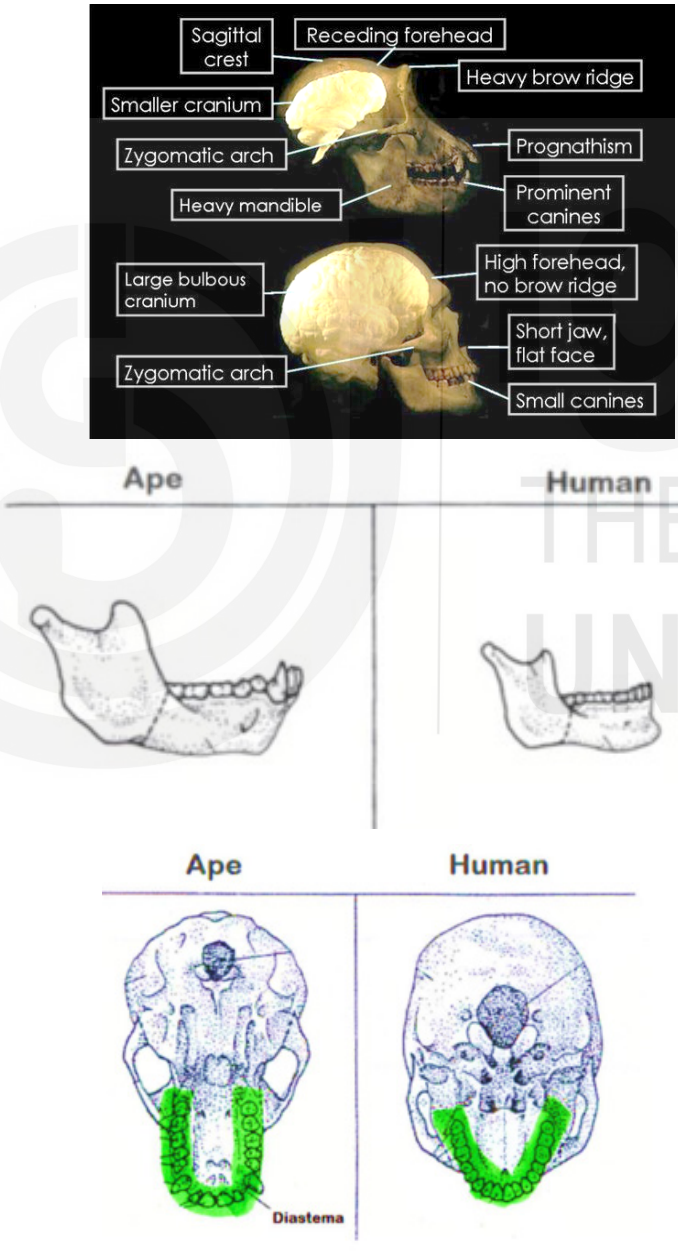

Fig. 10.1: Morphological Comparisons of Human and Ape Skull
(***Source:*** https://www.pathwayz.org/Tree/Plain/APE+VS.+HOMININ+SKULLS)

Check Your Progress

2) Explain the differences between Man and Ape skull.

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10.3.2 Foot

Characteristics of Human Foot

- The great toe is not opposable, and it is in the same line as the other digits. It is also the biggest digit.
- The lateral toes are reduced in size, and the fifth one is the smaller one.
- The dorsal and ventral surfaces of all the digits are the same way, the ventral surfaces being directed downwards.
- The first metatarsal presents a flattened area at its posterior end for articulation with the cuneiform bone.

Characteristics of Ape Foot

- In apes, the great toe is opposable. It is not well developed and also not in line of other digits.
- In apes, the lateral toes are well developed.
- The third digit is the largest digit in apes.
- In apes, the ventral surface of the great toe does not face downwards but laterally.
- In apes, the great toe is free from the common covering.
- In apes, the articular region of the cuneiform is convex, and that of the metatarsal is concave.

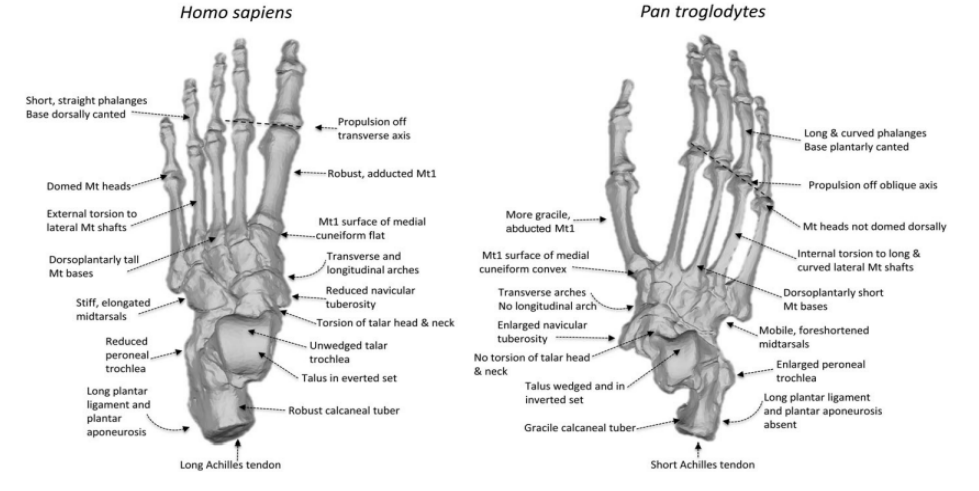

Fig. 10.2: Anatomical Comparisons of Human and Ape foot

(***Source:*** McNutt et al., 2018)

10.3.3 Vertebral Column and Rib Bones

In apes, the vertebral column is a single curve – C shaped. In contrast, it is S-shaped in humans, which is also an essential characteristic of erect posture. S-shaped vertebral column enables to carry the bodyweight nearly directly above the hip joints. Humans, Chimpanzees, and Orangutans vary in modal vertebral formulae, with 12 thoracic and five lumbar vertebrae in humans, 12 thoracic and four lumbar vertebrae in Orangutans, and 13 thoracics and 3 to 4 lumbar vertebrae in Chimpanzees. The rib cage is barrel-shaped in humans while it is flared, making it conical or funnel-shaped.

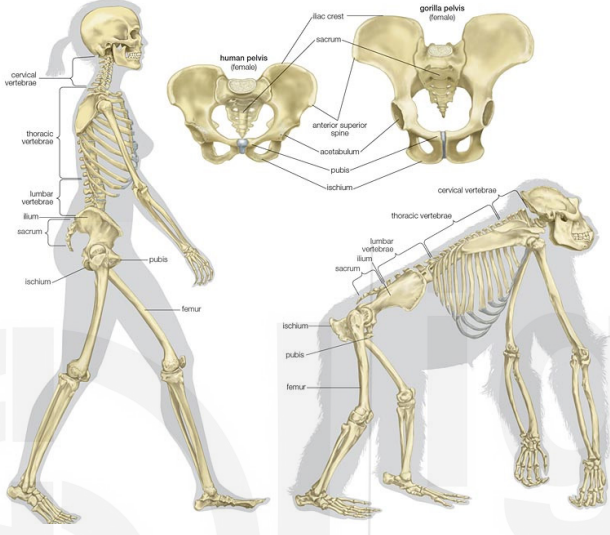

Fig. 10.3: Anatomical Differences of Human and Ape Axial Skeleton including Vertebrae Column, Pelvic girdle

(***Source:*** https://www.pathwayz.org/Tree/Plain/APES+VS.+HOMININ+SKELETONS)

10.3.4 Hip Bone or Pelvis

In apes, the hip bone or pelvis is extended due to quadrupedal posture. In contrast, in humans, it forms bowl-shaped and much broader to support the abdominal organs and to give stability when walking upright as it transfers the weight directly to the legs. The pelvis is another important area in which many changes have occurred due to hominid evolution. Pelvis has three distinct parts – ilium, ischium and pubis. Ilium appears to be the earliest one to evolve, followed by the other parts.

Contrasting features of the pelvic skeleton in apes and human	
Ape	**Human**
Elongated ilium	Short ilium
Moderately developed sacral area	Large sacral area
Long sacrum	Broad sacrum
Frontally oriented ilia	Medially oriented basin like ilia
Great distance between sacral area and acetabulum	Sacral area close to acetabulum
Long ischium	Short ischium

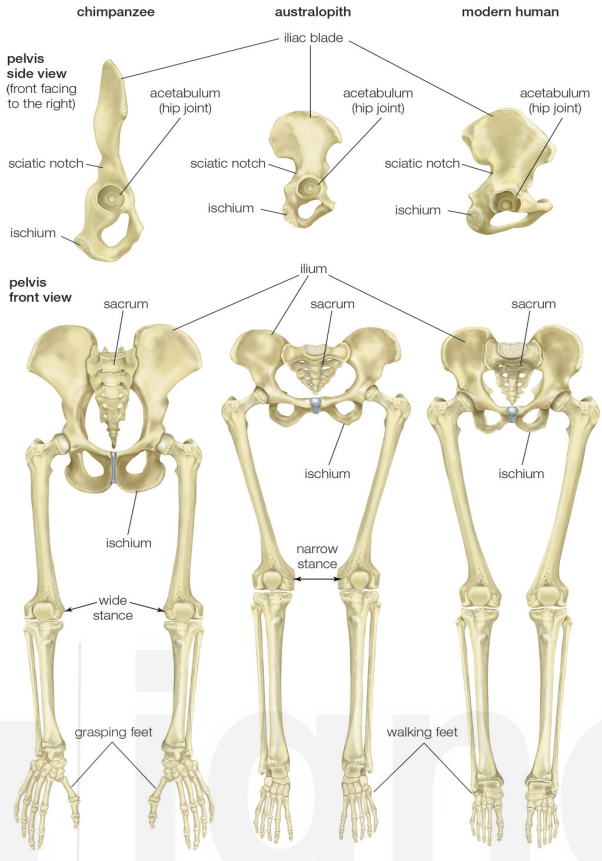

Fig. 10.4: Comparison of the Pelvis and lower limbs of a chimpanzee, an australopith, and a modern human

(***Source:*** Encyclopædia Britannica, Inc.: https://cdn.britannica.com/57/52957-050-0C501D65/Comparison-pelvis-chimpanzee-limbs-human-australopith.jpg)

Check Your Progress

3) What are the significant skeletal changes that occurred during the emergence of man from ape?

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10.3.5 Knee-valgus angle

Humans have a larger valgus angle (the angle the femur makes at the knee), i.e. our thigh slopes inward, bringing our feet closer to the centre of gravity, which shifts less weight when walking, making it more efficient. In the case of apes, the knee–valgus angle is small, and when they attempt to walk on two legs, they swing back and forth.

10.3.6 Hands

Among the apes, due to the habit of brachiating, the thumb became reduced as they used their fingers more as hooks. Whereas in humans, the thumb is

enlarged. Moreover, the first metacarpal is connected to the wrist by a 'saddle' joint, enabling the thumb to be brought across the hand to touch the tip of the first or any other finger. Only humans can achieve the full thumb tip to others fingertips.

10.3.7 Hand Proportions or Thumb-to-digit Ratio (IHPs)

Hand proportions of humans are usually compared with those of apes using the thumb-to-digit ratio (or IHPs), a good functional measure of thumb opposability and, thus, a proxy for manual dexterity. The humans can be easily distinguished from modern apes by the thumb relative to the other digits. The modern human IHP range is well above that of modern apes, linked directly to the human capability to perform an efficient pad-to-pad precision grasping. Chimpanzees and orangutans have significantly shorter thumbs than gorillas and hylobatidae.

10.3.8 Brain

The human brain is about three times larger than our closest living relative, the chimpanzee. Besides, a part of the cerebral cortex, which plays a crucial role in memory, attention, awareness, and thought – contains twice as many cells in humans as the same region in chimpanzees. The network of brain cells in the cerebral cortex also behaves differently in the two species (Mora-Bermudes et al., 2016). Using live microscopy it is seen that progenitor cells that form the human cerebral cortex spend around 50% more time in a stage of the cell division process called metaphase compared to the same cells from chimpanzees or orangutans. Progenitor cells are more likely to become neurons if they had shorter metaphase than the progenitor cells, more likely to remain proliferating as stem cells for longer (Mora-Bermudes 2007).

10.3.9 Blood Groups

Blood groups are defined by polymorphisms in immunogenic molecules in blood cells, plasma, and body secretors. Various blood groups or types can be found across species, including in humans and great apes. The human ABO blood group system is very significant in transfusion and transplantation. These antigens are oligosaccharides synthesised by glycosyltransferase enzymes, which create a pattern of sugars present on the outer membrane of cells or secreted glycoproteins. In type O blood group, a carbohydrate sequence called the H-antigen is present. Type A and Type B individuals modify the H-antigen by adding monosaccharides to produce corresponding A and B antigens. The presence of ABO polymorphism is highly variable across all primates. Chimpanzees have been found to have primarily type A blood, with type O less commonly. Gorillas appear to have type B exclusively, and orangutans express all three blood types. Another type of blood group known as the Rh blood group is essential in humans because of its role in the haemolytic disease of the newborn. Rh polymorphisms also exist in chimpanzees, where they were originally described in the R-C-E-F blood group system. Chimpanzees share some of the variants of Rh with humans. Still, the two species have additional variants that they do not share. Another blood group MNS also exist in a

chimpanzee in the form of V-A-B-D blood groups (https://carta.anthropogeny.org/moca/topics/blood-group-antigen-types-and-prevalence).

10.3.10 Comparative Overview

Sl. No.	Anatomy and Differences	Human	Ape
1	Cranial sutures	Human cranial sutures are highly serrated and fused very late and exist throughout one's life.	Cranial sutures of apes are less serrated and start to fuse at an early age.
2	Orbits	Human orbits have rectangular and rounded angles.	Chimpanzee has an elliptical orbit, Orangutan and Gorilla have rounded or oval orbits.
3	Nasal bones and bridges	The nasal bones in humans are short and broad with raised nasal bridges.	Apes lack a nasal bridge completely, and in Chimpanzee, it is short and flat while long in Orangutan and broad at the base and long in Gorilla.
4	Nasal sutures	Nasal sutures are present in humans.	It is present in Chimpanzee but often absent in Orangutan and Gorilla.
5	Skull size	The human brain is three-fold more than the average apes' brain size. It ranges from 1300 cc to 1450 cc.	The apes have a smaller brain, where chimpanzees and gorillas have between 400-500cc, orangutans have 365cc to 425cc.
6	Teeth and lower jaw	Human cranium is more prominent and protruding. Facial portion is smaller than the cranial portion.	They have a large facial portion than the cranium portion.
7	Forehead	Humans have a prominent and arched forehead.	Apes lack forehead.
8	Tuberosities	The frontal and parietal tuberosities become prominent in the human cranium	It is less prominent as compared to humans.
9	Occipital bones	Occipital bones are well arched and more protruding than apes.	It is more protruding than humans.

10	Foramen magnum	In human, foramen magnum is anteriorly placed.	In apes, the foramen magnum is placed towards the posterior.
11	Skull placement	Human skull is well mounted on the top of the vertebral column in a proper balance.	Ape's skull hangs on the vertebral column.
12	Nuchal region	The nuchal region is comparatively smooth in humans.	Apes have a rugged nuchal region.
13	Premaxilla	Premaxilla is fused to maxilla in humans.	It is well marked in apes.
14	Food habit and teeth	Humans eat tender and cooked food, so we have smaller teeth.	Apes have larger teeth; canine is also larger, pointed, sharped and projecting.
15	Legs and arms	Legs are longer than arms in humans.	Arms are longer than legs in apes.
16	Mandibles and chin	Humans have slender and lighter mandibles or lower jaw with distinct chins.	They have larger and massive mandibles without a chin.
17	Pelvis and birth canal	Pelvis and birth canal is basin or U – shaped with the broad and short ilium.	They have long and flat ilium placed on the back of the animal.
18	Vertebral column	Humans have an S-shaped vertebral column.	Apes have a C-shaped vertebral column.
19	Diastema	Diastema is not present in humans.	Diastema (the gap between the upper incisor) can be seen in most apes.
20	Hallux (Great toe)	Humans have non-opposable hallux, and lateral movement is restricted.	Apes have opposable hallux, and they can move it laterally.
21	Locomotion	Bipedal locomotion	Quadrupedal locomotion and brachiation.
22	Dental arch	U-shaped dental arch.	V-shaped dental arch.

23	Genetic difference	Humans have 46 chromosomes	Chimpanzee and other apes have 48 chromosomes
		The genetic difference between human individual is about 0.1%	The genetic difference between Chimpanzee is approximately about 1.2%.

Similarities between humans and apes	
1	Human and apes have similar bones and muscles, the same manner in the working of their nervous systems.
2	Humans and apes have the same number of fingers and toes.
3	The structure of the human female uterus is somewhat similar to a female ape.
4	Both human and apes have an opposable thumb.
5	Although not identical, apes have blood types similar to the human ABO blood group system.
6	Both female apes and female humans have regular menstrual cycles.

10.4 SUMMARY

There are lots of differences in the anatomy and morphological features between apes and humans. Human cranial sutures are highly serrated in the skull portion and fused very lately and exist throughout one's life compared to the cranial sutures of apes. In humans, the facial portion is smaller than the cranial portion. Whereas, apes have a more significant facial portion than the cranial portion. The human head is well mounted on the top of the vertebral column in a proper balance, thereby placing the foramen magnum anteriorly. In contrast, the foramen magnum is placed posteriorly in apes. Humans are erect in posture; therefore, they are characterised by bipedal locomotion, while brachiation and quadrupedal locomotion are the characteristics of apes. When it comes to the similarities between apes and humans then also we see that they both have similar bones and muscles, the same manner in the working of their nervous system and many other similar characteristics.

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10.6 ANSWERS TO CHECK YOUR PROGRESS

1) Hylobatidae (hylobatid apes) encompass the gibbons or lesser apes. "The term lesser apes" refers only to the fact that they are much smaller than bonobos, chimpanzees, gorillas, and orangutans. They are arboreal creatures. Slender body with extremely long arms; the hand touches the ground when standing in an erect position. They are the perfect brachiator and capable of precision grip. Their habitat is the rain forests of Southeast Asia, including Java, Sumatra, Philippines, etc.; they are also found in Northeast India and Myanmar (Burma). The head is large, elongated and narrow. The braincase is larger than the face, and the frontal elevation is low, with about 100 cranial capacities. The face is provided with a flat and broad nose and thin lips with pronounced eye orbits composed of thick bony rims. The supraorbital torus is not found. Another group of gibbons

known as Symphalangus or Siamang lives in Sumatra. The Siamang is larger in size than the common gibbon. They possess laryngeal air sacs. Some scholars have concluded that the Siamang represents an intermediate form between the Gibbons and Giant apes.

2) Humans and apes belong to the same order primates and suborder Anthropoidea. However, both belong to different families. For further details on the differences between the humans and apes skulls please refer to the sub-section 10.3.1

3) Ancestors of modern man and the living apes did not look like any living ape today because they developed in different lines and separated during the Oligocene period. Ape-Man line of development became differentiated from each other in Miocene. Changing ecological and environmental conditions during the ancestry of man, the vegetarian great apes were compelled to change their food habits thereby changing in the overall morphological and anatomical structure. Various changes took place in the hominid evolution and the changes are remarkably noticed in brain and skull, hand and limbs, vertebral column, ribs, pelvis etc. the major changes that occurred during the emergence of man from ape are given below.

- **Skull and Brain**
  - » Cranial portion of skull increased as compared to the facial portion.
  - » Increase in the size of the brain which is evident through increase in cranial capacity.
  - » Well-developed nasal bridges.
  - » Face became orthognathous with prominent chin.
  - » Supraorbital ridges were less developed.
  - » Foramen magnum shifted anteriorly and downwardly due to erect posture.
  - » Decreased sagittal crest.
  - » Dental arch became parabolic or elliptical.
- **Vertebral column**
  - » Vertebral column change to 'S' shaped from 'C' shaped.
- **Pelvis**
  - » From elongated ilium to shortened ilium.
  - » Moderately developed sacral area to largely developed sacral area.
  - » Long sacrum to broad sacrum.
  - » Frontally oriented ilia to medially oriented ilia.
  - » Long ischium to shorter ischium.
- **Hands and feet**
  - » Fore arms became shorter and hind limbs became longer.
  - » Femur angle became greater.
  - » Presence of Linea aspera at the back of femur which is one of the special characteristics for erect posture.